# Guidelines for Conducting Birth Defects Surveillance

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Copies of *Guidelines for Conducting Birth Defects Surveillance* can be viewed or downloaded from the NBDPN website at http://www.nbdpn.org/bdsurveillance.html.

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# Introduction

In January of 1999, the National Birth Defects Prevention Network (NBDPN) established a Surveillance Guidelines and Standards Committee (SGSC) in order to develop and promote the use of standards and guidelines for birth defects surveillance programs in the United States. This set of guidelines is designed to serve as an important first step in the documentation of this process and as the vehicle for dissemination of the committee's findings.

*The Guidelines for Conducting Birth Defects Surveillance* (henceforth referred to as *The Surveillance Guidelines*) were developed with three major long-term objectives in mind:

> ➢ To improve the quality of state birth defects surveillance data, including accuracy, comparability, completeness, and timeliness.

> ➢ To enhance the utility of state birth defects surveillance data for research on the distribution and etiology of birth defects.

> ➢ To encourage and promote the use of state birth defects surveillance data for the purposes of linking affected children with services and evaluation of those services.

The technical guidelines that make up this document provide a way of improving the quality of birth defects surveillance data, which in turn enhances their use in support of the latter two objectives. Fundamental to quality is ensuring that procedures for all aspects of data definition, collection, management, and analysis are established and followed. Because state-based surveillance systems operate with different objectives and data needs, it is clear that, with respect to procedures and standards, "one size does not fit all." It is also clear, however, that common guidelines can provide a basis for the development of system-specific operating procedures and supporting manuals.

Variation among surveillance programs is manifest along several dimensions. These include:

> ➢ *Objectives*, which can be very diverse but commonly include:

>> • Providing baseline data on occurrence

>> • Identifying populations at increased risk

>> • Monitoring changes in occurrence

>> • Investigating clusters

>> • Collaborating with research

>> • Estimating service needs

>> • Referring affected children to services

>> • Evaluating prevention programs

> ➢ *Case ascertainment methods*

>> • Active – case finding

>> • Passive – case reporting

>> • Combined

> ➢ *Organizational location*

  - Health department

  - University

  - Other

The first two dimensions – objectives and case ascertainment methods – are of particular significance in attempting to develop guidelines that have the breadth to be useful (i.e., universality), while at the same time making clear that there is not necessarily a common denominator across programs. Thus most of the guidelines in this volume are phrased as *recommendations* or "shoulds," as opposed to *standards*, which could be interpreted as "musts." The exception to the latter is Chapter 10, which refers the reader to information on how data are to be reported to NBDPN for the Annual Report. The relevance of organizational location to the guidelines is probably restricted to legislative issues, which are addressed in Chapter 2.

*The Surveillance Guidelines* consist of a series of chapters covering the fundamental aspects of developing, planning, implementing, and conducting surveillance for birth defects and using the resulting data. Although the focus is on birth defects, most of the principles described are relevant and applicable to surveillance for any health outcome. Just as the methods and strategies developed for birth defects in the Metropolitan Atlanta Congenital Defects Program provided a blueprint for the subsequent development of the Metropolitan Atlanta Developmental Disabilities Surveillance Program, the information included in these guidelines can provide a blueprint for the development of surveillance for developmental disabilities among the states.

On reviewing the guidelines, the reader will note that a number of the chapters are supported by appendices. In many instances these appendices are designed to provide additional information on technical issues considered. In some cases they provide extensive detail on procedures that are currently being used by surveillance programs. Because of their size, three documents cited as appendices will only be available in electronic format. These are the NBDPN Abstractor's Instructions (Chapter 3, Appendix 3.2) and the Texas Disease Index and the CDC Six-digit Codes (Chapter 5, Appendices 5.1 and 5.2, respectively). Information on how to access the electronic format is included in each appendix.

*The Surveillance Guidelines* are being published in two formats: as print copy and through the NBDPN website. The Surveillance Guidelines and Standards Committee anticipates updating and revising the guidelines over time. Whenever a revision is published, a revision date will appear in the chapter header to distinguish that page or pages from previous versions. Because we anticipate this will be a living document, we encourage comments, suggestions, and corrections. If you have such, please submit them through the link to the Surveillance Guidelines and Standards Committee on the NBDPN website.

This set of guidelines represents a great deal of work by a large number of individuals. The development of the document was carried out by the NBDPN Surveillance Guidelines and Standards Committee. A working group for each of the chapters did most of the writing. When chapters were completed in draft form, they were submitted to the SGSC Steering Group for review and suggested revisions. When a draft was considered acceptable to the Steering Group it was sent to Dr. Lowell Sever of Battelle Centers for Public Health Research and Evaluation, the editorial consultant for the reference manual. Dr. Sever then edited the chapter, returning it to the Steering Group, and working groups when necessary for clarification and acceptance of his revisions. Several of the chapters were also sent to specially assembled "Focus Teams" for review and assessment of the technical content. When the final content was agreed upon, the chapter was submitted to a Battelle technical writer and editor for finalization of structure and format.

We have compiled all of the contributors to this intensive process into a single acknowledgements page. *The Surveillance Guidelines* represent a significant and complex undertaking that could not have been accomplished without the contributions of this large number of individuals, and we thank them all.

We dedicate this milestone document to Larry Edmonds of the Centers for Disease Control and Prevention in recognition of his strategic vision, inspiring leadership, and steadfast commitment – both to the National Birth Defects Prevention Network and to the enhancement of birth defects surveillance generally – throughout the remarkable process of developing *The Surveillance Guidelines*.


*Carol Stanton, Chair*
*NBDPN Surveillance Guidelines and Standard Committee*


*Lowell E. Sever, Editor*                                              *Joanne Abed, Technical Editor*

# Chapter 1

# The Whys and Hows of Birth Defects Surveillance – Using Data

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# 1.1 Introduction

The ultimate value of any public health surveillance program lies in the ways in which the data collected are used to improve the health of the public. State birth defects surveillance programs are no exception; they exist to improve public health. Every program must have clear goals and objectives that drive how their surveillance data are used toward improving public health. Public input through partnerships with state agencies and organizations and the effective utilization of advisory committees are essential to establishing and revising program objectives and ensuring that the resources exist to meet them.

The purposes and objectives established by state birth defects surveillance programs are constantly evolving. Some objectives are traditional, such as those having to do with the epidemiologic purposes of surveillance; others have emerged more recently, serving to broaden the scope of surveillance programs. Birth defects surveillance programs increasingly use data for services planning and evaluation, for development and evaluation of prevention strategies, to inform parents of children with birth defects about available services, for studies of the societal impact of birth defects, for referral of families to needed services and resources, and for clinical research studies. The consistent theme among these emerging data uses is how birth defects surveillance may benefit other programs in the quest to improve the public's health. In the face of fluctuating resources for public health and obstacles resulting from concerns about confidentiality of health records, the need to incorporate public input in planning and priority-setting has never been greater. This chapter will attempt to address some of the issues in the forefront as we plan for the future of birth defects surveillance.

In the remainder of this chapter we present the rationale for conducting birth defects surveillance (Section 1.2), key steps in establishing a state-based birth defects surveillance program (Section 1.3), and some important uses for birth defects surveillance data (Section 1.4). References cited in this chapter may be found in Section 1.5.

## 1.2 Rationale

When contemplating initiating or enhancing a birth defects surveillance program, a number of questions come to mind:

> ➢ What is the rationale for conducting birth defects surveillance?
>
> ➢ Why is birth defects surveillance important?
>
> ➢ How do birth defects surveillance data benefit other programs?
>
> ➢ What are the barriers to collection and full utilization of birth defects surveillance data?

In this chapter, we provide answers to these questions, which may help you advocate for and prepare to launch or expand a birth defects surveillance program in your area.

### 1.2.1 What is the rationale for conducting birth defects surveillance?

CDC defines public health surveillance as:

> *The ongoing, systematic collection, analysis, and interpretation of health data essential to the planning, implementation and evaluation of public health practice, closely integrated with the timely dissemination of these data to those who need to know. The final link of the surveillance chain is the application of these data to prevention and control. A surveillance system includes a functional capacity for data collection, analysis, and dissemination linked to public health programs (Centers for Disease Control, 1988).*

It is clear from this definition that a birth defects surveillance program must establish goals and objectives for how data are to be collected, analyzed, disseminated, and used. It is through the latter (i.e., data use) that the efforts from the former are translated into public health action and health improvement. Thus, using data to meet a program's objectives is the most important aspect of any public health surveillance program; merely collecting data is not enough. How data are being used is also what programs tout when they need to showcase their activities to agency officials and legislators.

Because of the essential relationship of the ultimate uses of data to the design and conduct of birth defects surveillance, we begin these guidelines with a consideration of fundamental data-related issues, considering not only the rationale for birth defects surveillance but the key steps for establishing state-based birth defects surveillance programs, followed by a discussion of the use of surveillance data for improvement of the public's health. Every surveillance program should have a plan for data utilization that incorporates public input on all phases of the process – from data development, through data collection, to data dissemination to the public. Suggestions for developing a data utilization plan are presented in Section 1.2 below.

### 1.2.2 Why is birth defects surveillance important?

States have many reasons for conducting birth defects surveillance. The value of birth defects surveillance lies in how the data are collected and how they are used, with respect to the goals of the program. All programs should establish goals and objectives, which make it clear that the ultimate rationale for conducting public health surveillance is to have data that can be used to improve the health of the public. Reporting the

data certainly qualifies as "using the data," yet this should never be considered sufficient as it fails to meet the definition of public health surveillance cited above.

The objectives of state birth defects surveillance programs have evolved over the past 40 years. Lynberg and Edmonds (1992) assessed the objectives that had been established by surveillance programs by the early 1990s. Table 1.1 organizes these objectives under broad purposes originally suggested by Reed and Meaney (1988) with some slight modifications. A review of the table highlights the potentially broad mission of birth defects surveillance, providing state programs with a way of assessing how they are utilizing data currently and possible new uses.

**Table 1.1 Purposes and Objectives of Birth Defects Surveillance**

| Purposes | Objectives |
|---|---|
| Epidemiologic | Develop timely baseline birth defects rates |
| | Monitor trends and relationships to environmental factors |
| | Perform cluster investigations |
| | Provide basis for ecologic and etiologic studies |
| | |
| Planning/Prevention | Provide data for services planning |
| | Provide basis for prevention strategies |
| | Evaluate efficacy of preventive services |
| | |
| Educational/Social | Inform public about public health importance |
| | Inform parents about resources and care facilities |
| | Provide data for studies of economic impact |
| | Provide data for follow-up studies of long-term effects |
| | |
| Healthcare and human services | Refer children to services and resources |
| | Evaluate services utilization |
| | |
| Clinical | Provide basis for clinical research |

Adapted from Lynberg and Edmonds (1992) and Reed and Meaney (1988) with modifications.

### 1.2.3 How do birth defects surveillance data benefit other programs?

The benefits of birth defects surveillance data to human service programs include: identifying children in need of services to ensure that they and their families are referred appropriately; evaluating service utilization by children with birth defects and their families; and planning the location of services for particular conditions in areas of highest frequency. An important use of surveillance data is monitoring birth defects trends following the initiation of prevention programs in order to evaluate their effectiveness.

One of the public health benefits of the computer age is enhanced capacity for record linkage. Record linkage using public health data has a longer history than most people realize, beginning in the 1950s with the availability of computers in university settings. Pioneering investigators like Harold B. Newcombe (1962) recognized the utility of linking vital records data in studying human populations. The potential now exists for extensive computerized record linkage in birth defects surveillance programs, allowing for the tracking of children with a health-related condition from the point of identification through access to services. Many computer-based systems already exist for documenting health care delivery, including diagnostic and procedure codes. Birth defects surveillance records have been linked to many other public health program databases. These include, for example, newborn screening to conduct epidemiologic studies, special education data to predict the need for services for children with mental retardation, and early intervention

program data to assess the overlap and utility of a birth defects surveillance program as a "child find" resource.

In the final section of this chapter we describe a number of applications of these approaches that can serve as models for states developing birth defects surveillance programs, as well as for programs considering expansion of the current uses of their data. To date, the potential for applications of these types exceeds available resources to support them and to overcome some of the obstacles discussed immediately below.

## 1.2.4  What are the barriers to collection and full utilization of birth defects surveillance data?

While improved methods and technological advances have increased our ability to collect data, there have been intensified efforts to protect the confidentiality of records and the information they contain. Many birth defects surveillance programs – based both in health departments and in other institutions such as universities – have encountered increasing concerns and pressures as a result of Health Insurance Portability and Accountability Act (HIPAA) regulations and issues surrounding their interpretation and implementation. A variety of HIPAA-related issues are discussed in Chapter 2 of these guidelines. Even though the HIPAA regulations include public health exclusions regarding access to records without a patient's consent, programs have seen increased awareness and concerns on the part of hospitals and clinics reporting cases and data. These concerns are magnified when a surveillance program attempts to expand data usage through linkage to other databases covered under HIPAA regulations.

Prior to HIPAA, concerns often surfaced about data sharing among officials in different programs within the same state agency or among programs located in different agencies. Such concerns were usually due to program-specific regulations about data use. Program regulations frequently impede attempts to link records between case-finding databases and service-delivery databases. As a result, attempts to meet the very reasonable public health goal of ensuring access to services by those in need may be thwarted. Thus, programs are strongly urged to consider strategies for surmounting these problems well in advance of undertaking data collection and record linkage.

## 1.3  Synopsis of Key Steps in Establishing State-Based Birth Defects Surveillance Programs

In this section we outline some of the key steps in establishing a birth defects surveillance program. These include:

➢ Defining the objectives and purposes of the program

➢ Considering legal issues

➢ Engaging external support

➢ Leveraging resources

➢ Considering record linkage

Time devoted upfront to serious consideration of these issues will be well spent and will ensure that the resultant program is established on a firm footing.

### 1.3.1  Defining the Objectives and Purposes of the Program

The success of a birth defects surveillance program is likely to be highly dependent on the host agency's commitment and support. Without programmatic commitment and resource support at the agency level, programs are apt to languish in circumstances that do not allow much beyond the collection and reporting of data. In these situations, using data in ways other than the calculation of rates and their dissemination in reports is usually not possible. Programs committed to expanding how birth defects surveillance data are used must establish programmatic objectives and demonstrate to agency officials how the data could be used. This involves prioritizing what uses would be of greatest utility in terms of meeting agency goals and objectives, demonstrating (or "marketing") to the agency how beneficial these data uses could be, and working to achieve commitment of additional agency resources.

Another strategy for increasing support from the agency in which the surveillance program resides is to gather support from other intra-agency programs and from external agencies that could benefit from the use of birth defects surveillance data to meet their own programmatic goals. Often other programs and agencies, given enough information about birth defects surveillance and the objectives of the program, will see potential uses of the data that are beyond the current scope of the surveillance program.

There has been an increase in intra-agency collaboration during the last ten years through the availability of federal support for data linkage and integration. A prime example of data collaboration would be linking birth defects surveillance databases with Children with Special Health Care Needs (CSHCN) program databases that collect data on program enrollment and services. These linked data sets could then be used to evaluate the rates at which this long-term maternal and child health program is utilized. Such applications have been accomplished in some states through grant support from the Maternal and Child Health Bureau (MCHB) of the Health Resources and Services Administration (HRSA) and through cooperative agreements with the Centers for Disease Control and Prevention (CDC). Interagency collaboration in linking birth defects surveillance program databases with services databases (such as those for early intervention programs or developmental disabilities) have begun in a few states. The benefits to be gained in this way – i.e., by utilizing birth defects surveillance data as a means of identifying children eligible for special programs, such

as early intervention – is clearly a "selling point" that can lead to additional resource allocation, either from within the host agency for the birth defects program or from an external agency in need of the data.

Most birth defects surveillance programs experience cyclical problems with availability of state resources, leading them to define precisely what they can and cannot do given the resources available to them. While it is certainly necessary for programs to realistically budget their resources to ensure continued viability, programs also need to engage both intra-agency and interagency support for their goals and objectives as a means to maintain and expand a surveillance program. At a minimum, programs should allocate personnel time to educate officials of their own agency and other agencies about birth defects surveillance and its importance and potential uses in the public health field.

## 1.3.2  Considering Legal Issues

To the extent possible, programs should consider the inclusion of references to data use in the legislation that authorizes birth defects surveillance. Given the relative ease with which rules – as compared to laws – can be changed, it is generally desirable to make references to potential data uses for surveillance data more general in the statute and more specific in the rules. Rules and regulations that refer to the authorizing statutes are the obvious choice as to where best to specify detailed uses to which surveillance data will be put. Relevant issues and legal considerations are discussed extensively in Chapter 2 of these guidelines.

## 1.3.3  Engaging External Support

Beyond seeking intra-agency and interagency support for a new surveillance program or for expansion of an existing surveillance program, program staff should also seriously consider means to attract the support of both non-governmental partnering organizations and the public.

**Partnering organizations.** The importance of building partnerships with organizations such as the local March of Dimes can never be sufficiently stressed. In recent years, the success story of the birth defects surveillance program in North Carolina is arguably without peer. The program has consistently credited the partnership it built with the March of Dimes as a major contributor to its success in garnering additional resources for the program. In Texas, the March of Dimes was also instrumental recently in restoring funds to maintain the Texas Birth Defects Monitoring Division, funds that had not been requested in the budget put forward by the Texas Department of Health. These kinds of partnerships should be entered into with clear and consistent agreement among the players regarding the objectives of the program relative to data usage, prioritization of data uses, and planning toward future applications of the data. In other words, the contribution of organizations such as the March of Dimes can be beneficial from the design of data utilization plans through to the reporting of actual outcomes.

Advisory committees with agency, organizational, and public representation, including political officials, are another means of obtaining input regarding uses of birth defects surveillance data. The available computer technologies such as listservs and webpages decrease the need for face-to-face meetings among interested parties, while increasing the frequency with which information about a program can be communicated and feedback solicited. New ideas about potential uses to which a program's data can be put and the resources needed to accomplish programmatic activities can be shared with advisory committee members for immediate feedback as to the feasibility of the idea and its potential for success.

Programs should create opportunities for formal input from advisors on a regular basis to ensure the availability of support in times of fiscal crises. Advisory group members' knowledge of surveillance data collection activities and uses for surveillance data can be critical to securing resources for a program in times when limited resources require justification for program continuation.

**Public involvement.** Birth defects surveillance programs generally have not engaged consumer and parent participation other than through advisory group representation. Members of the public, including parents of children identified through these programs, are often not well informed about public health surveillance activities. If not already doing so, birth defects surveillance programs should engage both consumers – here defined as adults with birth defects, and parents and caretakers of children with birth defects – in the planning and implementation of any and all programmatic changes. There are a number of advocacy and parent support groups, such as the Spina Bifida Association of America, Family Voices, and the Alliance of Genetic Support Groups, that can play important roles in planning and conducting birth defects surveillance programs.

Programs should embrace the concept of *participatory action research* (PAR) (Whyte, 1991). PAR is a way to obtain public input into programmatic activities from design though dissemination of results. PAR ensures input from the community members most affected by potential data uses. Again, as discussed with respect to advisory group input, computer technology can be immensely beneficial in obtaining feedback on new initiatives and more importantly in soliciting input about programmatic activities from community members.

## 1.3.4  Leveraging Resources

For birth defects surveillance, as for other public health surveillance programs, the ways in which data are used will influence continued availability of program resources. In the age of evidence-based medicine and increased emphasis on demonstrating program efficacy for continued support, birth defects surveillance programs should work toward expanding data use. Fiscal trends in states suggest that the likely survivors in times of increasingly fewer tax-based resources will be programs that adapt by reinventing themselves in terms of data utilization. While emphasizing the application of surveillance data to improving human services and then evaluating their impact will not ensure the survival of a program, it should increase its chances.

Surveillance programs (particularly those housed in health departments) may be given adequate resources for data collection and management, but often do not have adequate personnel or resources for data analysis beyond simple descriptive reporting. Program managers and staff often use lack of adequate resources as an excuse to minimize the number of new initiatives they undertake, but this may well be a short-sighted approach. We have already discussed the importance of partnerships, advisory groups, and public involvement in increasing the probability of acquiring additional resources. While programs must, realistically, work within the limits of available resources, partnerships with agencies and institutions can represent a means to extend and enhance programmatic achievements. Universities, particularly those with public health training programs or medical schools, will have faculty and trainees potentially interested in birth defects. What a birth defects surveillance program lacks in resources for data analysis and research often can be compensated for through partnerships with interested faculty members willing to direct student theses and dissertations that focus on birth defects. New programs and programs that do not currently have such partnerships should give serious consideration to forming these types of collaborations, which can lead to additional resources through contracts and grants.

## 1.3.5  Considering Record Linkage

As touched upon in Section 1.3.1, the potential to link records and consolidate information from different databases contributes to the public health applications of surveillance data. For example, data from birth defects surveillance programs can be used to determine whether reported cases of birth defects represent existing cases in other databases, such as records in interdisciplinary clinics and schools with programs to assist children with disabilities. The ability to link records on individuals in more than one database can

streamline the treatment and referral processes and help maintain a certain level of fidelity and trust in prevalence data. Record linkage can streamline the research process by consolidating several different databases. Another utility of record linkage is the ability to supply crucial data required for various research efforts. Specifically, the data located in one database can be used to elicit information from a second.

# 1.4  Uses of Surveillance-based Birth Defects Data

Most US states have implemented birth defects surveillance programs that monitor and disseminate information regarding birth defects. Public health staff and researchers nationwide have used these data in a variety of ways. The actual and potential uses of birth defects data are discussed and exemplified in the following sections. Data from birth defects surveillance programs can be employed to define the magnitude of a problem, to support research, as well as to assess the efficacy of prevention and treatment, playing a key role in the core public health function of *assessment* (Institute of Medicine, 1988).

For convenience, the uses of birth defects surveillance data can be grouped into the following categories:

> ➢ Prevalence studies

> ➢ Epidemiologic studies

> ➢ Mortality assessment

> ➢ Needs assessment for services

> ➢ Referral to clinics and services

> ➢ Program evaluation

> ➢ Clinical research

Each of these categories of use will be discussed in further detail below. While comprehensive coverage of works in each of these categories is beyond the scope of this chapter, we have selected published studies that exemplify the kinds of research that can be conducted in each category. Naturally, what an individual program is able to do depends ultimately on its goals and objectives. When programs are faced with limited resources to conduct data analysis and research, collaborations with universities or contractors with epidemiologic expertise can often yield mutually satisfactory results.

## 1.4.1  Prevalence Studies

A common use of data produced by birth defects surveillance programs is to describe the occurrence (***prevalence at birth***) of the monitored conditions. Such uses of surveillance data include identification of trends in birth defects occurrence, definition and evaluation of clusters of congenital defects, and assessment of the need for resources and interdisciplinary services.

Khoury et al. (1986) is an example of an early study by a state surveillance program that used data in this way. This study was the outcome of a partnership between the state health department-based surveillance program and university-based researchers. Khoury and co-workers used 1984 data collected from the Maryland Birth Defects Reporting and Information System (BDRIS) to determine rates of occurrence and to identify potential trends. The prevalence at birth of "sentinel" defects, as determined from the Maryland BDRIS data, was 52.7 per 10,000 qualified births. Furthermore, trends in the occurrence of several specific birth defects were identified. The study revealed an association of low birth weight and prematurity with birth defects, an association between twinning and the rate of birth defects, racial differences in the prevalence of neural tube defects, and a relationship between Down syndrome and advanced maternal age. The importance of determining prevalence at birth is that the data can be compared with similar data collected from other birth defects monitoring systems to assess differences in rates that may exist among

surveillance areas and to direct further research efforts in an attempt to identify the reasons behind the differences.

An example of a more recent prevalence study is one reported by Ethen and Canfield (2002), who investigated the effects of including elective pregnancy terminations, prior to 20-weeks gestational age, on birth defects prevalence. In many surveillance programs, pregnancies ending prior to 20 weeks gestational age, including elective terminations, are not ascertained to be included among reported cases. The researchers concluded that when elective terminations at less than 20 weeks were considered, the prevalence of some congenital defects increased, while others remain unchanged. Specifically, anencephaly, spina bifida, and encephalocele increased substantially, while cleft palate did not change. The underlying assumption is that pregnancies resulting in debilitating or potentially terminal conditions are more likely to be terminated electively than those resulting in less severe or treatable malformations.

These two studies show the potential usefulness of prevalence data to reveal important trends and associations. These types of data often provide the impetus to initiate subsequent research. A consequence of producing birth defects prevalence data is that it frequently opens other avenues of exploration. Quite simply, without basic prevalence data to lead inquiry, many research investigations never would be conceptualized, much less carried out.

## 1.4.2  Epidemiologic Studies

Cases from birth defects surveillance programs have played key roles in conducting etiologic research in the United States and internationally. Cases from the Metropolitan Atlanta Congenital Defects Program (MACDP) have provided the basis for numerous research studies that have shed light on both the causes (Khoury et al., 1982; Oakley, 1984; Erickson, 1991; Dott et al., 2003) and prevention (Roberts et al., 1995; Olney et al., 2002) of birth defects. Similarly, the California Birth Defects Monitoring Program (CBDMP) has been the source of cases and etiologic research that has resulted in dozens of seminal papers on a variety of specific congenital malformations and their risk factors (Croen et al., 1991; Shaw et al., 1996; Ritz et al., 2002). Other state programs have contributed cases for epidemiologic studies leading to a growing number of multi-state investigations of specific risk factors (for example, Olney et al., 1995). Reference to the annual report of the International Clearinghouse for Birth Defects Monitoring Systems (International Centre for Birth Defects, 2002) demonstrates the large number of studies based on individual surveillance systems and collaborative projects among programs.

An example of an early methodological study, based on surveillance data, is a study by Khoury et al. (1988) that assessed the patterns of maternal residential mobility between conception and delivery. The authors' rationale was that most epidemiologic studies of environmental risk factors are based on maternal residence at the time of delivery. Such an assessment would be invalid, however, in instances where the mother had moved prior to delivery. The researchers examined demographic data for infants born with congenital defects. Both the demographic data as well as the birth defect data were taken from the Maryland BDRIS in 1984. The researchers concluded that, on average, 21% of all mothers whose pregnancies resulted in a child affected by one of the birth defects included in the Maryland BDRIS had moved between conception and delivery. This is important for several reasons. First, it is well understood that the effects of environmental teratogens occur early in embryogenesis; so assessing the influence of environmental exposures must be related temporally to conception. In addition, potential exposures to teratogenic environmental factors could possibly be misrepresented if examined at delivery rather than around the time of conception. Maternal mobility could also skew data regarding geographic clusters of birth defects. This study was made possible because the Maryland BDRIS determines the residence of the mother not only at the time of delivery, but also at the time of conception. This is an important aspect of the Maryland BDRIS that is not common to all birth defects surveillance programs.

Examples of surveillance-based etiologic research of associations between maternal exposures and congenital defects include studies of cigarette smoking and orofacial clefts. Among the earliest research efforts investigating this association was a study by Khoury et al. (1987) using data collected in 1984 from the Maryland BDRIS. A case-control study examined the history of cigarette smoking among mothers of infants with orofacial clefts and a group of control mothers. The researchers concluded that odds ratios for cleft palate (2.39, CI 1.04-5.45) and cleft lip with and without cleft palate (2.56, CI 1.13-5.78) were increased for women who smoked. Furthermore, the researchers identified a dose-response effect. Khoury and his co-workers also took into account possible confounding factors, including race, gender, residence, maternal age, parity, and several pregnancy exposures or complications. None of these affected the results significantly. This is a classic example of how surveillance-based birth defects data can be used to examine etiologic factors through the use of simple epidemiologic techniques. Sometimes the importance of earlier epidemiologic studies is not appreciated when comparing them to more recent research. It is worth noting that the association between maternal cigarette smoking and orofacial clefts has been corroborated through more recent studies using several surveillance-based investigations. The paper by Khoury et al. (1987) has been cited in many contemporary research publications (Shaw et al., 1996; Lieff et al., 1999).

Some states have used surveillance data to look for associations between environmental factors that are known to cause specific birth defect syndromes and other birth defects. For example, maternal alcohol use during pregnancy is a known cause of the fetal alcohol syndrome, but its role in more common, isolated, craniofacial defects is not well understood. A population-based, case-control study of orofacial clefts was conducted in Iowa based on births from 1987-1991 (Munger et al., 1996). Cases were identified by the Iowa Birth Defects Registry and classified as having a cleft lip with or without cleft palate (CLP) or cleft palate only (CP) and as to whether the cleft was isolated or occurred with other birth defects. Controls were selected from normal Iowa births. Maternal alcohol use during pregnancy was classified according to self-reported drinks consumed per month. Compared to women who did not drink alcohol during pregnancy, the relative odds of isolated CLP rose with increasing level of maternal drinking as follows: 1-3 drinks per months, 1.5; 4-10 drinks per month, 3.1; more than 10 drinks per month, 4.7 (chi-square test for trend, P = 0.003). Adjustment for maternal smoking, vitamin use, education, and household income did not substantially alter the results. No association was found between alcohol use and isolated cleft palate or clefts in children with multiple birth defects. Based on these data, alcohol use during pregnancy may be a cause of isolated cleft lip with or without cleft palate.

As described, epidemiologic investigation is an important area of research supported by birth defects surveillance data. In the past, this research effort primarily focused on environmental exposures as possible etiologic factors. However, with the recent explosion of molecular genetics and a more thorough understanding of molecular biology, the avenues of epidemiologic investigation have widened significantly. Investigators now have an enhanced ability to examine the contributions of both maternal and fetal genotypes to disease risk. Examination of the interplay between genetic predispositions/susceptibilities and environmental exposures is a growing area of study, with potential major implications with respect to understanding birth defects etiology. This is illustrated by the genetic component of the National Birth Defects Prevention Study, a multicenter case-control study being conducted by CDC and participating state surveillance programs (Yoon et al., 2001; Rasmussen et al., 2002).

Continuing with the study of the association between smoking and clefts, epidemiologic studies have focused on the relationship between certain alleles of a transforming growth factor and maternal cigarette smoking with regard to risk of orofacial clefts. The most promising associations are seen in polymorphisms of the transforming growth factor alpha (TGFα) gene taq1 and maternal cigarette smoke exposure. An example is a study by Hwang et al. (1995), supported by surveillance data, that examined this association. The data on infants born with orofacial clefts were taken from the Maryland BDRIS. The Maryland BDRIS was not only able to supply cases of orofacial clefts, but also information about maternal prenatal behaviors, including maternal smoking during pregnancy. Cases were genotyped and screened for the rare C2 taq1 polymorphism.

The researchers concluded that the C2 genotype, combined with maternal smoking, significantly increased the risk of orofacial clefts. Using data collected through a birth defects surveillance program, they were able to identify a possible interaction between an environmental exposure and a genetic predisposition with respect to risk for orfacial clefts.

Studies like this represent another generation of epidemiologic research. The power of these molecular epidemiologic studies lies in their ability to elicit possible etiologies of birth defects beginning with prevalence data, demographic information, and biologic samples. While the epidemiologic research methods have evolved significantly, the ultimate goal of these studies has remained constant: namely, to identify, define, and associate birth defects with possible etiologic factors. The development and application of molecular genetic methods serve as stepping stones to future research based on surveillance-derived cases.

### 1.4.3  Assessing Mortality Associated with Birth Defects

A 1995 Texas study assessed survival rates for selected birth defects among babies born between January 1, 1995 and December 31, 1997, by linking two databases: the state's active birth defects registry and the infant death registry (Nembhard et al., 2001). The goal of the study was to determine mortality among cases with various birth defects identified through the birth defects surveillance system by matching those cases against infant death files. Specifically, the researchers found the birth defects with the lowest survival were anencephaly (0%) and trisomy 13 (7.4%), while the birth defects with the highest survival were gastroschisis (92.9%) and trisomy 21 (92.3%). These survival data were only for the first year of life.

Another example of a mortality study is that carried out by Druschel et al. (1996), who examined infant mortality among children with orofacial clefts, comparing their mortality rates to those of children with no congenital malformations. In the absence of malformations in other organ systems (isolated clefts), mortality was not increased among children with orofacial clefts. The study revealed, however, that many children with orofacial clefts have other malformations that increase their risk of death. These findings suggest the need for careful evaluation of possible additional malformations among children with orofacial clefts as these children may be at higher risk of death.

### 1.4.4  Estimating the Need for Services

Estimating service needs based on birth defects prevalence has significant direct social consequences. Accurately predicting the demand for various interdisciplinary clinics and social and educational services is critical for children born with birth defects. Estimating future service needs allows for capacity building to ensure that necessary resources will be accessible and that appropriate professionals will be available to provide the services.

Brewster et al. (1992) linked demographic and diagnostic data from 1980 – 1982 in a birth defects surveillance program database (the Arkansas Reproductive Health Monitoring System) with education databases. The data were first used to estimate the percentages of infants with specific birth defects who were at risk for developmental disabilities and mental retardation. Once prevalence rates were determined, two clinicians estimated the various services that would be needed by children with the various birth defects most likely to contribute to developmental disabilities. This included academic and other services these infants would require as they matured. The researchers estimated that between 32% and 56% of all children in schools who were classified as mentally retarded were also identified by the Arkansas Reproductive Health Monitoring System.

This study showed that recognition of children with mental retardation, who were also identified years earlier as having congenital defects, allowed researchers to refine their estimates of the birth defects that will contribute most significantly to mental retardation in school-aged children. This is useful in improving the ability of health care professionals to predict accurately future needs of the current cohort of newborns with birth defects.

## 1.4.5  Referral to Services

Information collected as part of birth defects surveillance can be used to refer specific children and their families to appropriate services. Established referral networks serve as a resource for children and their families to learn about available medical services, community programs, and social support. Affected children and their families can be connected with appropriate services in a timely fashion.

Many papers have been written detailing the process of identification and ultimate service referral. One of the first papers on this topic comes from the Maryland BDRIS, where investigators examined the referral of children identified with orofacial clefts through the surveillance program to the Maryland Crippled Children's Service Program in the 1960s (White, 1981). This study examined referral rates to services. A more recent paper on referral and treatment patterns for orofacial clefts comes from Florida, where referral and treatment patterns of live-born Florida infants diagnosed with orofacial clefts identified through the Florida Birth Defects Registry were determined (Williams et al., 2003).

Another example is a paper describing service referrals in Colorado that use birth defects data taken from their birth defects surveillance program (Montgomery and Miller, 2001). The Community Notification and Referral Program (CNRP), operating from within the state's health department, uses birth defects data to link affected infants with an organization that can refer them and their families to agencies and interdisciplinary clinics. In 1998, 259 families were referred for services as a result of being identified through the birth defects registry. There are a number of services to which patients are commonly referred, including developmental screening and evaluation, public health programs, early intervention programs, financial assistance, parenting classes, medical services, recreational programs, and family support groups. Additionally, the effectiveness of this program has been assessed through the use of surveys and questionnaires.

A review of the use of surveillance data relative to provision of early intervention services can be found in a recent paper on identification and referral programs by Farel and colleagues (2003). Having agencies use birth defects data to link patients with appropriate services is a critical data use that has immediate and direct impact on the lives of those affected. Although epidemiologic and laboratory efforts may illuminate etiologies and possible preventive measures for future use, the fact remains that effective therapeutic efforts in the present can significantly improve the lives of persons with birth defects; scientific studies take years to complete and primarily aid future patients. Meanwhile, there are people who require immediate assistance, and service referral is an important mechanism through which they can receive that help.

## 1.4.6  Program Evaluation

Another use of birth defects surveillance data is program evaluation. Typically, this use is employed subsequent to research efforts, many of which were also based on surveillance data and may represent a baseline from which post-intervention improvement can be measured. Program evaluation is a valuable and desired area of activity with important scientific, academic, social, and policy applications. Program evaluation can focus on different aspects of surveillance program activities, such as case referrals and clinical interventions. First, evaluating a program for service referral can give investigators information on the efficacy of their referral agencies or the appropriateness of the services offered. Second, evaluating clinical

intervention studies allows researchers to assess both the effectiveness of the intervention and the validity of their clinical assumptions.

One study involving program evaluation of a clinical intervention using birth defects surveillance data was performed in Nuevo León, México (Martinez de Villarreal et al., 2002). The investigators assessed the effectiveness of a folic acid campaign in reducing the occurrence of neural tube defects. Investigators first developed a base rate for neural tube defects prior to administration of the folic acid and counseling services. An intervention was then initiated that included five mg of folic acid supplementation per week, as well as counseling and social services. After 28 months, the rates of neural tube defects were ascertained again. From the baseline in 1999 (95 cases of neural tube defects), neural tube defects declined by 50% in the next two years (59 cases in 2000, 55 cases in 2001).

This study illustrates the wide range of uses for birth defects surveillance data in evaluation. First, data were used to assess an initial rate of neural tube defects and at the conclusion of the intervention to assess its appropriateness and efficacy. In addition, the study demonstrated the efficacy of folic acid supplementation in reducing the occurrence of neural tube defects and the fact that the methods of administration were clinically appropriate and effective.

In another example of the use of surveillance data in program evaluation, Meyer and Oakley (2000) used data from the North Carolina Birth Defects Monitoring Program to assess the folic acid fortification mandates of the federal government. The results suggested that the decline in the occurrence of neural tube defects was marginal and not the predicted 50% decrease. The authors' recommendation was to increase the folic acid fortification standards on a national level.

## 1.4.7  Clinical Research

Recently a group of researchers in the United Kingdom carried out surveillance in one Health Region using multiple sources to identify all individuals with specific conditions (Holland et al., 1998; Whittington et al., 2001). The condition the researchers captured that is of greatest relevance to birth defects is Prader-Willi syndrome (PWS). The first step was to conduct population-based surveillance in the Cambridge Health District (eight English counties with a base population of 280,000 individuals) (Whittington et al., 2001). The birth prevalence of PWS was estimated to be 1:22,000 and the mortality rate more than 3% per year. The next step was to carry out population-based clinical research about phenotypic features, including the prevalence of behavioral and health problems in PWS. Clarke et al. (2002) reported the prevalence of compulsive and similar behaviors among individuals with PWS in this population. Butler et al. (2002) presented data on the prevalence of comorbidities in PWS that could contribute to reduced life expectancy for persons with this condition. Most recently Holland et al. (2003) reported on the specific behaviors that comprise the proposed behavioral phenotype in PWS.

Although this work represents a non-traditional method of surveillance compared to state surveillance programs in the United States, it is important in terms of clinical research that has been conducted and the potential for conducting similar work using state-based surveillance data. A major advantage of these clinical studies is that they are population based. Even though all individuals identified through the surveillance work did not participate in the collection of behavioral and health data, the sample of individuals with PWS who participated in the clinical research can be compared to the total population of ascertained individuals to evaluate how representative the sample is of individuals in the Health Region who have PWS. Usually this is not possible using common methods of clinical research.

# 1.4.8  Using Birth Defects Data in the Future

For the data collection process itself, abstracting methods continue to be refined. Quality assurance procedures and ongoing training, aimed at increasing data accuracy and validity, are being implemented in order to assure a certain level of fidelity and trust in the data collected. Improving and standardizing these procedures are among the objectives of these guidelines.

The future uses of birth defects surveillance data are related to scientific advances in other areas of research. Several developing scientific fields will utilize birth defects data in novel ways. For example, our understanding of molecular biology has developed exponentially. With the successful sequencing of the human genome, the resulting information will provide significant information on genetic factors influencing disease risk. Consequently, these discoveries will be investigated for certain genetic regulatory mechanisms and environmental triggers. Using birth defects surveillance data, investigators will be able to examine possible environmental exposures that are etiologically associated with birth defects in the presence of a particular genetic background. Discoveries of gene-environment interaction will allow researchers to understand etiologic associations. Additionally, the way in which these environmental conditions regulate gene expression will further illuminate these associations.

Future advancements in research supported by birth defects data will benefit from the integration of electronic medical records. Current methods for obtaining birth defects data are laborious. They frequently involve extensive abstraction procedures, reporting cases to the respective health department, entering the abstracts into the database, and categorizing the data. These methods will be streamlined, as medical records and birth defects surveillance systems are maintained electronically. This will have two general effects: first, it will help facilitate the abstraction process by eliminating bulky charts containing information not necessarily applicable to the birth defects surveillance program and, second, it will allow researchers to access these information-rich databases more quickly and efficiently. Furthermore, database search functions will allow researchers to identify cases of interest instantaneously without physically sifting through thousands of reported cases. Ultimately, researchers will be given access to the electronic surveillance database. Using surveillance systems researchers will able to search for cases of interest and refine their cohort by filtering cases by demographics, location, or maternal prenatal behaviors. A study that currently takes weeks to conclude would be completed in the course of several hours.

Researchers continuously find new and exciting uses of the data from birth defects surveillance programs. Given the breakthroughs achieved through earlier studies using surveillance data, the possibilities of future revelations are staggering. In their relatively short existence, birth defects surveillance programs have changed the ways in which professionals view birth defects both clinically and socially. The importance of the impact of birth defects surveillance programs on clinical and public health research cannot be overstated, as such research is revolutionizing the way scientists, clinicians, and health care professionals approach, treat, and manage infants affected by birth defects, while also advancing our understanding of preventive measures.

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# *Chapter 2*

# *Legislation*

# Table of Contents

# Appendices

# Acronyms

CDC        Centers for Disease Control and Prevention

CE          Covered Entity

FERPA     Family Educational Rights and Privacy Act

DHHS      Department of Health and Human Services

HIPAA      Health Insurance Portability and Accountability Act

OCR        Office of Civil Rights

PHA        Public Health Authority

PHI         Protected Health Information

PR          Privacy Rule

# 2.1  Introduction

Legislation supporting birth defects surveillance activities is important for several reasons. For example, legislation serves to define the purposes of surveillance activities, specifies the kinds of data or information to be collected, and designates who is responsible for this activity. The first birth defects legislation was passed in New Jersey in 1926. During the past 20 years, the majority of states have enacted legislation mandating reporting of birth defects to the health department. As of April 2004, 41 states had existing legislation or rule related to birth defects surveillance.

Although there are a number of advantages to having legislation that supports birth defects surveillance, some limitations may also accrue. Early in their planning process, new or relatively new state programs should consider both the benefits and the possible limitations of birth defects surveillance legislation. At this early stage in a program's development, the opportunity exists to advocate for and perhaps assist in crafting clearly written, effective legislation that will serve the needs of the program in years to come.

In this chapter we discuss the distinction between the terms 'legislation', 'regulation', and 'authority' (Section 2.2); key elements of model legislation (Section 2.3); and federal laws that can affect birth defects surveillance programs (Section 2.4). References cited in this chapter may be found in Section 2.5.

To assist those interested in drafting or revising state legislation concerning birth defects surveillance, we append sample legislation from Arkansas, California, New Jersey, New York, Oklahoma, and Texas (see Appendix 2.1). Additional appendices include a table of birth defects legislation (Appendix 2.2), definitions used to determine 'covered entity' status under the Privacy Rule (Appendix 2.3), and an excerpt from the text of the Health Insurance Portability and Accountability Act (Appendix 2.4).

## 2.2  Legislation, Regulation, and Authority

'Authority' to mandate the reporting of birth defects to a surveillance program can be granted through 'legislation' or 'regulation'. In this section we explore distinctions among these and other related terms.

***Legislation*** is the process of enacting laws by a legislative body. The type of law depends on the legislative authority granted. State legislatures and Congress have complex processes to enact legislation. These processes vary from state to state. In the simplest terms, state and federal legislative bodies create ***statutory law***, also called a ***legislative act***. These terms denote a bill that has been passed by one house in a bicameral legislature. After enactment by both houses, the terms 'law' and 'act' may be used interchangeably. A ***statute*** is the formal written enactment of a legislative body, whether federal, state, city, or county.

State and federal agencies are arms of the executive branch of the government. Such agencies have broad power granted under state and federal law to make ***regulations*** that govern activities for which they are responsible. Leaders of public health and other state agencies are not elected, but rather appointed by the executive, usually the governor of a state. Under current public health legislation, public health authorities may make regulations that can be mandatory, voluntary, directive, or prohibitive.

In sum, the term 'legislation' refers to a law enacted by an elected body, whereas 'regulations' are created by agencies.

For an agency, such as a state public health department, to establish a regulation mandating the reporting of birth defects, the health department must have the power or the ***authority*** to establish that type of regulation. This power can be based on state law or on an act of the executive power of the state, such as the governor. If the health department does not already have such regulatory power, then two options exist, namely, proposing a state law mandating birth defects reporting or proposing a state law granting authority to the health department to establish a regulation.

A state reporting law is straightforward and more democratic because it is enacted by elected representatives and gives an agency clear power or authority to do whatever the law states. However, a state reporting law also places the power to modify or change the law in the hands of the legislative body, despite the fact that the legislature may not be well informed about public health matters. Because most legislative bodies recognize the expertise of the people who run public health agencies, they generally grant them the necessary authority to conduct their work properly. Thus, the legislative bodies of many states have given the health department power to enact the regulations they deem necessary to protect the public health and welfare.

# 2.3  Key Elements of Model Legislation

Birth defects legislation should be considered early in the developmental phase of a surveillance program, if possible. This allows for legislation to be written clearly to support facilitation of surveillance activities. Language should be broad and flexible enough to cover all of the areas necessary to meet programmatic objectives, yet not to be so vague as to be confusing or meaningless. Well-written legislation that facilitates birth defects surveillance should address the key elements outlined in the Sections 2.3.1 through 2.3.8 below. These include:

- ➢ Designation of agency authority
- ➢ Purpose and priorities
- ➢ Access to data and records
- ➢ Ability to share data while maintaining confidentiality
- ➢ Terminology and definitions
- ➢ Opt-out clauses
- ➢ Advisory committee
- ➢ Funding

## 2.3.1  Designation of Agency Authority

Model state legislation for birth defects surveillance should specify the agency that has the overall grant of authority for the system. This authority usually resides within the department of health, which has the power to enact rules and regulations, establish criteria for reportable conditions, and implement and oversee procedures for reporting. In most cases, there is no need to detail the specific regulations in the legislation. However, legislation should specify that the department has the authority to enact and enforce the regulations.

## 2.3.2  Purpose and Priorities

The purpose of the program will drive decision-making about its scope and activities. The purpose will also help states define outcomes, ages to be covered, and the most important sources of data to be included. Language should clearly articulate what the system should do and what its priorities should be. For example, Hawaii's legislation contains the following language:

> *"The department of health shall establish the statewide birth defects program to:*
>
> *1)  Collect surveillance information on birth defects and other adverse reproductive outcomes;*
>
> *2)  Report the incidence, trends and causes of birth defects and other adverse reproductive outcomes;*
>
> *3)  Report information for the development of prevention strategies to reduce the incidence of birth defects and other adverse reproductive outcomes; and*
>
> *4)  Develop strategies to improve the access of children with birth defects to health and early intervention services." (Hawaii Revised Statutes, Chapter 321, §321)*

### 2.3.3  Access to Data and Records

Legislation should grant the birth defects surveillance program the authority to access hospital discharge data and medical records or to require reporting with access for follow-up as needed. Legislation that provides for access to medical records grants surveillance programs the opportunity to obtain more complete and reliable reporting of birth defects, while also ensuring that surveillance data sets are large enough to be useful to researchers and service providers.

California's birth defects surveillance law states that:

> *"… The director shall require health facilities, with 15 days' notice, to make available to authorized program staff the medical records of children suspected or diagnosed as having birth defects, including the medical records of their mothers. In addition, health facilities shall make available the medical records of mothers suspected or diagnosed with stillbirths or miscarriages and other records of persons who may serve as controls for interview studies about the causes of birth defects …" (California Health and Safety Code, Part 2, Chapter 1, §103830)*

Legislation with mandated reporting should include language that allows a program to access medical records for follow-up to ensure data quality. For example, New Jersey's legislation stipulates that:

> *"The Commissioner of Health, in consultation with the Public Health Council, shall require the confidential reporting to the Department of Health of all cases ..." (New Jersey, Chapter 26:8-40.2)*

> Then, in its regulations, the department of health addresses the follow-up component:

> *"Every health facility and independent clinical laboratory shall allow access to, or provide necessary information on infants with birth defects …" (New Jersey Rules, Chapter 20, Subchapter 1, 8:20-1.2j)*

### 2.3.4  Ability to Share Data While Maintaining Confidentiality

Legislation should specify who can have access to the data and how the confidentiality of the data will be protected. Many states have specific guidelines regarding the use of data for research purposes, and legislation may stipulate that persons who violate rules about data use or confidentiality are subject to civil penalties. For example, Texas' legislation states that:

> *"(a) Access to the central registry information is limited to authorized department employees and other persons with a valid scientific interest who are engaged in demographic, epidemiological, or other studies related to health and who agree in writing to maintain confidentiality.*

> *(b) The department shall maintain a listing of each person who is given access to the information in the central registry. The listing shall include:*

> *(1) the name of the person authorizing access;*

> *(2) the name, title, and organizational affiliation of each person given access;*

> *(3) the dates of access; and*

> *(4) the specific purpose for which the information was used.*

*(c) The listing is public information, is open to the public under the open records law, Chapter 424, Acts of the 63rd legislature ... and may be inspected during the department's normal hours of operation." (Texas Health and Safety Code, Subchapter D, § 87.062)*

## 2.3.5  Terminology and Definitions

Terminology should be defined clearly, but not in an overly narrow or restrictive manner. For instance, it is more effective to specify surveillance for the general category of 'birth defects' rather than for a narrow or finite list of specific defects such as spina bifida, anencephaly, Down syndrome, and so on.

The state of California defines **birth defect** as:

*"... any medical problem of organ structure, function, or chemistry of possible genetic or prenatal origin." (California Health and Safety Code, Chapter 1, §103825 [a])*

The legislation also specifies that **health facilities** are:

*"... general acute care hospitals, and physician-owned or operated in clinics ... that regularly provide services for the diagnosis or treatment of birth defects, genetic counseling, or prenatal diagnostic services." (California Health and Safety Code, Chapter 1 §103830)*

Broader language is more flexible, inclusive, and comprehensive than narrow language and allows for future modifications in program priorities or activities, whereas revising or amending narrowly written legislation can be a lengthy and difficult process. Legislating surveillance of specific defects may prove to be problematic in the long run as conditions change or as it becomes necessary or desirable to collect data on additional defects or combinations of defects. Definitions should be in the agency's regulations, not in the enabling legislation.

## 2.3.6  Opt-out Clauses

In most cases, parental consent is not required in order for a surveillance program to be able to collect data on children with birth defects from schools or health care providers. Some states, however, do require written consent from parents. Because obtaining written consent from parents can be problematic, some states handle this issue with an opt-out clause.

For example, Ohio's opt-out clause states that the health department shall adopt rules that will:

*"Establish a form for use by parents or legal guardians who seek to have information regarding their children removed from the system and a method of distributing the form to local health departments ... and to physicians. The method of distribution must include making the form available on the internet." (Ohio, House Bill No.534, § 3705.35[e])*

Opt-out clauses assume consent unless otherwise stated, allowing the surveillance program to collect data unless a child's parent or legal guardian submits a written request that their child's information be removed from the surveillance system. Opt-out clauses eliminate the need for providers and surveillance program staff to obtain written consent from parents and contribute to more complete data collection.

## 2.3.7  Advisory Committee

States that consider the potential impact of legislation in the planning stages of their programs have the advantage of influencing the development of legislation that can support the overall growth and development of the program. In some states, for example, legislation calls for establishing an advisory committee to provide guidance and oversight for the design and implementation of birth defects surveillance. Advisory committees made up of experts from fields such as epidemiology, hospital administration, biostatistics, maternal and child health, and public health can develop recommendations and provide the expertise necessary to ensure that the program meets well-defined standards and goals. Some advisory committees also include parents of children with birth defects. For example, Vermont's legislation calls for the establishment of a 'birth information council'.

> *"(a) The commissioner of health, in collaboration with the March of Dimes, shall appoint a birth information council to advise on the need for and implementation of a comprehensive, integrated, and confidential birth information system.*
>
> *(b) The council shall be composed of nine members, who represent each of the following interests:*
>
>> *(1) obstetrics and gynecology;*
>>
>> *(2) pediatrics and genetics;*
>>
>> *(3) the Vermont Children's Health Improvement Program;*
>>
>> *(4) a parent of a child with special medical needs;*
>>
>> *(5) an adult with special medical needs;*
>>
>> *(6) the commissioner of health, or his or her designee;*
>>
>> *(7) the Family, Infant, and Toddler Program;*
>>
>> *(8) the Vermont chapter of the March of Dimes; and*
>>
>> *(9) the Vermont Program for Quality Health Care." (Vermont, H.636, § 5084)*

## 2.3.8  Funding

Cost can be an impediment to establishing a birth defects surveillance system.

Some states have legislation mandating special funds to cover the operating expenses of their birth defects surveillance program. Sources of special funds include marriage license, birth certificate, and newborn screening fees. For example, Iowa's special fund is supported through birth registration fees:

> *"It is the intent of the general assembly that the funds generated from the registration fees be appropriated and used as follows:*
>
>> *(1) Beginning July 1, 2003, and ending June 30, 2005 ... five dollars of each fee for the birth defects institute central registry established pursuant to section 136A.6.*
>>
>> *(2) Beginning July 1, 2005, ... ten dollars of each fee for the birth defects institute central registry established pursuant to section 136A.6." (Iowa Code, §144.13A)*

In summary, paying due consideration to how legislative language can affect the design, implementation, and operation of the surveillance program and further ensuring that the birth defects surveillance program itself has input into legislative language from the time the program is established can have a significant impact on the long-term success of the program.

## 2.4  Federal Laws

A broad range of federal laws must be considered when planning state legislation, local regulations, or new birth defects surveillance programs. While state laws will govern most of the activities of the program, the impact of federal privacy regulations must also be considered. Depending upon how the birth defects program is structured, it may need to follow the Health Insurance Portability and Accountability Act (HIPAA) discussed in Section 2.4.1, the Family Educational Rights and Privacy Act (FERPA) discussed in Section 2.4.2, and other federal regulations such as the Privacy Act (Section 2.4.3), the Public Health Service Act (Section 2.4.4), and the Freedom of Information Act (Section 2.4.5). The following sections provide basic information about major federal laws that must be considered when setting up a birth defects surveillance program. In Section 2.4.6 we discuss the supportive role that can be played by state health officials or staff of the Centers for Disease Control and Prevention (CDC) in conjunction with planning state legislation or local regulations for birth defects surveillance programs.

## 2.4.1  Health Insurance Portability and Accountability Act (HIPAA)

The Health Insurance Portability and Accountability Act was passed in 1996 to protect consumers of the insurance industry. The Privacy Rule (or PR, also referred to as the Rule), which implements the Act, became effective on April 14, 2001, and creates national standards to protect an individual's medical records and other personal health information, known as ***protected health information*** (or PHI). The Rule gives patients more control over their health information and establishes appropriate safeguards that health care providers and other ***covered entities*** (or CEs) must establish to protect the privacy of PHI. Violators are subject to civil and criminal penalties if they violate patients' privacy rights as stated in the Privacy Rule. The Rule allows for disclosure of some forms of data for activities carried out by ***public health authorities*** (or PHAs) but limits release of information to the minimum necessary for the purpose of the disclosure. In addition, the covered entity may rely on the public health authority for what constitutes the 'minimum necessary'.

The Privacy Rule requires health care providers who are covered entities to provide information to patients about their privacy rights and how their information can be used, to adopt clear privacy procedures and adequately train employees in these procedures, and to designate an individual to be responsible for seeing that the privacy procedures are adopted and followed. Privacy protections should not, however, interfere with a patient's access to health care or the quality of health care delivered.

### Basic Provisions of the Privacy Rule That Affect Birth Defects Reporting

A state, county, or local health department that performs functions that make it a covered entity, or otherwise meets the definition of a covered entity, may elect to call itself a ***hybrid entity***. For example, a state Medicaid program is a covered entity (i.e., a health plan) as defined in the Privacy Rule. Some health departments operate health care clinics and thus are health care providers. If these health care providers transmit health information electronically, in connection with a transaction covered in the HIPAA Transactions Rule, they are covered entities.

Most of the requirements of the Privacy Rule apply only to the hybrid entity's health care provider component(s). If a health department elects to be a hybrid entity, there are restrictions on how its health care component(s) may disclose protected health information to other components of the health department. Birth defects surveillance components that provide genetic counseling and other types of

health care services will most likely be required to comply with the Rule's 'covered entities' provisions, if they bill electronically for their services. (See 45 C.F.R. § 164.504 (a) – (c) for more information about hybrid entities.)

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*For further information, see the definitions of 'covered entity', 'health care provider', 'health plan', and 'health care clearinghouse' in 45 C.F.R. §160.103. See also, the "Covered Entity Decision Tools" posted at:*

**http://www.cms.gov/hipaa/hipaa2/support/tools/decisionsupport**

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## Uses and Disclosures for Which an Authorization or Opportunity to Agree or Object Is Not Required

Section 164.512 of the Privacy Rule sets forth the conditions under which a covered entity, as defined previously, may disclose protected health information without the individual's consent or authorization. Below is a discussion of the application of the Rule to the birth defects surveillance system. The actual text of the regulation can be found in Appendix 2.4.

**Consent and notice.** The US Department of Health and Human Services (DHHS) made changes to the Privacy Rule effective August 14, 2002, to protect privacy while eliminating barriers to treatment. The notice requirement was strengthened, making consent for routine health care delivery purposes optional. The Rule requires covered entities to provide patients with notice of a patient's privacy rights and the privacy practices of the covered entity. The strengthened notice requires direct treatment providers to make a good faith effort to obtain patients' written acknowledgement of the notice of privacy rights and practices. The modified Rule removes mandatory consent requirements while providing covered entities with the option of developing a consent process that works for that entity. The Rule also allows consent requirements already in place to continue, but does not mandate any particular standard.

In states where data collection for birth defects surveillance is ongoing and there is no mandatory reporting law, it would be helpful to approach the data source with a request to have the public health authority listed in the privacy notice that is provided to patients. Note, however, that this does not circumvent the accounting provisions of the Rule for the covered entity.

**Mandatory reporting – 'Required by law' versus 'permitted'.** Extensive discussion has ensued within the public and private health care sectors regarding the need for mandatory reporting laws in states in order for birth defect surveillance programs to collect data. Note that this section of the Rule, §164.512, has two subsections.

> (a) Standard: uses and disclosures required by law.

> (b) Standard: uses and disclosures for public health activities.

Subsection (a) is the provision for disclosures that are required by law. If a state has a mandatory birth defects reporting law, then this is the provision in the Privacy Rule that allows that law to remain intact. The definitions in the section below explain what 'required by law' means under the Privacy Rule. However, if a state health department meets the definition below of a public health authority, then the

health department may have authority to collect birth defects data based on the department's broad grant of authority from the state to protect and promote health, prevent and control disease, or other activity.

As noted earlier, each state health department has specific authority granted it under the laws of that state. Most health departments do have some regulatory authority and can, therefore, make birth defects reporting mandatory under that authority. If the health department does not have the present authority to make such a regulation, or conduct such activity, then the health department may request that this authority be granted by the legislature, after which the department may promulgate its regulation. This method is acceptable under the Privacy Rule.

The most significant distinction to make is that subsection (a) is for reporting ***required by law***, whereas subsection (b) is for reporting ***authorized by law***. Although there is no definition of 'authorized by law' in the Rule, DHHS has sought to make this point more clearly in the Preamble to the Rule (64 FR, page 59929):

> *"When we describe an activity as 'authorized by law,' we mean that a legal basis exists for the activity. The phrase 'authorized by law' is a term of art that includes both actions that are permitted and actions that are required by law."*

In addition to this comment, new Office of Civil Rights (OCR) guidelines state:

> *"The HIPAA Privacy Rule permits disclosures that are required by law. Furthermore, disclosures to public health authorities that are authorized by law to collect or receive information for public health purposes are also permissible under the Privacy Rule."(OCR HIPAA Privacy Dec 3, 2002, http://www.hhs.gov/ocr/hipaa)*

In short, public health authorities have two different paths by which to access data for surveillance, a mandatory reporting law, or the regulatory or program authority to collect the data. (See Appendix 2.4 for OCR HIPAA privacy regulation text.)

## Data Sharing and Public Health Authorities

A public health authority that has either a mandatory reporting law, or a regulation, or some other grant of authority to collect data under the previously discussed §164.512, may use those data in any way that is permitted under state and federal law. Data that are collected by a third party, such as a university, under a grant or a contract on behalf of a public health authority, such as the Centers for Disease Control and Prevention (CDC), whether a bona fide agent or not of that health department, falls under the Privacy Rule definition of a 'public health authority':

> *"'Public health authority' means an agency or authority of the United States, a State, a territory, a political subdivision of a State or territory, or an Indian tribe, or a person or entity acting under a grant of authority from or contract with such public agency, including the employees or agents of such public agency or its contractors or persons or entities to whom it has granted authority, that is responsible for public health matters as part of its official mandate."(45 CFR §164.512(b)(l)(i))*

The Rule does not comment on what the public health authority may or may not do with the data it has legally collected. HIPAA seeks to regulate the release and use of protected health information by covered entities, and a public health authority is not a covered entity under the Rule (unless they have designated themselves as such). The grantee, holder of a cooperative agreement, or contractor conducting a public health activity, as a public health authority, as defined above, may share the data in ways that comport with all previously promulgated laws and regulations. Once data are in the possession of a public health

authority, the Rule should not be an issue for the PHA because the Rule does not regulate the use or disclosure of protected health information by a PHA.

A number of health departments have designated some of their components as covered components because they provide health care as defined in the Rule. In this case, the entire health department may be called a 'hybrid entity'. The consequences for data sharing are the same as if the designated component, or covered entity, were any other health care provider. The covered entity component of the health department can share the data it collects from individuals with the non-covered PHA component of the health department. The covered entity would have to provide the individual with the 'notice of privacy practices', which would include information to the effect that the covered entity was sharing data with other components of the health department. The covered component would also have to comply with all other provisions of the Rule, including accounting for disclosures to public health authorities. Some health departments may even provide consents to the individual based on the requirements of a state or local requirement, or to increase public confidence in the health department.

Nor is the data-sharing that flows from a public health authority to a covered entity after data collection regulated by the Privacy Rule. In cases where the public health authority wishes to refer a case to another covered entity, such as a health care provider, for a public health intervention, and the covered entity may report back its findings, remember that the definition of 'public health activities' includes the following:

> *"A public health authority that is authorized by law to collect or receive such information for the purpose of preventing or controlling disease, injury, or disability, including, but not limited to, the reporting of disease, injury, vital events such as birth or death, and the conduct of public health surveillance, public health investigations, and public health interventions."(45 CFR §164512(b)(l)(i))*

When requesting data from a covered entity, it is also important to note that even though public health authorities are exempted from the need for the authorization of the person for disclosure, the covered entity is only required to provide for the minimum necessary information to accomplish the public health mission of the PHA. In addition, the covered entity may, under the Rule, reasonably rely on the representation of the PHA for what constitutes the 'minimum necessary' information.

Some state grantees conducting birth defects and other kinds of surveillance funded by CDC have asked what kind of proof of identification (ID) they need to show to the covered entity to assure them that they are in fact a PHA and have the authority to obtain the data they seek from the CE. Business cards, government identification badges, letterhead, or other types of official representation are sufficient. Because there are so many different types of ID, DHHS chose to be very broad in this area by not specifying one type.

## Data Clearinghouses and Business Associates

Some state health departments do not carry out actual surveillance and data collection; instead, hospitals voluntarily report birth defects data to a data collection entity or clearinghouse that compiles the data and then reports the information in some form to the health department. In these cases, the hospital and clearinghouse are required to execute a data use agreement, and the covered entity must disclose this information in the privacy notice provided to patients. The clearinghouse may provide the data to the public health authority under that Rule just as the covered entity could do, without the authorization or consent of the person for purposes of public health activities, surveillance, and, under some circumstances, research.

**Surveillance versus research under the Privacy Rule.** Research is covered under a separate section of the Privacy Rule. Unlike the public health authority provisions discussed above, the research provisions do not exempt public health authorities from compliance with the Rule as research is not a public health activity as defined in the Privacy Rule. The Rule defines *research* as:

> *"A systematic investigation, including research development, testing, evaluation, designed to develop or contribute to generalizable knowledge." (45CFR 164.501)*

The recent revision in the Privacy Rule sought to bring the definition of 'research' in the Rule in line with the definition for the same term in the Common Rule. The Common Rule definition of 'research' is the one used by CDC (45 CFR 46.102[e]).

**De-identified data use.** For research purposes, a covered entity may always use or disclose health information that has been de-identified (45 CFR 164.502(d) and 164.514[a]-[c]). The Rule has a very strict definition of 'de-identified' that truly eliminates all possibility of re-identification of the individual. However, a covered entity may enter into a data use agreement with a researcher that would allow the CE to disclose to the researcher a limited data set for the purposes of research, public health, or health care operations (45 CFR 164.514[e]). A *limited data set* is specifically defined in the Privacy Rule to exclude certain direct identifiers; however, the limited data set contains sufficient geographical and vital information – such as birth, death, admit and discharge data – that it can be very useful for birth defects research. In addition, there are other specific requirements that must be included in the data use agreement. These include:

➢ Stating the permitted uses and disclosures of the limited data set

➢ Limiting who can receive the data

➢ Requiring the researcher to agree to:

— Abide by and not violate a data use agreement

— Protect the data from re-disclosure

— Report any unauthorized use or disclosure

➢ Binding all contractors or agents to the data use agreement

➢ Refraining from identifying or contacting the individual

Another way to obtain access to protected health information for research without authorization from the individual is to obtain documented Institutional Review Board (IRB) or Privacy Board approval for an exemption (45 CFR 164.512[i][l][i]). This provision is most practical for conducting records searches when use of de-identified data is not useful. There are extensive requirements under this section of the Rule that must be adhered to. Another way to obtain access to data for research without authorization of the individual is when preparing a research protocol preparatory to research (45 CFR 164.512 [i][l][ii]). Except for these limited exceptions, the disclosure or use of protected health information for research purposes requires the written authorization of the individual.

## 2.4.2  Family Educational Rights and Privacy Act (FERPA)

The Family Educational Rights and Privacy Act (20 U.S.C. § 1232g; 34 CFR Part 99) is a federal law that protects the privacy of student education records. The law applies to all schools that receive funds under an applicable program of the US Department of Education. There are some privately funded schools to which FERPA does not apply.

FERPA gives parents specific rights with respect to their children's educational records. These rights transfer to the student when he or she reaches the age of 18 or attends a school beyond the high school level. Students to whom the rights have transferred are defined as ***eligible students*** in FERPA.

➢ Parents or eligible students have the right to inspect and review the student's education records maintained by the school.

➢ Parents or eligible students have the right to request that a school correct records that they believe to be inaccurate or misleading.

➢ Generally, schools must have written permission from the parent or eligible student in order to release any information from a student's education record.

However, FERPA allows schools to disclose those records, without consent, to the following parties or under the following conditions (34 CFR § 99.31):

➢ School officials with legitimate educational interest

➢ Other schools to which a student is transferring

➢ Specified officials for audit or evaluation purposes

➢ Appropriate parties in connection with financial aid to a student

➢ Organizations conducting certain studies for or on behalf of the school

➢ Accrediting organizations

➢ Appropriate officials in cases of health and safety emergencies

➢ State and local authorities, within a juvenile justice system, pursuant to specific state law

➢ To comply with a judicial order or lawfully issued subpoena

Access to educational records can be necessary to a birth defects surveillance program for follow-up and early intervention services. FERPA generally prohibits access to educational records without the prior written consent of the parent or guardian.

**Surveillance versus research under FERPA.** For compliance with FERPA, there is no distinction made between surveillance and research. The issue in FERPA is who holds the data and who wants access to the data and why. The fact that the information in the educational record is medical, behavioral, sociological, or psychological in nature in no way alters the inability to access the information without parental consent. All information, other than student directory information, in an educational record maintained by a school, regardless of the nature of the information, is considered to be an educational record. It is important to note that HIPAA specifically states that nothing in HIPAA in any way alters FERPA. As a result, FERPA, unlike HIPAA, defines its 'protected records' simply by who possesses them, whereas in HIPAA the analysis of what is protected and the exceptions are more complex.

# 2.4.3  Privacy Act

The Privacy Act of 1974, 5 U.S.C. § 552a (2000), which has been in effect since September 27, 1975, can generally be characterized as an omnibus 'code of fair information practices' that attempts to regulate the collection, maintenance, use, and dissemination of personal information by federal executive branch agencies. However, the Act's imprecise language, limited legislative history, and somewhat outdated regulatory guidelines have rendered it a difficult statute to decipher and apply. Moreover, even after more than 25 years of administrative and judicial analysis, numerous Privacy Act issues remain unresolved or unexplored. Adding to difficulties in interpretation is the fact that many Privacy Act cases are unpublished district court decisions. The general rule contained in the Privacy Act is:

> *"No agency shall disclose any record which is contained in a system of records by any means of communication to any person, or to another agency, except pursuant to a written request by, or with the prior written consent of, the individual to whom the record pertains [subject to 12 exceptions]." (5 U.S.C. § 552a[b])*

States have adopted similar laws that should be considered when drafting legislation for a birth defects surveillance program. For further information, see the Department of Justice website at http://www.doj.gov.

# 2.4.4  Public Health Service Act

The Public Health Service Act of July 1, 1944 (42 U.S.C. §201), consolidated and substantially revised all existing legislation relating to the US Public Health Service, of which the CDC is a part. The Public Health Service Act is a broad compilation of authorities under which CDC administers national and international programs for the prevention and control of communicable and vector-borne diseases and other preventable conditions. The Public Health Service Act is only applicable to federal agencies within the Public Health Service.

Title III of the Public Health Service Act sets forth the general powers and duties of the Public Health Service. Within this title, Sections 301, 307, 311, and 317 provide CDC and other agencies within the Service with general operating authorities, including but not limited to:

➢ Encourage, cooperate with and render assistance to other appropriate public health authorities, scientific institutions, and scientists in the conduct and promotion of activities relating to the causes, diagnosis, treatment, control, and prevention of physical and mental diseases.

➢ Make grants-in-aid to universities, hospitals, laboratories, and other public and private research institutions.

➢ Participate with other countries in cooperative endeavors in biomedical research, health care technology, and health services research for the purpose of advancing the status of health sciences in the United States.

➢ Cooperate with and assist states and their political subdivisions in the prevention and suppression of communicable diseases and other public health matters.

In regard to provisions of the Public Health Service Act which promote, encourage, and influence activities in the area of birth defects study and prevention, Section 317C was added to the Public Health Service Act by the Children's Health Act of 2000. Section 317C provides the general operating authority for the National Center on Birth Defects and Developmental Disabilities (NCBDDD), a center within the

CDC. This authority was recently renewed in accordance with the Birth Defects and Developmental Disabilities Prevention Act of 2003. In part, Section 317C allows NCBDDD to:

➤ Collect, analyze, and make available data on birth defects and developmental disabilities.

➤ Operate regional centers for the conduct of applied epidemiological research on the prevention of such defects and disabilities.

➤ Provide information and education to the public on the prevention of such defects and disabilities.

The Public Health Service Act is codified in Title 42 of the United States Code.

## 2.4.5  Freedom of Information Act 5 USC §522 (FOIA)

All federal agencies are generally required under the Freedom of Information Act (FOIA) to disclose records they maintain when requested in writing by any person. Most states have adopted state laws that mirror the federal law. Therefore, it is important for a state birth defects surveillance program to be aware of the state law and know which records they may have to provide to the public when requested. However, federal agencies may withhold information pursuant to nine exemptions and three exclusions contained in the statute, and states have generally adopted similar exemptions. The exemptions that are most pertinent here are the FOIA exemptions 3 and 6.

Exemption Number 3:

> *Specifically exempted from mandatory disclosure by statute (other than the Privacy Act), provided that such statute:*

> *(i) Requires that the matters be withheld from the public in such a manner as not to leave any discretion on the issue, or*

> *(ii) Establishes particular criteria for withholding or refers to particular criteria for withholding or refers to particular types of matters to be withheld.*

This exemption is useful for protecting birth records in surveillance programs when the authorizing legislation specifically exempts the information in the statute.

Exemption Number 6:

> *Personnel and medical files and similar files the disclosure of which would constitute a clearly unwarranted invasion of personal privacy.*

The FOIA applies only to federal agencies and does not create a right of access to records held by Congress, the courts, or by state or local government agencies. Each state has its own public access laws that should be consulted for access to state and local records. Each federal agency is responsible for meeting its FOIA responsibilities for its own records. Likewise, each federal agency component is responsible for processing FOIA requests for the records that it maintains. For more information and a list of FOIA federal contacts, see the Department of Justice website at http://www.doj.gov.

# 2.4.6 Advocacy

In this section we discuss advocacy for the development and implementation of surveillance systems in terms of both the state's role and CDC's role in such advocacy.

**The role of the state in advocacy.** State health officials and surveillance staff can be important partners for advocates in the development and implementation of surveillance systems. While state employees may be limited in terms of what activities they can participate in within advocacy, they can work together with advocates throughout the process in order to create or improve birth defects systems. State officials and health department surveillance staff bring planning, technical assistance, and an understanding of the political environment to the planning and implementation process.

**The role of the CDC in advocacy.** The CDC can also work with states and with advocates to provide technical assistance in the design, planning, and implementation stages of a birth defects surveillance system and can make recommendations for improving ongoing programs. CDC can also play a substantial role in educating policymakers and the public about the benefits of a birth defects surveillance program.

# 2.5  References

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*Appendix 2.1*

*Sample State Legislation*

# Appendix 2.1 – Sample State Legislation

***Appendix 2.1.1***

***Arkansas Legislation***

**Arkansas Code of 1987 Annotated**
**Title 20. Public Health and Welfare**
**Subtitle 2. Health and Safety**
**Chapter 16. Reproductive Health**
**Subchapter 2. Arkansas Reproductive Health Monitoring System**


**20-16-201 Establishment- Purpose**
a) The Arkansas Reproductive Health Monitoring System is established and is to be administered within the Arkansas Children's Hospital.
b) The purpose of the system is to collect and analyze data from a number of sources to describe trends in the occurrence of reproductive endpoints such as congenital anomalies, fetal death, developmental disorders, etc., and to correlate those trends and investigate and report on the suspected causes of unexpected deviations in those trends.
**History**. Acts 1985, No. 214, § 1; A.S.A. 1947, § 82-4608.


**20-16-202 Definitions**
As used in this subchapter, unless the context otherwise requires:
1) "Board" means the technical advisory board established in § 20-16-204;
2) "Commission" means the advisory commission established in § 20-16-203; and
3) "System" means the Arkansas Reproductive Health Monitoring System.
**History**. Acts 1985, No. 214, § 3; A.S.A. 1947, § 82-4610.


**20-16-203 Advisory Commission- Members- Functions**
a) The Arkansas Reproductive Health Monitoring System shall be administered with the advise of an advisory commission appointed to one-year renewable terms by the Medical Director of the Arkansas Children's Hospital.
b) The functions of the commission are to:
    1) Advise the medical director as to the adequacy of policies, procedures, and performance of the system;
    2) Appoint members of the board upon the recommendations of the medical director;
    3) Promote the purposes of the system and assist in identification of appropriate funding sources;
    4) Promote interagency cooperation toward the goals of this system;
    5) Advise the medical director regarding requests for data dissemination; and
    6) Review mechanisms ensuring the maintenance of the confidentiality of personal data.
c) This commission shall be composed of the following state agencies, professional members, and public members:
    1) The medical director of the Arkansas Children's Hospital;
    2) The chancellor of the University of Arkansas for Medical Science;
    3) The director of the Department of Health;
    4) The director of the Department of Human Services;
    5) The director of the Arkansas Department of Environmental Quality;
    6) The director of the National Center for Toxicological Research;
    7) One (1) representative of the Arkansas Medical Society;
    8) One (1) representative of the Arkansas Academy of Pediatrics;

9) One (1) representative of the Arkansas Society of Obstetrics & Gynecology;
10) One (1) representative of the Arkansas Hospital Association;
11) One (1) representative of the State Plant Board;
12) Two (2) consumer representatives;
13) One (1) member from the Senate Public Health, Welfare, and Labor Committee and one (1) member from the House Public Health, Welfare, and Labor Committee; and
14) Up to four (4) additional members at large may be appointed.

d) Members of the commission who are not employees of the state may receive expense reimbursement in accordance with § 25-16-901 et seq.

**History.** Acts 1985, No 214, §§ 4, 11; A.S.A. 1947, §§ 82-4611; Acts 1997, No. 250, § 188; 1999, No. 1164, § 173.

**Amendments.** The 1997 amendment rewrote (d). The 1999 amendment substituted "Environmental Quality" for "Pollution Control and Ecology" in (c) (5).

### 20-16-204 Technical Advisory Board- Members- Functions

a) There shall be a technical advisory board whose function shall be to:

b) (1) This board shall be appointed to one-year renewable terms by the Medical Director of the Arkansas Children's Hospital upon recommendation of the commission and the director.

(2) It shall be comprised of a maximum of ten (10) regular members drawn from fields of expertise such as: medicine, industrial hygiene and toxicology, agriculture, environmental sciences, and epidemiology and statistics.

(3) At the discretion of the board and the director, ad hoc members of the board may be appointed for specific periods to advise on special needs or problems, which have been identified.

c) Members of the board who are not employees of the state may receive expense reimbursement in accordance with § 25-16-901 et seq.

**History.** Acts 1985, No 214, §§ 5, 11; A.S.A. 1947, §§ 82-4612, 82-4618; Acts 1997, No 250, § 189.

**Amendments.** The 1997 amendment rewrote (c).

### 20-16-205 Director- Appointment- Power and duties

a) The Arkansas Reproductive Health Monitoring System shall be administered by a director appointed by the Medical Director of the Arkansas Children's Hospital from among the professional staff of the Arkansas Children's Hospital.

b) The director shall:
1) Supervise the work of the system and administer the budget;
2) Appoint and remove such other employees as may be necessary to perform the duties and responsibilities of the system; and
3) Select and retain the services of consultants whose advice is considered necessary to carry out the system's mandate.

**History.** Acts 1985, No 214, § 2; A.S.A. 1947, § 82-4609.

**20-16-206 Authority to contract for information**
a) The Arkansas Reproductive Health Monitoring System is expressly authorized to contract for the production of any information, which its technical advisory board determines to be relevant to monitoring reproductive health from any department or agency of the state.
b) Information shared under this section includes, but is not limited to, information identified by the name or other personal identifier, including information concerning any system by which such data or information is identified or classified if required to decipher the information.
**History.** Acts 1985, No 214, § 6; A.S.A. 1947, § 82-4613.

**20-16-207 Information confidential- Exception**
The Arkansas Reproductive Health Monitoring System is expressly exempted and prohibited from supplying any information by individual name or other personal identifier or in a form other than a statistical report or other appropriate form which protects the confidentiality of individuals except to any state agency or department which originally supplied the information to the system unless both the originating agency and the system grant release of this information for a specific purpose.
**History.** Acts 1985, No 214, § 7; A.S.A. 1947, § 82-4614.

**20-16-208 Furnishing of information by hospitals**
a) All hospitals with patient records containing information pertaining to reproduction and development are required to share information in those records with the Arkansas Reproductive Health Monitoring System.
b) No hospital shall be required to furnish information under this section until appropriate reimbursement in return for the service has been determined by the advisory commission and funds are available to pay the compensation.
**History.** Acts 1985, No 214, § 8; A.S.A. 1947, § 82-4615.

**20-16-209 Furnishing of information by physician, clinic, etc.**
a) Any physician, clinic, person, or organization may provide information relative to reproductive health to the Arkansas Reproductive Health Monitoring System.
b) No liability of any kind or character for damages or other relief shall arise or be enforced against any person or organization by reason of having provided the information or by reason of having released or published the findings of the system in order to reduce morbidity and mortality or to advance medical research or medical education.
**History.** Acts 1985, No 214, § 9; A.S.A. 1947, § 82-4616.

**20-16-210 Intergovernmental agreements**
The Arkansas Reproductive Health Monitoring System shall have the power to enter into agreements with neighboring states and the federal Centers for Disease Control and Prevention consistent with the requirements and restrictions of this subchapter in order to obtain relevant information for the system concerning Arkansas residents who receive health-related services outside the state.
**History.** Acts 1985, No 214, § 10; A.S.A. 1947, § 82-4617.

**20-16-211 Funding and implementation**
a) The Arkansas Reproductive Health Monitoring System shall have the power to receive and expend grants, donations, and funds from public and private sources to carry out its responsibilities under this subchapter.
b) The Arkansas Children's Hospital is not required to implement this system unless sufficient funds are available as determined by the Medical Director of the Arkansas Children's Hospital.
c) The system may be implemented in stages or phases.
**History.** Acts 1985, No 214, § 13; A.S.A. 1947, § 82-4620.

**20-16-212 Reports**
The Arkansas Reproductive Health Monitoring System shall periodically prepare reports of its findings for dissemination to appropriate agencies and interested persons.
**History.** Acts 1985, No 214 § 14; A.S.A. 1947, § 82-4621.

**20-16-213 Rendering of patient care and regulatory activity prohibited**
The Arkansas Reproductive Health Monitoring System is expressly prohibited from rendering patient care, promulgating any rule or regulation, or engaging in any regulatory activity.
**History.** Acts 1985, No 214, § 13; A.S.A. 1947, § 82-4620.

**20-16-214 No actionable right, presumptions, or findings created**
a) Persons other than the state or Arkansas Reproductive Health Monitoring System shall not acquire any actionable right by virtue of this subchapter
b) A determination by this system that a source is suspected of causing adverse reproductive health outcomes shall not create by reason thereof any presumption of law or finding of a fact which shall inure to, or be for, the benefit of any person other than the state.
**History.** Acts 1985, No 214, § 12; A.S.A. 1947, § 82-4619.

**Arkansas Code of 1987 Annotated**
**Title 20. Public Health and Welfare**
**Subtitle 2. Health and Safety**
**Chapter 16. Reproductive Health**
**Subchapter 4. Reproductive Health Information**

**20-16-402 Information from state agencies**
a) (1) Any bona fide appropriately licensed medical facility, including, but not limited to, county hospitals, participating in recognized research in Arkansas and the federal Centers for Disease Control and Prevention are expressly authorized to contract for the production of any information relevant to monitoring reproductive health from any department or agency of the state.
(2) Information acquired under this subsection (a) includes, but is not limited to, information identified by name or other personal identifying information including the methods by which the information was compiled or tabulated.
b) The University of Arkansas for Medical Sciences, Arkansas Children's Hospital, other participating medical facilities as described in subsection (a) of this section, and the federal Centers for Disease Control and Prevention are expressly prohibited from supplying any information obtained pursuant to subsection (a) of this section by individual name or other personal identifying information or in a form other than a statistical report or other appropriately form which protects the confidentiality of individuals.
c) Information obtained pursuant to subsection (a) of this section may be returned to any state agency or department from which it was originally obtained.
**History.** Acts 1983, No 773, §§ 1, 3; A.S.A. 1947, §§ 82-4601, 82-4603.

*Appendix 2.1.2*

*California Legislation*

SCIENTIFIC

FOUNDATIONS

SERIES

CALIFORNIA
BIRTH DEFECTS
MONITORING
PROGRAM

# STATUTORY

# AUTHORITY

OCTOBER 1998

# STATUTORY AUTHORITY

*Recognizing that birth defects are a public health problem about which too little is known, the State Legislature in 1982 created the California Birth Defects Monitoring Program. From 1982-1990, seven pieces of legislation were passed and enacted, mandating the Program to:*

■ *Maintain an ongoing birth defects monitoring program statewide*

■ *Track birth defects rates and trends*

■ *Evaluate whether environmental hazards are associated with birth defects*

■ *Investigate other possible birth defects causes*

■ *Develop birth defects prevention strategies*

■ *Conduct interview studies about causes*

■ *Operate by contract with a qualified entity.*

*This document includes the Program's current statutory authority in the Health & Safety Code.*

HEALTH & SAFETY CODE, DIVISION 102, PART 2, CHAPTER 1

## CHAPTER 1. BIRTH DEFECTS MONITORING PROGRAM

**Section**

103825.   Legislative findings and declaration.
103830.   Collection of information;  system establishment;  medical records.
103835.   Scope of program;  assessment of resources.
103840.   Investigative studies.
103845.   Advisory committee;  membership.
103850.   Confidentiality of information;  research; review and approval;  civil penalty.
103855.   Contract for establishment and implementation of program.

*Chapter 1 was added by Stats.1995, c. 415 (S.B. 1360), § 4.*

**Historical and Statutory Notes**   Legislative findings relating to the nonsubstantive effect of Stats.1995, c. 415 (S.B. 1360), and the legislative intent not to create any new rights, see Historical and Statutory Notes under Health and Safety Code § 100100.

### § 103825.  Legislative findings and declaration

The Legislature hereby finds and declares that birth defects, stillbirths, and miscarriages represent problems of public health importance about which too little is known; that these conditions lead to severe mental anguish on the part of parents and relatives and frequently to high medical care costs; and that a system to obtain more information about these conditions could result in development of preventive measures to decrease their incidence in the future.  Therefore, it is the intent of the Legislature in enacting this section to accomplish all of the following:

(a)  To maintain an ongoing program of birth defects monitoring statewide.  "Birth defect" as used in this chapter means any medical problem of organ structure, function, or chemistry of possible genetic or prenatal origin.

(b)  To provide information on the incidence, prevalence, and trends of birth defects, stillbirths, and miscarriages.

(c)  To provide information to determine whether environmental hazards are associated with birth defects, stillbirths, and miscarriages.

(d)  To provide information as to other possible causes of birth defects, stillbirths, and miscarriages.

(e)  To develop prevention strategies for reducing the incidence of birth defects, stillbirths, and miscarriages.

(f)  To conduct interview studies about the causes of birth defects.

(g)  To affirm the authority of the state department to contract with a qualified entity to operate the birth defects monitoring program statewide.

(Added by Stats.1995, c. 415 (S.B., 1360), § 4.)

**Historical and Statutory Notes**

**Derivation:** Former §10800, added by Stats.1982, c. 204, § 1, amended by Stats.1985, c. 1137, § 1; Stats.1989, c. 8, § 1; Stats.1990, c. 122, § 1.

### § 103830.  Collection of information; system establishment; medical records

The director shall maintain a system for the collection of information,  necessary to accomplish the purposes of this chapter.  The director shall require health facilities, with 15 days' notice, to make available to authorized program staff the medical records of children suspected or diagnosed as having birth defects, including the medical records of their mothers.  In addition,  health facilities shall make available the medical records of mothers suspected or diagnosed with stillbirths or miscarriages and other records of persons who may serve as controls for interview studies about the causes of birth defects.  If it is necessary to photocopy records made available under this section, copying expenses shall be paid by the state department.

"Health facilities" as used in this section means general acute care hospitals, and physician-owned or operated clinics, as defined in Section 1200, that regularly provide services for the diagnosis or treatment of birth defects, genetic counseling, or prenatal diagnostic services.

(Added by Stats 1995, c. 415 (S.B.1360),  § 4.)

**Historical and Statutory Notes**

**Derivation:** Former §10801, added by Stats.1982, c. 204, § 1, amended by Stats.1989, c. 8, § 2; Stats.1990, c. 122, § 3.

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### § 103835. Scope of program; assessment of resources

The birth defects monitoring program shall operate statewide. It is the intent of the Legislature that the adequacy of program resources shall be assessed annually, and that the annual assessment shall include a consideration of at least all the following factors:

(a) The numbers of births in the state.

(b) The scope of program activities.

(c) Any urgent situation requiring extraordinary commitment of present or planned program staff or resources.

(Added by Stats.1995, c. 415 (S.B. 1360), § 4.)

**Historical and Statutory Notes**

**Derivation:** Former § 10802, added by Stats.1982, c. 204, § 1, amended by Stats.1990, c. 122, § 4.

### § 103840. Investigative studies

The director shall use the information collected pursuant to Section 103830 and information available from other reporting systems and health providers to conduct studies to investigate the causes of birth defects, stillbirths, and miscarriages and to determine and evaluate measures designed to prevent their occurrence. The department's investigation of poor reproductive outcomes shall not be limited to geographic, temporal, or occupational associations, but may include investigation of past exposures.

(Added by Stats.1995, c. 415 (S.B.1360), § 4.)

**Historical and Statutory Notes**

**Derivation:** Former § 10803, added by Stats.1982, c. 204, § 1, amended by Stats.1990, c. 122, § 7.

### § 103845. Advisory committee; membership

The director shall appoint an advisory committee to advise on the implementation of this chapter. Each of the disciplines of epidemiology, hospital administration, biostatistics, maternal and child health and public health shall be represented on the committee. At least one of the members shall be a representative of the manufacturing industry.

(Added by Stats.1995, c. 415, (S.B.1360), § 4.)

**Historical and Statutory Notes**

**Derivation:** Former § 10804, added by Stats.1982, c. 204, § 1, amended by Stats.1990, c. 122, § 8.

### § 103850. Confidentiality of information; research; review and approval; civil penalty

(a) All information collected and analyzed pursuant to this chapter shall be confidential insofar as the identity of the individual patient is concerned and shall be used solely for the purposes provided in this chapter. Access to the information shall be limited to authorized program staff, and persons with a valid scientific interest, who meet qualifications as determined by the director, who are engaged in demographic, epidemiological or other similar studies related to health, and who agree, in writing, to maintain confidentiality.

(b) The department shall maintain an accurate record of all persons who are given access to the information in the system. The record shall include: the name of the person authorizing access; name, title, and organizational affiliation of persons given access; dates of access; and the specific purpose for which information is to be used. The record of access shall be open to public inspection during normal operating hours of the state department.

(c) All research proposed to be conducted by persons other than program staff, using the information in the system, shall first be reviewed and approved by the director and the State Committee for the Protection of Human Subjects. Satisfaction of the terms of the director's rules for data access shall be deemed to establish a valid scientific interest for purposes of subdivision (a), entitling the researcher to review records collected pursuant to Section 103830 and to contact case subjects and controls.

(d) Whenever program staff, pursuing program objectives, deems it necessary to contact case subjects and controls, program staff shall submit a protocol describing the research to the director and to the State Committee for the Protection of Human Subjects. Once

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a protocol is approved by that committee, program staff shall be deemed to have established a bona fide research purpose, and shall be entitled to complete the approved project and contact case subjects and controls without securing any additional approvals or waivers from any entity.

(e)  Nothing in this section shall prohibit the publishing by the department of statistical compilations relating to birth defects, stillbirth, or miscarriage that do not in any way identify individual cases or individual sources of information.

(f)  Any person who, in violation of a written agreement to maintain confidentiality, discloses any information provided pursuant to this section, or who uses information provided pursuant to this section in a manner other than as approved pursuant to this section may be denied further access to any confidential information maintained by the department.  That person shall also be subject to a civil penalty of five hundred dollars ($500).  The penalty provided in this section shall not be construed as restricting any remedy, provisional or otherwise, provided by law for the benefit of the department or any person.

(Added by Stats.1995, c. 415, (S.B.1360), § 4.)

**Historical and Statutory Notes**

**Derivation:** Former § 10805, added by Stats.1982, c. 204, § 1, amended by Stats.1989, c. 8, § 4; Stats.1990, c. 122, § 9.

**Library References**

Records key 31. WESTLAW Topic No. 326. C.J.S. Records § § 74 to 92.

### § 103855. Contract for establishment and implementation of program

The department may enter into a contract for the establishment and implementation of the birth defects monitoring program.  The contract shall include provisions requiring full compliance with all the requirements of this chapter. The term of the contract may be in excess of one year, but no longer than three years.  Funds shall be allocated in accordance with the state Budget Act.  Funds withheld from the contractor at the conclusion of a fiscal year until specified tasks are completed shall be released promptly on proof of

substantial completion, and shall not be offset against any funding for the subsequent fiscal year.

(Added by Stats.1995, c. 415, (S.B.1360), § 4.)

**Historical and Statutory Notes**

**Derivation:** Former § 10806, added by Stats.1985, c. 1147, § 2, amended by Stats.1989, c. 8, § 5; Stats.1990, c. 122, § 10.

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CALIFORNIA
BIRTH DEFECTS
MONITORING
PROGRAM

The California Birth Defects Monitoring Program—
a public health program devoted to finding causes of birth defects—
is funded through the California Department of Health Services
and jointly operated with the March of Dimes Birth Defects Foundation.

For more information about the Program, please call
**(559)224-2212.**

*Appendix 2.1.3*

*New Jersey Legislation*

## REGISTRATION - VITAL STATISTICS 26:8-40.22

**26:8-40.21.  Birth defects registry**

The State Department of Health shall establish and maintain a birth defects registry which shall contain a confidential record of all birth defects that occur in New Jersey and any other information that the department deems necessary and appropriate in order to conduct thorough and complete epidemiologic surveys of birth defects that occur in this State and plan for and provide services to children with birth defects and their families.

L1983, c.291, § 2.

### Historical Note

Effective date, see Historical Note under § 26:8-40.20.

### Library References

Health and Environmental 34.
C.J.S. Health and Environmental § 41.

**26:8-40.2.    Confidential reports of abortions of fetus with or infant affected by birth defects**

a.  The Commissioner of Health, in consultation with the Public Health Council, shall require the confidential reporting to the Department of Health of all cases where a pregnancy results in a naturally aborted fetus or infant affected by a birth defect, and an electively aborted fetus that exhibits or is known to have a birth defect after 15 weeks of gestation.  The reporting requirement shall apply to all infants from birth through one year of age.

b. The Commissioner of Health shall determine the health care providers and facilities which shall be required to report all birth defects, the types of conditions or defects that shall be reported, the type of information that shall be contained in the confidential reprot and the method for making the report.  In reports concerning all fetuses with anomalies, the name of the mother shall not be submitted.

### Historical Note

Efective date, see Historical Note under § 26:8-40.20.

### Library References

Health and Environmental 34.
C.J.S. Health and Environmental § 41.

**26:8-40.23  Confidentiality of reports**

The confidential reports made pursuant to this act are to be used only by the Department of Health and other agencies that may be designated by the Commissioner of Health and shall not otherwise be divulged or made public so as to disclose the identity of any person to whom they relate; and to that end, such reports shall not be included under materials available to public inspection pursuant to P.L. 1963, c.73 (C.47:1A-1 et seq.).

L.1983, c.291, § 4.

**Historical Note**

Effective date, see Historical Note under § 26:8-40.20.

**Library References**

Health and Enviornment 34.
Records 30 et seq., 50 et seq.
C.J.S. Health and Environment § 41.
C.J.S. Records §§ 34 to 38.

**26:8-40.24.  Nonliability for divulging confidential information**

No individual or organization providing information to the Department of Health in accordance with this act shall be deemed to be or held liable for divulging confidential information.

L.1983, c.291, § 5.

**Historical Note**

Effective date, see Historical Note under § 26:8-40.20.

**Library References**

Health and Environmental 34.
Records 30 et seq., 50 et seq.
C.J.S. Health and Environmental § 41.
C.J.S. §§ 34 to 38.

**26:8-40.25.  Act not to be construed to compel submission to medical examination or to supervision by department of health**

Nothing in this act shall be construed to compel any individual to submit to a medical examination or to Department of Health supervision.

L.1983, c.291, §§ 6.

## Historical Note

Effective date, see Historical Note under § 26:8-40.20.

## Library References

Health and Environmental 34.
C.J.S. Health and Environmental § 41.

### 26:8-40.26.  Rules and regulations

The Commissioner of Health shall promulgate rules and regulations necessary to effectuate the purposes of this act.

L.1983, c.291, § 7.

## Historical Note

Effective date, see Historical Note under § 26:8-40.20.

## Library References

Administrative Law and Procedure 381 et. Seq.
Health and Environment 7(3), 20, 39.
C.J.S. Health and Environmental §§ 2 to 13, 40 to 51, 62 to 64, 106, 125 to 137, 155, 156.
C.J.S. Public Administrative Law and Procedure §§ 87 to 91.

## CHAPTER 20

## BIRTH DEFECTS REGISTRY

**Authority**

N.J.S.A. 26:8–40 et seq., specifically 26:8–40.26.

**Source and Effective Date**

R.2000 d.99, effective February 10, 2000.
See: 31 N.J.R. 2863(a), 32 N.J.R. 802(a).

**Executive Order No. 66(1978) Expiration Date**

Chapter 20, Birth Defects Registry, expires on February 10, 2005.

**Chapter Historical Note**

Chapter 20, Birth Defects Registry, was adopted as R.1985 d.92, effective March 4, 1985. See: 16 N.J.R. 3118(a), 17 N.J.R. 591(a).

Pursuant to Executive Order No. 66(1978), Chapter 20, Birth Defects Registry, was readopted as R.1990 d.187, effective March 2, 1990. See: 21 N.J.R. 3636(a), 22 N.J.R. 1134(c).

Pursuant to Executive Order No. 66(1978), Chapter 20, Birth Defects Registry, was readopted as R.1995 d.182, effective March 2, 1995. See: 27 N.J.R. 269(a), 27 N.J.R. 1410(b).

Pursuant to Executive Order No. 66(1978), Chapter 20, Birth Defects Registry, was readopted as R.2000 d.99, effective February 10, 2000. See: Source and Effective Date. See, also, section annotations.

### CHAPTER TABLE OF CONTENTS

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## SUBCHAPTER 1.   LIVE BIRTHS

### 8:20–1.1   Definitions

The following words and terms when used in this document shall have the following meanings unless the context clearly indicates otherwise.

"Birth defect" means an abnormality of the body's structure or inherent function which is present at birth, whether such abnormality is manifest at the time of delivery or becomes apparent later in life.

"Infant" means a child from birth to one year of age.

### 8:20–1.2   Reporting requirements

(a) Any infant who is born to a resident of the State of New Jersey, or who becomes a resident of the State before one year of age, and who is diagnosed as having a birth defect either at birth or any time during the first year of life shall be reported to the State Department of Health and Senior Services, Special Child, Adult and Early Intervention Services Program as follows:

1. The conditions listed as Congenital Anomalies (Diagnostic Codes 740.00 through 759.90) in the most recent revision of the International Classification of Diseases, Clinical Modification, shall, except as specified in (a)1ii below, be reported to Special Child, Adult and Early Intervention Services. In addition, there are several other conditions considered to be defects that are not listed under Diagnostic Codes 740.00 through 759.90 which describe Congenital Anomalies. The birth defects listed in (a)1i below shall also, in every case, be reported to Special Child, Adult and Early Intervention Services. The minor conditions listed in (a)1ii below shall not be reported to Special Child, Adult and Early Intervention Services in every case, but only as required in (a)1iii, iv and v below.

i. Congenital anomalies, including, but not limited to, the following:

(1) Anencephalus and similar anomalies, such as craniorachischis and inencephaly.

(2) Spina bifida with and without mention of hydrocephalus.

(3) Other congenital anomalies of the nervous system, such as: encephalocele; microcephalus; reduction deformities of the brain; congenital hydrocephalus; congenital cerebral palsies, congenital muscular dystrophies; and other anomalies, congenital diseases, lesions and any other deformities of the brain, nervous system or spinal cord.

(4) Congenital anomalies of the eye, such as: anophthalmos; microphthalmos; buphthalmos; congenital cataract and lens anomalies; coloboma and other anomalies of the anterior or posterior segment; congenital anomalies of eyelids, lacrimal system and orbit; and any other anomalies of the eye.

(5) Congenital anomalies of the ear, face and neck, such as: anomalies of the ear causing impairment of hearing; accessory auricle and any other anomalies of the ear; branchial cleft cyst or fistula; preauricular sinus; webbing of the neck; and any other anomalies of face and neck.

(6) Bulbus cordis anomalies and anomalies of cardiac septal closure such as: common truncus; transposition of great vessels; Tetralogy of Fallot; common ventricle; ventricular septal defect; ostium secundum type atrial septal defect; endocardial cushion defects; cor biloculare; and any other defects of septal closure.

(7) Other congenital anomalies of the heart, such as: anomalies of pulmonary valve; congenital tricus-

pid atresia and stenosis; Ebstein's anomaly; congenital stenosis of aortic valve; congenital mitral stenosis of aortic valve; congenital mitral stenosis or insufficiency; hypoplastic left heart syndrome; and any other structural anomalies of the heart.

(8) Other congenital anomalies of circulatory system, such as: patent ductus arteriosus (only in infants larger than 2,500 grams); coarctation of aorta and other anomalies of the aorta, aortic arch or atresia and stenosis of the aorta; anomalies of pulmonary artery; anomalies of great veins, absence or hypoplasia of umbilical artery; other anomalies of peripheral vascular system; or other unspecified anomalies of circulatory system.

(9) Congenital anomalies of respiratory system, such as: choanal atresia; other anomalies of nose; webbing of larynx; other anomalies of larynx, trachea and bronchus; congenital cystic lung; agenesis, hypoplasia and dysplasia of lung; other anomalies of the lung; and other unspecified anomalies of respiratory system.

(10) Cleft palate and cleft lip.

(11) Other congenital anomalies of upper alimentary tract, such as: anomalies of the tongue; anomalies of mouth and pharynx; tracheoesophageal fistula, esophageal atresia, and stenosis and other anomalies of esophagus; congenital hypertrophic pyloric stenosis, congenital hiatal hernia; other anomalies of stomach; and other unspecified anomalies of upper alimentary tract.

(12) Other congenital anomalies of digestive system, such as: Meckel's diverticulum; atresia and stenosis of small intestine, large intestine, rectum and anal canal; Hirschsprung's disease and other congenital functional disorders of colon; anomalies of intestinal fixation; other anomalies of intestine, gall bladder, bile ducts, liver and pancreas; disorders of tooth formation, development and eruption, dentofacial anomalies, and other unspecified anomalies of the digestive system.

(13) Congenital anomalies of genital organs, such as: anomalies of ovaries, fallopian tubes and broad ligaments; doubling of uterus and other anomalies of uterus; anomalies of cervix, vagina and external female genitalia; undescended testicle; hypospadias and congenital chordee; indeterminate sex and pseudohermaphroditism; and other unspecified anomalies of the genital system.

(14) Congenital anomalies of urinary system, such as: renal agenesis and dysgenesis; cystic kidney disease; obstructive defects of renal pelvis and ureter; other anomalies of kidney and ureter; exstrophy of urinary bladder; atresia and stenosis of urethra and bladder neck; anomalies of urachus; other anomalies of bladder and urethra; and other unspecified anomalies of the urinary system.

(15) Certain congenital musculoskeletal deformities, such as: of skull, face and jaw; of sternocleidomastoid muscle; of spine; congenital dislocation of hip; congenital genu recurvatum and bowing of long bones of leg; varus and valgus deformities of feet; other congenital deformities of feet such as talipes cavus, calcaneus or equinus; and other specified nonteratogenic anomalies such as pectus excavatum, pectus carinatum; club hand; congenital deformity of chest wall; dislocation of elbow; generalized flexion contractures of lower limbs; spade-like hand.

(16) Other congenital anomalies of limbs, such as: syndactyly; reduction deformities of upper limb; reduction deformities of lower limb; other anomalies of upper limb, including shoulder girdle; and other anomalies of lower limb, including pelvic girdle.

(17) Other congenital musculoskeletal anomalies, such as: anomalies of skull and facial bones; anomalies of spine; cervical rib; other anomalies of ribs and sternum; chondrodystrophy; osteodystrophies; anomalies of diaphragm; anomalies of abdominal wall such as prune belly syndrome; other specified anomalies of muscle, tendon, fascia and connective tissue; and other unspecified anomalies of musculoskeletal system.

(18) Congenital anomalies of the integument, significant anomalies of skin, subcutaneous tissue, hair, nails and breast, such as birthmarks or nevi measuring four inches or greater in size, multiple skin tags (more than five in number).

(19) Chromosomal anomalies, such as: Down's syndrome; Patau's syndrome; Edwards' syndrome; autosomal deletion syndromes and other conditions due to autosomal anomalies; gonadal dysgenesis; Klinefelter's syndrome; and other conditions due to sex chromosome anomalies or anomalies of unspecified chromosome.

(20) Other and unspecified congenital anomalies, such as: anomalies of spleen, situs inversus; conjoined twins; tuberous sclerosis; other hamartoes; multiple congenital anomalies; and other congenital anomalies including congenital malformation syndromes affecting multiple organ systems including Laurence–Moon–Biedl syndrome, Marfan's syndrome and Prader–Willi syndrome.

(21) Certain endocrine, nutritional and metabolic diseases and immunity disorders, includes congenital hypothyroidism; congenital hypoparathyroidism; hypopituitarism; diencephalic syndrome; adrenogenital syndrome; testicular feminization syndrome; phenylketonuria; albinism; maple syrup urine disease; argininosuccinic aciduria; glycogen storage diseases; cystic fibrosis; alpha–1 antitrypsin deficiency; DiGeorge's syndrome; congenital deficiencies of humoral immunity; cell-mediated immunity; combined immunity deficiencies; and other specified and unspecified disorders of the immune mechanisms.

(22) Certain diseases of the blood and blood forming organs, includes hemolytic diseases of the newborn; G-6PD deficiency; hemophilia (all types); Von Willebrand's disease; and sickle-cell anemia or other hemoglobin-apathies.

(23) Certain diseases of the nervous system and sense organs, includes hereditary and degenerative diseases of the central nervous system such as Tay Sachs disease and familial degenerative CNS diseases; Werdnig–Hoffmann disease; cerebral palsy; Moebius syndrome; hereditary retinal dystrophies, and chorioretinitis.

(24) Certain diseases of the circulatory system, includes endocardial fibroelastosis; congenital Wolfe–Parkinson–White syndrome; and Budd–Chiari syndrome.

(25) Certain diseases of the digestive system, includes abnormalities of jaw size, micrognathiae and macrognathia; congenital inguinal hernia with gangrene (only in females), congenital, inguinal hernia with obstruction with no mention of gangrene (only in females), congenital, inguinal hernia without obstruction with no mention of gangrene (only in females), umbilical hernia (only if not covered by skin), epigastric hernia.

(26) Certain complications of pregnancy childbirth, and the puerperium, includes amniotic bands, amniotic cyst.

(27) Certain diseases of the skin and subcutaneous tissue, pilonidal sinus.

(28) Certain conditions originating in the perinatal period, includes fetal alcohol syndrome, probable fetal alcohol syndrome (includes facies), fetal alcohol effects, fetal hydantoin (Dilantin) syndrome, bronchopulmonary dysplasia, unspecified TORCH infection and certain congenital infections including congenital syphilis, congenital rubella, cytomegalovirus, toxoplasmosis, hepatitis, herpes simplex.

(29) Neoplasms, includes lipomas of skin and subcutaneous tissue of face and other skin and subcutaneous tissue, intrathoracic and intra-abdominal organs, spermatic cord, other specified sites, lumbar, sacral, paraspinal, and other unspecified sites; benign neoplasms of skin includes blue nevus, pigmented nevus (include if greater than four inches in diameter), papilloma, dermatofibroma, syringoadenoma, dermoid cyst, hydrocystoma, syringoma; other benign neoplasms of lip, eyelid, ear, external auditory canal, skin and other unspecified parts of face, scalp, skin of neck, skin of trunk, skin of upper limb, skin of lower limb, other specified and unspecified sites including hairy nevus; hemangioma (include if: greater than four inches in diameter, multiple, more than five in number or cavernous hemangioma) of skin and subcutaneous tissue, intracranial, intra-abdominal cystic

hygroma; lymphangioma of any site, hemangioma of other and unspecified site; and certain malignant neoplasms including Wilm's tumor, retinoblastoma, other congenital neoplasms including neuroblastoma, medulloblastoma, teratoma, fibrosarcoma, histiocytosis (malignant), neurofibromatosis.

ii.    Minor conditions, as follows:

Accessory auricle
Accessory nipple (supernumerary nipple, or skin tag)
Anal fissure—never a defect
Anal tags
Bat ear
Bell's Palsy
Bent nose, deviation of septum
Big lips
Blue sclera (babies <2500 grams)
Brachial palsy
Breast Hypertrophy—never a defect
Cafe-au-lait spots (register if five or more)
Caput succedaneum
Cardiac murmur [1]
Cauliflower ear
CNS hemorrhage
Cephalhematoma—never a defect
Cervical rib
Chalasia (gastroesophageal reflux)—never a defect
Clinodactyly (incurving of fifth finger)
Congenital hydrocele
Conjunctivitis—never a defect
Cryptorchidism (undescended testicle) [2]
Darwin's tubercle
Diastasis recti—never a defect
Downward eyeslant (antimongoloid)
Ear tags, preauricular tags
Elfin ear
Epicanthal folds
Epulis—never a defect
Erb's palsy
Erythema toxicum
Esotropia
Exotropia
Facial palsy
Flammeus nevus or port wine stain (<four inches in diameter)
Flat bridge or nose
Fontanel (large or small)
Fractured clavicle
Fused eyelids (not a defect if birth weight is <1001 grams)
Gastroesophageal reflux—never a defect
Gum cysts—includes epulis, ranula, mucocele—never a defect
Hemangioma—<four inches in diameter [3]
Hepatomegaly
Hipclick—without follow-up or therapy—not a defect
Hydrocele
Hydrocephaly; acquired
Hymenal tags
Hypoglycemia, idiopathic
Hypoplastic scrotum
Imperforate hymen
Incurving finger (clinodactyly)
Inguinal hernia in male (Note: do not report in females)

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Infant of a diabetic mother; asymptomatic
Intussusception
Lanugo, excessive or persistent
Large fontanel
Laryngomalacia or tracheomalacia—never a defect
Long fingers and/or toes
Lop ear
Low set ears
Macrocheilia (big lips)
Meckel's diverticulum
Meconium peritonitis
Meconium plug
Meconium stained skin or nails—never a defect
Metatarus adductus—never a defect
Metatarus varus
Microcheilia (small lips)
Mongolian spots
Mucocele—never a defect
Nasal lacrimal duct obstruction
Nail defects
Natal teeth
Neonatal acne—never a defect
Nystagmus
Orthopedic positional anomalies [4]
Overlapping toes
Overriding (overlapping) sutures—never a defect
Partial syndactyly second and third toes—web extends
    <one-third length of second toe
Patent ductus arteriosus (PDA) in infants <2500
    grams or resolved prior to or at discharge
Patulous lips (wide lips)
Persistent fetal circulation
Petechiae—never a defect
Phimosis—never a defect
Pilonidal dimple
Pilonidal cyst
Pixie-like ear
Pneumothorax
Pointed ear
Polydactyly (postaxial, type B)—skin tags on hands or
    feet
Posteriorly rotated ears
Preauricular sinus
Pylorospasm (intermittent)
Ranula—never a defect
Rectal fissure
Redundant foreskin
Rockerbottom feet
Sacral dimple
Sebaceous cysts
Simian crease (transverse palmar crease)
Single umbilical artery
Skin cysts
Small fontanel
Small lips
Splenomegaly
Thymic hypertrophy
Tibial torsion
Tongue-tie
Torsion of spermatic cord
Torsion of testes
Tracheomalacia—never a defect
Umbilical cord atrophy
Umbilical hernias (completely covered by skin)
Undescended testicle [2]
Upturned nose
Upward eyeslant (mongoloid)

Vaginal cysts
Vaginal tags
Webbing of neck
Wide nasal bridge
Widely spaced nipples
Widely spaced first and second toes

iii.  If a condition or defect listed in (a)1ii above
appears as a single defect, a registration form shall not
be completed.

iv.  If two or more of the conditions listed in (a)1ii
above appear, a registration form shall be completed.

v.  If a condition or defect listed in (a)1ii above
accompanies a condition or defect listed in either Diag-
nostic Codes 740.00 through 759.90 in the most recent
revision of the International Classification of Diseases,
Clinical Modification, or in (a)1i above, a registration
form shall be completed.

(b) Any live born infant with a birth defect who has not
been previously registered and has expired shall be reported.
Such reports shall indicate that the infant has expired.

(c) The administrative officer of every health care facility
shall be responsible for establishing the reporting proce-
dures for that facility.  The reporting procedures must
insure that every infant who is initially diagnosed as having a
birth defect shall be reported to the Department.  All
presumptive, tentative, pending, or rule out diagnoses will
be reported at the time of discharge, if the child will be
diagnosed at a later time or if test results are pending.

(d) Every physician, dentist, certified nurse midwife, ad-
vanced practice nurse, and other health care professionals
who diagnose or confirm birth defects shall report to the
Department each infant diagnosed as having a birth defect
not known to be previously reported.

(e) The director of every clinical laboratory shall report
to the Department results of postmortem examination from
any infant indicating the existence of a birth defect, not
known to be previously reported.

(f) The information to be reported shall be provided
upon forms supplied by the State Department of Health and
Senior Services:

    Special Child, Adult and Early Intervention Ser-
        vices
    PO Box 364
    Trenton, New Jersey 08625–0364

(g) The reports made pursuant to these rules are to be
used only by the Department of Health and Senior Services
and other agencies that may be designated by the Commis-
sioner of Health and Senior Services and shall not otherwise
be divulged or made public so as to disclose the identity of
any person; and such reports shall be included under materi-
als available to public inspection pursuant to P.L. 1963, c.73
(N.J.S.A. 47:1A–1 et seq.).

## BIRTH DEFECTS REGISTRY                                    8:20–1.2

(h) Cytogenetic laboratories shall report the results of all postnatal chromosomal abnormalities.

(i) When a live infant is registered, the Department shall inform the parent or legal guardian of the registration.

(j) Every health care facility and independent clinical laboratory shall allow access to, or provide necessary information on infants with birth defects and other patients specified by characteristics for research studies related to birth defects conducted by the State Department of Health and Senior Services and which have been approved by the State Commissioner of Health and Senior Services after appropriate review for assuring protection of human subjects by the Department's Institutional Review Board. This shall include patients who came under the care of the health facility prior to March 4, 1985.

(k) Any agency designated by the Commissioner to receive reports pursuant to this chapter shall provide to Special Child, Adult and Early Intervention Services any updated diagnostic and/or demographic information.

Amended by R.1987 d.361, effective September 8, 1987.
See: 19 N.J.R. 909(b), 19 N.J.R. 1642(b).

Subsection (a) added a list of congenital anomalies and other conditions which also constituted reportable birth defects.
Amended by R.1990 d.187, effective April 2, 1990.
See: 21 N.J.R. 3636(a), 22 N.J.R. 1134(c).
Reporting requirements for certain conditions specified further; reporting requirements for sickle-cell anemia and other hemoglobinopathies added; all presumptive, tentative, pending and rule out diagnoses to be reported at discharge; cytogenetic laboratories to report postnatal chromosomal abnormality test results to the Department.
Amended by R.1991 d.414, effective August 5, 1991.
See: 23 N.J.R. 820(a), 23 N.J.R. 2335(a).
In (a)1, added ii. through v.
Amended by R.1992 d.184, effective April 20, 1992.
See: 24 N.J.R. 171(a), 24 N.J.R. 1494(b).
Minor conditions added at (a)1ii.
Amended by R.2000 d.99, effective March 6, 2000.
See: 31 N.J.R. 2863(a), 32 N.J.R. 802(a).
In (a) and (f), substituted references to Special Child, Adult and Early Intervention Services for references to Special Child Health Services; rewrote (d); in (j), inserted a reference to the Department's Institutional Review Board at the end of the first sentence, and substituted a reference to March 4, 1985 for a reference to the effective date of the regulations at the end of the last sentence; and added (k).

[1] Do not register innocent or functional murmurs: register only if there is a definitive cardiac anomaly or register as a rule out condition if the cause of murmur is not identified at the time of discharge.
[2] Register only if there is clinical evidence of congenital absence.
[3] Register cavernous hemangiomas and multiples of five or more.
[4] Do not register if defect can be corrected passively and does not require casting or bracing.

*Appendix 2.1.4*

*New York Legislation*

# Pertinent Public Health Laws and Regulations

The following laws and regulation establish the legal authority to collect information on birth defects and genetic diseases, to perform studies, and to maintain the confidentiality of the information and limits its use to research and the improvement of quality of care.

## Section 206 (1) of the Public Health Law

1. The Commissioner shall:

   (d) investigate the causes of disease, epidemics, the sources of mortality, and the effects of localities, employments and other conditions, upon the public health;

   (e) obtain, collect and preserve such information relating to marriage, birth,  mortality, disease and health as may be useful in the discharge of his duties or may contribute to the promotion of health or the security of life in the state,

   (j) cause to be made such scientific studies and research which have for their purpose the reduction of morbidity and mortality and the improvement of the quality of medical care through the conduction of medical audits within the state.  In conducting such studies and research, the commissioner is authorized to receive reports on forms prepared by him and the furnishing of such information to the commissioner, or his authorized representatives, shall not subject any person, hospital, sanitarium, rest home, nursing home, or other person or agency furnishing such information to any action for damages or other relief.  Such information when received by the commissioner, or his authorized representatives, shall be kept confidential and shall be used solely for the purposes of medical or scientific research or the improvement of the quality of medical care through the conduction of medical audits.  Such information shall not be admissible as evidence in any action of any kind in any court or before any other tribunal, board, agency or person.

## Section 225(5)(t) of the Public Health Law.

5.   The Sanitary code may:

   (t) facilitate epidemiological research into the prevention of environmental diseases, when such research is conducted pursuant

to paragraph (j) of subdivision one of section two hundred six of this chapter, by establishing regulations designating as environmentally related diseases those pathological conditions of the body or mind resulting from contact with toxins, or teratogens in solid, liquid or gaseous form, or in the form of ionizing radiation or nonionizing electromagnetic radiation, and by requiring the reporting of such diseases or suspected cases in such diseases to the department by physicians, medical facilities and clinical laboratories.  Any information provided to the department pursuant to such regulations shall be in the form required by the department and shall be kept confidential and used by the commissioner pursuant to the provisions of paragraph (j) of subdivision one of section two hundred six of this chapter, and other applicable laws relating to the confidential treatment of patient and medical data.

### Section 2733 of the Public Health Law

1. Birth defects and genetic and allied diseases shall be reported by physicians, hospitals, and persons in attendance at birth in the manner on and such forms as may be prescribed by the commissioner.

2. Such reports and information shall be kept confidential and shall not be admissible as evidence in an action or proceeding in any court or before any other tribunal, board, agency or person. The commissioner may, however, publish analyses of such information from time to time for scientific and public health purposes, in such manner as to assure that the identities of the individuals concerned cannot be ascertained.

### State Sanitary Code: Part 22 – Environmental Diseases
### (Statutory authority: Public Health Law, §§ 225 [5][t], 206 [1][j])

22.1    Supplementary reports of spontaneous abortions and fetal deaths for epidemiologic surveillance; filing.  Every physician and hospital shall file a supplemental report with the State Commissioner of Health of each spontaneous abortion or other fetal death occurring naturally.  Such report shall be filed within 10 days of the occurrence of such event on such fo9rms as may be prescribed by the commissioner to facilitate epidemiologic investigation and surveillance.

22.2    Supplementary reports of low birth weights for epidemiologic surveillance; filing.  Every physician, hospital, and person in attendance at live births shall file a supplementary report with the State commission of Health of each live birth for which the birth weight is 2,500 grams(5.2 pounds) less.

Such report shall be filed within 10 days of the birth and shall be on such formed as may be prescribed by the commissioner to facilitate epidemiologic investigation and surveillance.

**Regulation specifically establishing the CMR**

22.3    Supplementary reports of certain congenital anomalies for epidemiological surveillance; filing. Every physician and hospital in attendance on an individual diagnosed within two years of birth as having one or more of the congenital anomalies listed in this section shall file a supplementary report with the State Commissioner of Health within 10 days of diagnosis thereof. Such report shall be on such forms as may be prescribed by the commissioner to facilitate epidemiological investigation and surveillance.

22.9    Reports: place of filing. All reports required by this part shall be filed with the Director of the Bureau of Environmental Epidemiology and Occupational Health, Division of Epidemiology, New York State Department of Health, Empire State Plaza, Tower Building, Albany, NY 12237.

*Appendix 2.1.5*

*Oklahoma Legislation*

## PUBLIC HEALTH AND SAFETY                    63 § 1-550.2

### Historical and Statutory Notes

The repealed section, derived from Laws 1985, c 60, § 1; Laws 1986, c. 49, § 1, related to the reporting of cases of birth defects.

See, now, §§ 1-550.1, 1-550.2 of this title.

### § 1-550.1  Definitions

As used in this act:[1]

1. "Birth defect" means any physical or chemical abnormality present at birth;
2. "Commissioner" means the Commissioner of Health;
3. "Department" means the Oklahoma State Department of Health;
4. "ICD-9-CM diagnostic code categories" means the International Classification of Disease which assigns numbers to each of the congenital anomalies; and
5. "Poor reproductive outcomes" includes but is not limited to stillbirths and miscarriages.

Added by Laws 1987, c. 199, § 1, eff. Nov. 1, 1987.

[1] Section 1-550.1 et seq. of this title.

### Historical and Statutory Notes

Section 4 of Laws 1987, c. 199 provides for an effective date.

### § 1-550.2  Birth defects surveillance program

A. It is hereby found that the occurrence of a birth defect is a tragedy for the child, the family and the community, and a matter of vital concern to the public health. A system to obtain more information about these conditions could result in their prevention, treatment and management. Therefore, it is the intent of the Oklahoma State Legislature, in enacting this section, to:

1. Obtain information on the incidence and trends of birth defects and poor reproductive outcomes;
2. Obtain information to determine whether environmental hazards are associated with birth defects and poor reproductive outcomes;
3. Obtain information as to other possible causes of birth defects and poor reproductive outcomes; and
4. Develop prevention strategies for reducing the incidence of birth defects, and poor reproductive outcomes.

B. The Commissioner of Health may establish a system for the collection and verification of information concerning birth defects and other poor reproductive outcomes. In establishing the system, the Commissioner may require general acute care hospitals to maintain a list of patients up to six (6) years of age who have been diagnosed with birth defects incorporated within the ICD-9-CM diagnostic code categories 740 through 759.9 or such other information as the Commissioner deems appropriate, and all women discharged with a diagnosis of stillbirth or miscarriage. The list shall be made available to the Commissioner upon request and shall be used solely for purposes provided in this section.

C. The Commissioner may require general acute care hospitals, and other sources as deemed necessary, to make available to the State Department of Health the medical records of those patients who have been diagnosed with birth defects or poor reproductive outcomes as required in this section.

D. The system shall be implemented statewide.

E. The Commissioner may use the information collected pursuant to subsection B of this section and information available from other reporting systems and health providers to conduct studies to:

1. Investigate the causes of birth defects and poor reproductive outcomes;

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63 § 1-550.2                                                PUBLIC HEALTH AND SAFETY

2. Determine and evaluate measures to prevent their occurrences; and

3. Where possible ensure delivery of services for children identified with birth defects. The Department's investigation of poor reproductive outcomes shall include geographic, time-related or occupational associations, as well as investigations of past exposure to potentially harmful substances.

F. Where Commissioner may appoint an advisory committee of health professionals who shall advise on the implementation of this section. Advisory committee members shall serve without compensation.

G. If the Commissioner finds it is necessary to collect information from sources other than general acute care hospitals, the Commissioner shall first submit for approval to the advisory committee a proposal stating the need for such information.

H. All information collected and analyzed pursuant to this section shall be confidential insofar as the identity of the individual patient is concerned and shall be used solely for the purpose provided in this section. Access to such information shall be limited to the State Department of Health, provided that the Commissioner may provide access to those scientists approved by the advisory committee who are engaged in demographic, epidemiological or other similar studies related to health, and who agree, in writing as nonstate employees, to be identified and coded while maintaining confidentiality as described herein.

I. The Department shall maintain an accurate record of all persons who are given access to the information in the system. The record shall include:

1. The name of the persons authorizing access;
2. The name, title and organizational affiliation of persons given access;
3. The dates of access;
4. The specific purpose for which the information is to be used; and
5. The results of the independent research.

J. Nothing in this section shall prohibit the publishing of statistical complications relating to birth defects or poor reproductive outcomes which do not in any way identify individual cases or individual sources of information.

K. Any person who, in violation of a written agreement to maintain confidentiality, willfully discloses any information provided pursuant to this section shall be denied further access to any confidential information maintained by the Department. That person shall also be deemed guilty of a misdemeanor, and upon conviction thereof shall be punished by a fine of Two Hundred Dollars ($200.00) or imprisonment in the county jail for not more than thirty (30) days, of by both such fine and imprisonment.

L. The State Board of Health is authorized to adopt, amend and repeal rules and regulations for the purpose of carrying out the provisions of this section.

Added by Laws 1987, c. 199, § 2, eff. Nov. 1, 1987. Amended by Laws 1992, c. 123, § 1, emerg. eff. April 23, 1992.

**§ 1-551. Repealed by Laws 1987, c. 197, § 2, eff. Nov. 1, 1987**

### Historical and Statutory Notes

The repealed section, derived from Laws 1985, c. 60, § 2: Laws 1986, c. 49, § 2, related to the reporting cases of cancer.

See, now, § 1-551.1 of this title.

### § 1-551.1 Tumor registry

A. The State Commissioner of Health shall establish and maintain an up-to-date tumor registry to ensure an accurate and continuing source of data concerning such cancerous, precancerous and tumorous diseases as the State Board of Health may by rule specify. Such registry may include data necessary for epidemiological surveys and scientific research, and other data which is necessary and proper to further the recognition, prevention, control, treatment and cure of cancer, precancerous and tumorous diseases.

*Appendix 2.1.6*

*Texas Legislation*

**Texas legislation**

HEALTH & SAFETY CODE

CHAPTER 87. BIRTH DEFECTS

SUBCHAPTER A. GENERAL PROVISIONS

Sec. 87.001. DEFINITIONS. In this chapter:

(1) "Birth defect" means a physical or mental functional deficit or impairment in a human embryo, fetus, or newborn resulting from one or more genetic or environmental causes.

(2) "Communicable disease" has the meaning assigned by Section 81.003.

(3) Repealed by Acts 1995, 74th Leg., ch. 76, Sec. 8.134, eff. Sept. 1, 1995.

(4) "Environmental causes" means the sum total of all the conditions and elements that make up the surroundings and influence the development of an individual.

(5) "Harmful physical agent" has the meaning assigned by Section 503.001.

(6) "Health professional" means an individual whose:

(A) vocation or profession is directly or indirectly related to the maintenance of health in another individual; and

(B) duties require a specified amount of formal education and may require a special examination, certificate, or license or membership in a regional or national association.

(7) "Health facility" includes:

(A) a general or special hospital licensed by the department under Chapter 241;

(B) a physician-owned or physician-operated clinic;

(C) a publicly or privately funded medical school;

(D) a state hospital or state school maintained and managed by the Texas Department of Mental Health and Mental Retardation;

(E) a genetic evaluation and counseling center;

(F) a public health clinic conducted by a local health unit, health department, or public health district organized and recognized under Chapter 121;

(G)   a physician peer review organization;  and

(H)   another facility specified by board rule.

(8)   "Midwife" has the meaning assigned by Section 203.002, Occupations Code.

(9)   "Local health unit" has the meaning assigned by Section 121.004.

(10)   "Toxic substance" has the meaning assigned by Section 503.001.

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993. Amended by Acts 1995, 74th Leg., ch. 76, Sec. 8.134, eff. Sept. 1, 1995;  Acts 2001, 77th Leg., ch. 1420, Sec. 14.774, eff. Sept. 1, 2001.

Sec. 87.002.  CONFIDENTIALITY.  (a)  Except as specifically authorized by this chapter, reports, records, and information furnished to a department employee or to an authorized agent of the department that relate to cases or suspected cases of a health condition are confidential and may be used only for the purposes of this chapter.

(b)  Reports, records, and information relating to cases or suspected cases of health conditions are not public information under Chapter 552, Government Code, and may not be released or made public on subpoena or otherwise except as provided by this chapter.

(c)  The department may release medical, epidemiological, or toxicological information:

(1)  for statistical purposes, if released in a manner that prevents the identification of any person;

(2)  with the consent of each person identified in the information or, if the person is a minor, the minor's parents, managing conservator, guardian, or other person who is legally authorized to consent;

(3)  to medical personnel, appropriate state agencies, health authorities, regional directors, and public officers of counties and municipalities as necessary to comply with this chapter and board rules relating to the identification, monitoring, and referral of children with birth defects;

(4)  to appropriate federal agencies, such as the Centers for Disease Control of the United States Public Health

Service; or

   (5) to medical personnel to the extent necessary to protect the health or life of the child identified in the information.

  (d) A board member, the commissioner, another employee of the department, or an authorized agent may not be examined in a civil, criminal, special, or other proceeding as to the existence or contents of pertinent records of or reports or information about a child identified or monitored for a birth defect by the department without the consent of the child's parents, managing conservator, guardian, or other person authorized by law of this state or another state or by a court order to give consent.

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993. Amended by Acts 1995, 74th Leg., ch. 76, Sec. 5.95(88), 8.130, eff. Sept. 1, 1995.

  Sec. 87.003. CONTRACTS. The department may enter into contracts or agreements with persons as necessary to implement this chapter. The contracts or agreements may provide for payment by the state for supplies, equipment, data, and data collection and other services.

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993.

  Sec. 87.004. LIMITATION OF LIABILITY. A health professional, a health facility, or an administrator, officer, or employee of a health facility subject to this chapter is not civilly or criminally liable for divulging information required to be released under this chapter, except in a case of gross negligence or wilful misconduct.

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993.

  Sec. 87.005. COOPERATION OF GOVERNMENTAL ENTITIES. Another state board, commission, agency, or governmental entity capable of assisting the department in carrying out the intent of this chapter shall cooperate with the department and furnish expertise, services, and facilities to the program.

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993.

   SUBCHAPTER B. BIRTH DEFECTS MONITORING PROGRAM

  Sec. 87.021. SURVEILLANCE PROGRAM; REGISTRY ESTABLISHED. (a) The board shall establish in the department a

program to:

        (1)   identify and investigate certain birth defects in children;  and

        (2)   maintain a central registry of cases of birth defects.

    (b)   The board may authorize the department to implement a statewide program or to limit the program to a part or all of one or more public health regions, depending on the funding available to the department.  In establishing the program, the board shall consider:

        (1)   the number and geographic distribution of births in the state;

        (2)   the trained personnel and other departmental resources that may be assigned to the program activities;  and

        (3)   the occurrence or probable occurrence of an urgent situation that requires or will require an unusual commitment of the department's personnel and other resources.

    (c)   The board and the department shall design the program so that the program will:

        (1)   provide information to identify risk factors and causes of birth defects;

        (2)   provide information on other possible causes of birth defects;

        (3)   provide for the development of strategies to prevent birth defects;

        (4)   provide for interview studies about the causes of birth defects;

        (5)   together with other departmental programs, contribute birth defects data to a central registry;

        (6)   provide for the appointment of authorized agents to collect birth defects information;  and

        (7)   provide for the active collection of birth defects information.

    (d)   The board shall adopt rules to govern the operation of the program and carry out the intent of this chapter.  At a minimum, the rules shall:

        (1)   use a medically recognized system to specify the

birth defects to be identified and investigated;

       (2)  select a system for classifying the birth defects according to the public health significance of each defect to prioritize the use of resources;

       (3)  develop a system to select and specify the cases to be investigated;

       (4)  specify a system for selecting the demographic areas in which the department may undertake investigations;  and

       (5)  prescribe the training and experience a person must have for appointment as an authorized agent of the department.

    (e)  In adopting the rules required by Subsection (d), the board shall consider at least:

       (1)  the known incidence and prevalence rates of a birth defect in the state or portions of the state;

       (2)  the known incidence and prevalence rates of a particular birth defect in specific population groups who live in the state or portions of the state;

       (3)  the morbidity and mortality resulting from the birth defect;  and

       (4)  the existence, cost, and availability of a strategy to prevent and treat the birth defect.

    (f)  In addition to providing for the active collection of birth defects information under Subsection (c)(7), the board and the department may design the program to also provide for the passive collection of that information.

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993. Amended by Acts 2003, 78th Leg., ch. 1042, Sec. 1, eff. June 20, 2003.

    Sec. 87.022.  DATA COLLECTION.  (a)  To ensure an accurate source of data necessary to investigate the incidence, prevalence, and trends of birth defects, the board may require a health facility, health professional, or midwife to make available for review by the department or by an authorized agent medical records or other information that is in the facility's, professional's, or midwife's custody or control and that relates to the occurrence of a birth defect specified by the board.

    (b)  The board by rule shall prescribe the manner in which

records and other information are made available to the department.

    (c)    The board shall adopt procedural rules to facilitate cooperation between the health care facility, health professional, or midwife and a department employee or authorized agent, including rules for notice, requests for medical records, times for record reviews, and record management during review.

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993.

    Sec. 87.023.  REFERRAL FOR SERVICES.  A child who meets the medical criteria prescribed by board rule, and the child's family, shall be referred to the department's case management program for guidance in applying for financial or medical assistance available through existing state and federal programs.

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993.

    SUBCHAPTER C. INVESTIGATIONS AND INSPECTIONS

    Sec. 87.041.  INVESTIGATIONS.  (a)  The department may conduct investigations, including epidemiological or toxicological investigations, of cases of specified birth defects.

    (b)  The department may conduct these investigations to determine the nature and extent of the disease or the known or suspected cause of the birth defect and to formulate and evaluate control measures to protect the public health.  The department's investigation is not limited to geographic, temporal, or occupational associations and may include investigation of past exposures.

    (c)  A person shall provide medical, demographic, epidemiological, toxicological, and environmental information to the department under this chapter.

    (d)  A person is not liable in damages or other relief for providing medical or other confidential information to the department during an epidemiological or toxicological investigation.

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993.

    Sec. 87.042.  DEPARTMENTAL INVESTIGATORY POWERS.  To conduct an investigation under this chapter, the commissioner or the commissioner's designee has the same authority to enter, inspect, investigate, and take samples and to do so in the same manner as is provided for communicable diseases under Sections

81.061, 81.063, 81.064, and 81.065.

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993.

Amended by Acts 1995, 74th Leg., ch. 76, Sec. 8.132, eff. Sept. 1, 1995.

<div align="center">SUBCHAPTER D. CENTRAL REGISTRY</div>

Sec. 87.061. REGISTRY; CONFIDENTIALITY. (a) Information collected and analyzed by the department or an authorized agent under this chapter may be placed in a central registry to facilitate research and to maintain security. The department may also store information available from other departmental programs and information from other reporting systems and health care providers.

(b) The department shall use the registry to:

(1) investigate the causes of birth defects and other health conditions as authorized by Texas statutes;

(2) design and evaluate measures to prevent the occurrence of birth defects and other health conditions; and

(3) conduct other investigations and activities necessary for the board and department to fulfill their obligation to protect the health of the public.

(c) The department may store in the central registry information that is obtained from the section of the birth certificate entitled "For Medical and Health Use Only." This information may be used only as provided by Section 191.002(b), relating to the form and contents of the birth certificate.

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993.

Sec. 87.062. ACCESS TO INFORMATION. (a) Access to the central registry information is limited to authorized department employees and other persons with a valid scientific interest who are engaged in demographic, epidemiological, or other studies related to health and who agree in writing to maintain confidentiality.

(b) The department shall maintain a listing of each person who is given access to the information in the central registry. The listing shall include:

(1) the name of the person authorizing access;

(2) the name, title, and organizational affiliation of each person given access;

                    (3)   the dates of access;  and

                    (4)   the specific purpose for which the information was
used.

          (c)   The listing is public information, is open to the public
under Chapter 552, Government Code, and may be inspected during the
department's normal hours of operation.
Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993.
Amended by Acts 1995, 74th Leg., ch. 76, Sec. 5.95(88), eff. Sept.
1, 1995.

          Sec. 87.063.  RESEARCH;   REVIEW  AND  APPROVAL.  (a)  The
commissioner and the department's committee for the protection of
human subjects shall review each research proposal that requests
the use of information in the central registry.  The board shall
adopt rules establishing criteria to be used in deciding if the
research design should be approved.  A proposal that meets the
approval criteria is considered to establish a valid interest as
required by Section 87.062(a), and the commissioner and the
committee shall authorize the researcher to review the records
relevant to the research proposal and to contact cases and
controls.

          (b)  If an investigator using central registry data under a
research design approved under this section believes it is
necessary to contact case subjects and controls, the investigator
must submit a protocol describing the purpose and method to the
commissioner and the department's committee for the protection of
human subjects.   If the contact protocol is approved, the
investigator is considered to have established a bona fide
research, development, or planning purpose and is entitled to carry
out the contacts without securing additional approvals or waivers
from any entity.
Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993.
Amended by Acts 1995, 74th Leg., ch. 76, Sec. 8.133, eff. Sept. 1,
1995.

          Sec. 87.064.  REPORT  OF  CENTRAL  REGISTRY  ACTIVITIES  AND
FINDINGS.  (a)  The department shall publish an annual report of
activities using data contained in the central registry.  The
report shall include:

       (1)  a description of research projects in progress since the last report and the sponsors and principal investigators directing each project;

       (2)  results of the completed research projects either as an abstract or a complete scientific paper that has been reviewed and approved by an appropriate jury;

       (3)  a summary of the statistical information compiled in the registry, including a specific discussion of any clusters, high or low incidences, or prevalences or trends encountered;

       (4)  any policy, research, educational, or other recommendations the department considers appropriate;  and

       (5)  such other information the editors of the report find is appropriate.

    (b)  The department may publish periodic reports in addition to the annual report.

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993.

    Sec. 87.065.  COORDINATION WITH MEXICO.  In developing the central registry and conducting research in areas of this state that border Mexico, the department shall make every effort to coordinate its efforts with similar efforts and research programs in Mexico.

Added by Acts 1993, 73rd Leg., ch. 602, Sec. 1, eff. Sept. 1, 1993.

*Appendix 2.2*

*Table of Birth Defects Legislation*

# Appendix 2.2
# Table of State Birth Defects Legislation

| State Birth Defects Legislation – April 2004 | | | | |
|---|---|---|---|---|
| State | Name of Birth Defects Surveillance Program | Leg/ Rule | Year | Citation |
| **Alabama** | Alabama Birth Defects Surveillance and Prevention Program | Yes | 2001 | Code of Ala. § 22-10A-2 |
| **Alaska** | Alaska Birth Defects Registry (ABDR) | Yes | 1996 (enact) | 7AAC 27.012 |
| **Arizona** | Arizona Birth Defects Monitoring Program | Yes | 1988 (enacted); 1991 (adopted); 2001 (revised) | Statue: ARS § 36-133 Rule: Title 9, Chapter 4, Articles 1 &5 A.R.S. § 36-133 (2001) |
| **Arkansas** | Arkansas Reproductive Health Monitoring System | Yes | 1985 (enacted); 1999 (revised) | Bill 214  (1985) A.C.A. § 20-16-201 |
| **California** | California Birth Defects Monitoring Program | Yes | 1982 (enacted); 1996 (recodified) | Health and Safety Code, Division 102, Part 2, Chapter 1, Sections 103825-103855, effective 1982. Recodified 1996: § 103825, 103855, 103830, 103835, 125050, 103840, 103850, 125000 |
| **Colorado** | Colorado Responds To Children With Special Needs | Yes | 1985 (enacted) | Colorado Revised Statutes 25-1.5-101 - 25-1.5-105 |
| **Connecticut** | Connecticut Birth Defects Registry | Yes | 1991 (enacted) | Sec. 10a-132b transferred to sec 19a-56a in 1999 § 19a-56a, 19a-56b (2001) State has 2 statutes which mandate the reporting of children with birth defects. They are:  Sec. 19a-53. (Formerly Sec. 19-21). Reports of physical defects of children; Sec. 19a-54. (Formerly Sec. 19-21a). Registration of physically handicapped children; Sec. 19a-56a for Birth defects surveillance program, and Sec. 19a-56b, add Sec. 19a-56c Advisory committee. |
| **Delaware** | Delaware Birth Defects Surveillance Project | Yes | 1997 (enacted) | House Bill No. 197, an act to amend Title 16 of Del. Code 16 Code §203 (2000); §201, §202 |
| **District of Columbia** | District of Columbia Birth Defects Surveillance and Prevention Program | No | | |
| **Florida** | Florida Birth Defects Registry | Yes | 1999 | Sec 381.0031 (1, 2) provides for a list of reportable diseases/conditions in Florida.  Congenital anomalies were added in 1999. |
| **Georgia** | 1) Metropolitan Atlanta Congenital Defects Program (MACDP) 2) Georgia Birth Defects Reporting and Information System (GBDRIS) | Yes | 2002 (GBDRIS system) | MACDP: Official Code of GA (OCGA) 31-12-2 GBDRIS: Birth Defects reporting activated statewide in 2002; Citation:  Add GA 31-1-3.2 and DHR Rules 290-5-3-.02 and 290-5-24 |
| **Hawaii** | Hawaii Birth Defects Program | Yes | 1989 (enacted) 2002 (modified) | 8/15/1988 to 6/30/2002 - Hawaii Revised Statutes (HRS), Sections 321-31 and 338-2 in conjunction per Executive Chamber ruling by Governor on 6/16/1989. HRS Sections 324-1 and 324-2 for additional legislative authority (1990 Amendments).  7/1/02 to Present - Act 252 - Relating to Birth Defects (SB 2763, SD 2, HD 2, CD 1). |
| **Idaho** | No birth defects surveillance program | | | |

| State Birth Defects Legislation – April 2004 | | | | |
|---|---|---|---|---|
| State | Name of Birth Defects Surveillance Program | Leg/ Rule | Year | Citation |
| Illinois | Adverse Pregnancy Outcomes Reporting System | Yes | 1985 (enacted) | IL Health and Hazardous Substances Registry Act (410 ILCS 525); 235 ILCS 5/6-24a (2001) |
| Indiana | Indiana Birth Defects and Problems Registry | Yes | 2001 (enacted) | IC 16-38-4, Rule 410 IAC 21-3 |
| Iowa | Iowa Birth Defects Registry | Yes | 1983 (enacted); 2001, 2003 (revised) | IA Code 136A.1, 136A.2, 136A.3, 136A.5, 136A.6; 135.40 Administrative Code of IA 641-1.3(139A); 641-4.1, 641-4.7(136A) (revised 2001) The administrative rules for the Birth Defects Institute (which includes the Registry and other programs such as newborn screening) are undergoing revision in procedures for newborn screening & prenatal screening, but the revision does not affect the Registry's rules. |
| Kansas | Birth Defects Reporting System | Yes | 1979 (enacted) | KSA 65-102 |
| Kentucky | Kentucky Birth Surveillance Registry | Yes | 1992 (enacted); 1997 | KRS 211.651-211.670 (1992); HB372 KRS §211.660, §211.665, §211.670 (2001) KY SB 219 (2002) |
| Louisiana | Louisiana Birth Defects Monitoring Network | Yes | 2001 (enacted) | R.S. 40:31.41-40.31.48, Act No. 194 (2002) |
| Maine | Maine Birth Defects Program | Yes | 1999 | 22MRSA c. 1687 |
| Maryland | Maryland Birth Defects Reporting and Information System | Yes | 1982 | Health-General Article, Section 18-206; Annotated Code of MD |
| Massachusetts | Massachusetts Center For Birth Defects Research and Prevention, Birth Defects Monitoring Program | Yes | 1963 (enacted); 2002 (revised) | Massachusetts General Laws, Chapter 111, Section 67E. In 2002 the Massachusetts Legislature amended this statute, expanding the birth defects monitoring program. The new law: 1) increases mandated reporting up to age three; 2) requires physicians to report to MDPH within 30 days of diagnosis; 3) sets out requirements for the use of this data; 4) requires MDPH to promulgate regulations governing the operation of the Birth Defects Monitoring Program. |
| Michigan | Michigan Birth Defects Registry | Yes | 1988 | Public Health Act 236 of 1988 |
| Minnesota | Minnesota Birth Defects Information System | Yes | 2004 | MS 144.2215 |
| Mississippi | Mississippi Birth Defects Registry | Yes | 1997 | Sec. 41-21-205 of MS Code |
| Missouri | Missouri Birth Defects Registry | No | | |
| Montana | Montana Birth Outcomes Monitoring System | No | | |
| Nebraska | Nebraska Birth Defects Registry | Yes | 1972 | Laws 1972, LB 1203, §1, §2, §3, §4 (alternate citation: Public Health and Welfare [Codes] §71-645, §71-646, §71-647, §71-648, §71-649) |
| Nevada | Nevada Birth Defects Registry | Yes | 1999 | NRS 442.300-442.330; regulation - NAC 442 |
| New Hampshire | New Hampshire Birth Conditions Program | No | | |
| New Jersey | Special Child Health Services Registry | Yes | 1983 (enacted); 2000 (readopted) | Bill 757, NJSA 26:8, NJAC 8:20 (enacted 8-4-1983 with effective date of 3-4-1985) |

| State Birth Defects Legislation – April 2004 | | | | |
|---|---|---|---|---|
| State | Name of Birth Defects Surveillance Program | Leg/ Rule | Year | Citation |
| New Mexico | New Mexico Birth Defects Prevention and Surveillance System | Yes | 2000 (enacted) | In January 2000, birth defects became a reportable condition. These conditions are updated by the Office of Epidemiology. This did not involve legislation, only a change in regulations. |
| New York | New York State Congenital Malformations Registry | Yes | 1982 | Public Health Law Art. 2, Title, II, Sect 225(5)(t) and Art. 2 Title I, sect 206(1)(j): Codes, Rules and Regulations, Chap 1, State Sanitary Code, part 22.3 |
| North Carolina | North Carolina Birth Defects Monitoring Program | Yes | 1995; 2001 (adopted rules) | 15A NCAC 26C.0101-0106 NC adopted rules in 2001 to support the statute. |
| North Dakota | North Dakota Birth Defects Monitoring System | Yes | | ND Centry Code 50-10 |
| Ohio | Ohio Connections for Children with Special Needs | Yes | 2000 (enacted) | House Bill 354 The legislation authorizes the state to have a birth defects system, but until funding is identified/ secured, does not require the Ohio Dept. of Health to implement a system. The document is available at: www.legislature.state.oh.us/BillText123/123_HB_534_5 _Y.htm |
| Oklahoma | Oklahoma Birth Defects Registry | Yes | 1992 | 63 O.S. Sec 1-550.2 (1992) |
| Oregon | No birth defects surveillance program | | | |
| Pennsylvania | Pennsylvania Follow-Up Outreach, Referral and Education For Families | No | | |
| Puerto Rico | Puerto Rico Folic Acid Campaign and Birth Defects Surveillance System | No | | |
| Rhode Island | Rhode Island Birth Defects Surveillance Program | Yes | 2003 | House 5390, Senate 105 (Title 23, Chapter 13.3 of the General Laws) |
| South Carolina | South Carolina Birth Defects Surveillance And Prevention | No | | |
| South Dakota | No birth defects surveillance program | | | |
| Tennessee | Tennessee Birth Defects Surveillance Project | Yes | 2000 | TCA 68-5-506 |
| Texas | Texas Birth Defects Monitoring Division | Yes | 1993 (enacted) | Health and Safety Code, Title 2, Subtitle D, Section 1, Chapter 87. |
| Utah | Utah Birth Defect Network | Yes | 1999 | Birth Defect Rule (R398-5) |
| Vermont | Vermont Birth Information Network | Yes | 2003 | Act 32 |
| Virginia | Virginia Congenital Anomalies Reporting and Education system | Yes | 198 (enacted); 1986, 1988 (amended) | Bill 396, HL 32.1.69.1, Art. 8 1985 (1986) |
| Washington | Washington State Birth Defects Surveillance System | Yes | 2004 | HB1105, notifiable conditions WAC 246-101 (2000) |

| State Birth Defects Legislation – April 2004 | | | | |
|---|---|---|---|---|
| **State** | **Name of Birth Defects Surveillance Program** | **Leg/ Rule** | **Year** | **Citation** |
| **West Virginia** | West Virginia Congenital Abnormalities Registry, Education and Surveillance System | Yes | 1991 (enacted); 2002 (updated) | HB1747, SS Sec: 16-5-12a (1991); 16-40-1 (2002) *old legislation is still in effect but additional legislation that now calls for advisory committee to the Birth Defects Information System has been added in a different section of code. *this occurred during the 2002 session - SB 672. *new code citation is 16-40-1 *legislative rules are in process of being completed for inclusion during the 2004 session. |
| **Wisconsin** | Wisconsin Birth Defects Prevention and Surveillance Program | Yes | 2003 | 1999 WA 114 Statute 253.12; HFS 116--effective April 1, 2003 |
| **Wyoming** | No birth defects surveillance program | | | |

*Appendix 2.3*

**Definitions Used to Determine Covered Entity Status
Under the Privacy Rule**

# Appendix 2.3
# Definitions Used to Determine Covered Entity Status Under the Privacy Rule

**Covered Entity**              A health plan, a health care clearinghouse, or a health care provider who
                                conducts electronic transactions. These transactions are described at 45
                                C.F.R.164.

**Health Care**                 Care, services, or supplies related to the health of an individual. It includes,
                                but is not limited to, the following:

- Preventive, diagnostic, rehabilitative, maintenance, or palliative care,
  and counseling, service, assessment, or procedure with respect to the
  physical or mental condition, or functional status, of an individual or
  that affects the structure or function of the body

- Sale or dispensing of a drug, device, equipment, or other item in
  accordance with a prescription. See 45 C.F.R.160.103

**Covered Transactions**        Transactions for which the Secretary of Health and Human Services has
                                adopted standards and which can be found at 45 C.F.R. Part 162. If a health
                                care provider uses another entity (such as a clearinghouse) to conduct
                                covered transactions in electronic form on its behalf, the health care
                                provider is considered to be conducting the transaction in electronic form.

**Required by Law**             A mandate contained in law that compels an entity to use or to disclose
                                protected health information, and that is enforceable in a court of law.
                                Required by law includes, but is not limited to, court orders and court-
                                ordered warrants; subpoenas or summons issued by a court, grand jury, a
                                governmental or tribal inspector general, or an administrative body
                                authorized to require the production of information; a civil or an authorized
                                investigative demand; Medicare conditions of participation with respect to
                                health care providers participating in the program; and statutes or
                                regulations that require the production of information, including statutes or
                                regulations that require such information if payment is sought under a
                                government program providing public benefits.

**Research**                    Systematic investigation, including research development, testing, and
                                evaluation, designed to develop or contribute to generalizable knowledge.

*Appendix 2.4*

*Office of Civil Rights (OCR) HIPAA Privacy Regulation Text*

# Appendix 2.4

# Office of Civil Rights (OCR) HIPAA Privacy Regulation Text

Below is the actual text of the HIPAA privacy regulation, then the comments from the Privacy Rule's preamble, or the modification guidance issued with the Rule.

*Office for Civil Rights (OCR)/HIPAA Privacy Regulation Text*

§ 164.512 Uses and disclosures for which an authorization or opportunity to agree or object is not required.

A covered entity may use or disclose protected health information without the written authorization of the individual, as described in § 164.508, or the opportunity for the individual to agree or object as described in § 164.510, in the situations covered by this section, subject to the applicable requirements of this section. When the covered entity is required by this section to inform the individual of, or when the individual may agree to, a use or disclosure permitted by this section, the covered entity's information and the individual's agreement may be given orally.

(a) Standard: uses and disclosures required by law.

(1) A covered entity may use or disclose protected health information to the extent that such use or disclosure is required by law and the use or disclosure complies with and is limited to the relevant requirements of such law.

(2) A covered entity must meet the requirements described in paragraph (c), (e), or (f) of this section for uses or disclosures required by law.

(b) Standard: uses and disclosures for public health activities.

(1) Permitted disclosures. A covered entity may disclose protected health information for the public health activities and purposes described in this paragraph to:

(i)  A public health authority that is authorized by law to collect or receive such information for the purpose of preventing or controlling disease, injury, or disability, including, but not limited to, the reporting of disease, injury, vital events such as birth or death, and the conduct of public health surveillance, public health investigations, and public health interventions;…

[Balance of the regulation section omitted. The reader is referred to the OCR website for further details: http://www.os.dhhs.gov/ocr/hipaa/.]

# *Chapter 3*

# *Case Definition*

# Table of Contents

# Appendices

*This document may be reviewed or downloaded from the NBDPN website at:
http://www.nbdpn.org/bdsurveillance.html

# 3.1  Introduction

A *case definition* is a set of criteria that define the parameters of what is included for quantitative description and analysis. In birth defects surveillance a *case* refers to an individual with characteristics that fit into the defined parameters. Important characteristics in birth defects surveillance include the diagnosis, pregnancy outcome information, and demographics.

In the absence of a single national birth defects surveillance program in the United States, pooled data from state-based programs across the country serve to estimate national rates, indicate regional variations, and describe the epidemiology of defects that occur rarely. Because, at any given time, these programs may be in different stages of development, employ different methods of ascertainment, and have different goals and objectives, the elements of the case definition used by each must be clearly identified in order to make valid comparisons and to minimize birth defects rate variations across surveillance programs and among individual defects ascertained by the same program.

Therefore, it is necessary for a surveillance program to develop a clear, concise case definition. Consistent application of a standard definition facilitates the accurate monitoring of clinically relevant conditions, identification of true changes over time, and comparison among populations in order to meet surveillance goals.

In the remainder of this chapter we discuss what is meant by the term 'birth defect' (Section 3.2), some important terminology for case definition (Section 3.3), and case definition criteria (Section 3.4). The relationship between case definition and the two terms 'sensitivity' and 'specificity' is discussed in Section 3.5. References cited in this chapter may be found in Section 3.6. Appendices to this chapter include birth defects included in the NBDPN's case definition (Appendix 3.1), the NBDPN Abstractor's Instructions (Appendix 3.2, available in electronic format at http://www.nbdpn.org/bdsurveillance.html), examples of minor anomalies (Appendix 3.3), and conditions related to prematurity (Appendix 3.4).

# 3.2 What Is Meant by a 'Birth Defect'

The general term 'birth defect' may take on a variety of meanings depending on the context in which it is used and the perspective of the person using it. 'Congenital abnormality', 'congenital anomaly', and 'congenital malformation' are terms often used as synonyms for 'birth defect'. However, the word 'congenital' may describe any condition present at birth, regardless of its etiology or timing of occurrence. In the broadest sense, the term ***birth defect*** encompasses a diversity of conditions including physical malformations, sensory deficits, chromosomal abnormalities, metabolic defects, neurodevelopmental disorders, and complications related to prematurity and low birth weight, among others.

While such a broad definition may be very helpful when seeking legislation and funding for screening, intervention, or prevention programs, a more specific definition is needed for surveillance purposes. Traditionally, birth defects surveillance programs have monitored major structural and genetic defects that adversely affect health and development (Correa-Villaseñor et al., 2003). The specific conditions monitored by an individual program will vary depending on the goals and objectives of that program, the case ascertainment methods used, and the resources available.

# 3.3 Terminology [1]

## General Terminology

**Major anomaly**     A congenital abnormality that requires medical or surgical treatment, has a serious adverse effect on health and development, or has significant cosmetic impact. Individual major anomalies occur in less than 1 percent of the population. Together, they are seen in approximately 3 percent of births. Examples include cleft lip and tracheo-esophageal fistula.

**Minor anomaly**     A congenital abnormality that does not require medical or surgical treatment, does not seriously affect health and development, and does not have significant cosmetic impact. Individual minor anomalies generally occur in less than 4 percent of the population. The presence of multiple minor anomalies in the same child may provide clues to the timing of a prenatal insult and may indicate the presence of an undiagnosed major anomaly, syndrome, or functional deficit. Examples of minor anomalies are listed in Appendix 3.3.

**Normal variant**     A minor anomaly that occurs in approximately 4 percent or more of the population. Examples of normal variants include webbing of the second and third toes and a single umbilical artery in an otherwise normal infant.

## Terminology Related to the Formation of Major Anomalies

**Malformation**     A major anomaly that arises during the initial formation of a structure, i.e., during organogenesis. For most organs, this occurs during the first eight weeks after fertilization. The resulting structure may be abnormally formed, incompletely formed, or may fail to form altogether. Examples of malformations include spina bifida and hypoplastic left heart. The term 'congenital malformation' is also used more broadly to indicate any major anomaly.

**Disruption**     A major anomaly that results from alteration of a structure after its initial formation. The resulting structure may have an altered shape and configuration, abnormal division or fusion of its component parts, or loss of parts that were previously present. Examples of disruption defects include intestinal atresia and possibly gastroschisis.

**Deformation**     A major anomaly that results from molding of part of a structure, usually over a prolonged time, by mechanical forces after its initial formation. Examples of forces that may lead to a deformation include oligohydramnios (diminished amniotic fluid) and intrauterine crowding in twin, triplet, or higher order pregnancies. Examples of deformations include the compression (Potter's) facies seen with bilateral renal agenesis and some instances of clubfoot.

---

[1] Stevenson, et al., 1993; Jones, 1997; Cunningham et al., 2001; Moore, 1977; National Center for Health Statistics, 2002.

## Terminology Related to Patterns of Multiple Anomalies Occurring in a Single Child

**Syndrome**     A pattern of anomalies that form a specific diagnosis for which the natural history and recurrence risk are usually known. Use of the term 'syndrome' implies that the anomalies have a common specific etiology. Examples include Beckwith-Weidemann syndrome and Rubinstein-Taybi syndrome.

**Sequence**     A pattern of anomalies that results from a single primary anomaly or mechanical factor. The presence of the initial anomaly or factor leads to one or more secondary anomalies, which may then lead to one or more tertiary anomalies, etc., in cascade fashion. Examples include Robin sequence (micrognathia, posterior displacement of the tongue, cleft soft palate) and the oligohydramnios, or Potter, sequence (pulmonary hypoplasia, flattened facies, abnormal positioning of the limbs).

**Association**     A nonrandom pattern of anomalies that occur together more frequently than expected by chance alone, but for which no etiology has been demonstrated. Examples include *VACTERL* association (**V**ertebral, **A**nal, **C**ardiac, **T**racheo-**E**sophageal, **R**enal, and **L**imb anomalies) and *CHARGE* association (**C**olobomas, **H**eart defects, choanal **A**tresia, **R**etarded growth and development and/or central nervous system anomalies, **G**enital anomalies and/or hypogondaism, **E**ar anomalies and/or deafness). Use of the term 'association' does not indicate that a specific diagnosis has been made.

## Terminology Related to Tissue and Organ Formation

**Agenesis**     Failure of an organ to form.

**Dysgenesis**     Anomalous or disorganized formation of an organ.

**Aplasia**     Absence of a tissue or organ due to lack of cell proliferation.

**Dysplasia**     Disorganized cell structure or arrangement within a tissue or organ.

**Hypoplasia**     Undergrowth of a tissue or organ due to insufficient proliferation of otherwise normal cells.

**Hyperplasia**     Overgrowth of a tissue or organ due to excess proliferation of otherwise normal cells.

## Terminology Related to the Timing of Gestation and Delivery

**Embryonic period**     The first eight weeks after fertilization, during which most, but not all, organs are formed.

**Fetal period**     The period from the ninth week after fertilization through delivery.

**Neonatal (newborn) period**     The first 28 days following delivery of a live-born infant.

| Terminology Related to the Timing of Gestation and Delivery (continued) | |
|---|---|
| **Prenatal** | Before delivery. |
| **Perinatal** | Before, during, or after delivery. The exact time period may vary from 20 to 28 completed weeks of gestation through 7 to 28 days after delivery, depending on the context in which the term is used. |
| **Postnatal** | After delivery. |

| Terminology Related to Pregnancy Outcome | |
|---|---|
| **Live birth** | Spontaneous delivery of an infant that exhibits signs of life, including a heartbeat, spontaneous breathing, or movement of voluntary muscles. Transient cardiac contractions and fleeting respiratory efforts or gasps are not necessarily considered signs of life by all programs. |
| **Fetal death (stillbirth)** | Spontaneous delivery of an infant or fetus at 20 weeks or greater gestation that does not exhibit signs of life. Transient cardiac contractions and fleeting respiratory efforts or gasps are not necessarily considered signs of life by all programs. A late fetal death is a fetal death that occurs at 28 weeks or greater gestation. |
| **Spontaneous abortion (miscarriage)** | Spontaneous delivery of a fetus at less than 20 weeks gestation. |
| **Induced abortion (elective termination)** | The purposeful interruption of pregnancy with the intention other than to produce a live birth and which does not result in a live birth. |
| **Term infant** | An infant born after 37 completed weeks and before 42 completed weeks of gestation. |
| **Preterm infant** | An infant born before 37 completed weeks of gestation. |
| **Postterm infant** | An infant born after 42 completed weeks of gestation. |
| **Low birth weight** | Birth weight less than 2,500 grams, regardless of gestational age. |
| **Very low birth weight** | Birth weight less than 1,500 grams, regardless of gestational age. |
| **Extremely low birth weight** | Birth weight less than 1,000 grams, regardless of gestational age. |
| **Neonatal death** | Death of a live-born infant within the first 28 days after birth. ***Early neonatal death*** refers to death during the first 7 days. ***Late neonatal death*** refers to death after 7 days but before 29 days. |
| **Infant death** | Death of a live-born infant before 12 months of age. |

# 3.4  Case Definition Criteria

In this section we discuss the various components of the case definition, that is, the criteria a birth defects surveillance program uses to define a case. These include diagnoses to be included (Section 3.4.1), residence (Section 3.4.2), pregnancy outcome (Section 3.4.3), gestational age (Section 3.4.4), age at which defects are diagnosed (Section 3.4.5), as well as the issue of pregnancies resulting from assisted reproductive technology (Section 3.4.6). Each of these criteria is discussed further below.

## 3.4.1  Diagnoses to Be Included

For the purposes of generating and reporting birth defects surveillance data across multiple states, the National Birth Defects Prevention Network (NBDPN) recommends the 45 major anomalies listed in Appendix 3.1. These were chosen on the basis of their frequency, their impact on public health, the state of knowledge about their etiologies and risk factors, and other considerations. Individual surveillance programs may want to expand this list to include additional defects of interest. Programs with limited resources may need to ascertain a subset of this list. Descriptions of each of the 45 diagnoses, its ICD-9-CM and CDC/BPA codes, inclusion and exclusion criteria, and defect-specific information that may be helpful when abstracting medical records are provided in the NBDPN Abstractor's Instructions posted on the NBDPN website (see Appendix 3.2). Examples of conditions considered to be minor anomalies are provided in Appendix 3.3. Conditions related to prematurity that are not considered to be major anomalies are listed in Appendix 3.4.

## 3.4.2  Residence

When monitoring the frequency of any condition, it is critical to define the population in which the cases occur. This allows one to calculate rates within the population, evaluate changes in these rates over time, plan for prevention and intervention services, and assess program goals and effectiveness. Population-based birth defects surveillance programs should strive to ascertain defects that occur among the offspring of all women who reside within a defined geographic area at the time of pregnancy outcome.

While this charge for surveillance programs appears straightforward, there are some special considerations. One such consideration is the fact that women who reside in one state or community may travel outside that area – such as to an adjacent state, specialty care center, or military facility – for obstetric care. In these instances, the mother's place of residence at the time of delivery (rather than the actual location of the delivery) should be used to determine whether to include her pregnancy in the surveillance. Including in-area residents who deliver outside the surveillance area, and excluding out-of-area residents who deliver within the surveillance area, is essential in order to conduct comprehensive surveillance. Whether an individual program attains this level of comprehensiveness will depend on how frequently women travel outside the surveillance area for delivery, the magnitude of the potential impact this may have on defect rates, the staff and resources available, and, most importantly, the existence of data-sharing relationships with facilities and programs outside the surveillance area. Recent changes in regulations concerning the privacy of medical records under the Health Information Portability and Accountability Act (HIPAA) have added to the complexity of these data-sharing relationships. The HIPAA regulations are discussed further in Chapter 2 on Legislation.

Another consideration is the fact that a surveillance program may identify more than one residential address for an individual woman. For example, the address of the health insurance policyholder listed in a hospital delivery record may differ from the mother's address listed on a birth certificate. If a patient changes residence during pregnancy, programs that employ multisource ascertainment may identify one

address from prenatal or laboratory records and another from the hospital delivery record. For these reasons, surveillance programs should develop standard procedures for deciding which of multiple addresses to accept as the mother's residence at delivery. Usually, this is the address at the time of delivery as listed on the vital record. If a vital record is not available or is not generated, as when an elective termination is performed outside the hospital setting, considering the mother's address from the termination record or from the prenatal visit closest in time to the delivery may be appropriate alternatives.

A third consideration requires detailing the method of determining whether a particular address lies within the surveillance area, particularly if the population under surveillance is not that of an entire state. For example, zip codes often cross city or county boundaries, streets may be renamed, new zip codes may be added, and city or county boundaries may change over time. Addresses that contain only a post office box number do not provide information about a person's actual place of residence. For these reasons, surveillance programs should develop standard procedures for distinguishing addresses that lie within or outside their surveillance area. Potential reference sources include street maps, United States Postal Service listings, tax assessor records, census tracts, and the latitude and longitude of the surveillance area (geocoding). While the latter can be extremely precise, the accuracy of geocoding will depend upon the accuracy of the addresses to which the latitude and longitude are assigned.

# 3.4.3 Pregnancy Outcome

Ideally, for births defects surveillance to be comprehensive with high sensitivity, all defects occurring in a population should be ascertained regardless of whether a pregnancy ends in live birth, fetal death, or spontaneous abortion, or whether an elective termination is performed. It is estimated that approximately 10 to 15 percent of all recognized pregnancies end in spontaneous abortion, and approximately 6 to 7 percent of those that reach 20 weeks gestation end in fetal death (Gabbe et al., 1996; National Center for Health Statistics, 2001). Surveillance systems that ascertain defects only among live-born infants may report incomplete data for defects that occur frequently among these outcomes. However, it is important to recognize that even late fetal deaths may not be scrutinized for defects as closely or as systematically as are live births. Unless an autopsy (including internal examination) and chromosome analysis are performed routinely, defects present in fetal deaths, yet not immediately evident in the delivery room, may remain unidentified. Even if an autopsy and chromosome analysis are performed, the presence of minor defects may not be recognized and syndromes may not be diagnosed. Whether it is beneficial for an individual program to ascertain defects reported in outcomes other than live births will depend upon the program's goals and objectives, the staff and resources available, the accessibility of information about these outcomes, and the magnitude of the potential impact on individual defect rates of excluding them.

The development and widespread use of prenatal diagnostic technology has posed additional issues for birth defects surveillance. These procedures have provided women with the option of electively terminating affected pregnancies, particularly those with defects that are life-threatening or that are likely to result in significant mental or functional impairment, usually before 20 weeks gestation. In the absence of prenatal diagnosis, many of these pregnancies would end in live birth or fetal death and would be included in birth defects surveillance data from many programs. Failure to ascertain prenatal diagnoses among electively terminated pregnancies may, therefore, limit the comprehensiveness and sensitivity of surveillance programs for some defects, such as neural tube defects and chromosomal abnormalities, even when defects among fetal deaths are ascertained. And – because the availability and utilization of prenatal diagnosis and elective termination may vary among populations, across geographic regions, and over time – the ability to make valid comparisons of some defect rates may be compromised unless pregnancies electively terminated after prenatal diagnosis are regularly ascertained.

Unfortunately, including these prenatal diagnoses will likely require expansion of a program's case ascertainment sources to include settings such as prenatal diagnostic clinics and termination centers. Furthermore, as is the case with fetal deaths, pregnancies that are electively terminated may not be fully scrutinized for confirmation of the prenatal diagnosis or the presence of additional defects or syndromes upon completion of the procedure. Again, whether it is beneficial for an individual program to ascertain defects reported in these pregnancies will depend upon the program's goals and objectives, the staff and resources available, the accessibility of information about these outcomes, and the magnitude of the potential impact on individual defect rates of excluding them. Regardless, it is important for birth defects surveillance programs to clearly state which outcomes are included when reporting surveillance data and to include pregnancies electively terminated after prenatal diagnosis whenever possible.

## 3.4.4  Gestational Age

Another important component of the case definition is the gestational age at delivery of the cases included in the surveillance data. The frequency of some defects may vary by gestational age, leading to variations in their rates depending on the length of gestation. For example, some defects are identified more frequently among preterm infants (Rasmussen et al., 2001; Shaw et al., 2001). Others, such as patent ductus arteriosus and undescended testes, may be abnormal in term infants but physiologically normal in preterm infants. Some ventricular septal defects that are present at birth in preterm infants might have closed during the last weeks of gestation if the pregnancy had continued to term. If surveillance systems differ in the gestational age at delivery of cases they include, or in their use of exclusion criteria based on gestational age, their rates of some defects may not be comparable.

Again, the inclusion of pregnancies electively terminated after prenatal diagnosis poses additional issues. Many of these pregnancies would have delivered spontaneously at a considerably later gestational age had they not been terminated. In order not to underestimate the frequency of defects for which elective termination may be performed, pregnancies terminated after prenatal diagnosis should be included in surveillance data regardless of the gestational age at which they were terminated. However, this may slightly overestimate the frequency of some defects relative to their frequency in the absence of prenatal diagnosis. For example, the majority of pregnancies electively terminated before 20 weeks gestation would have otherwise continued beyond 20 weeks to be included in birth defects surveillance programs that monitor pregnancies of 20 weeks or greater. However, a small proportion might have ended in spontaneous abortion before 20 weeks and would not appropriately be included in data from these programs. While the frequency of spontaneous abortion for pregnancies with Down syndrome has been estimated for each week of gestation, the natural history of pregnancies with other defects has not been well described (Hook et al., 1995). The frequency of spontaneous abortion by gestational week probably varies depending on the defect. Unfortunately, this effect is likely to be greater the earlier in gestation that affected pregnancies are terminated.

For the purposes of generating and reporting birth defects surveillance data across multiple states, the NBDPN recommends monitoring defects among live births and fetal deaths of 20 weeks or greater and among pregnancies electively terminated after prenatal diagnosis at any gestational age. Gestational age may be derived in various ways based on the date of the last menstrual period, measurement of the fetus by prenatal ultrasound, or the newborn clinical exam. Because these methods may not be equally accurate and may yield conflicting results, an important consideration is which method to use to determine whether a case fulfills the gestational age criterion for inclusion in surveillance data (Alexander et al., 1990; Hall, 1990). The methods below are listed in descending order of their generally accepted accuracy for calculating gestational age:

> ➢ Prenatal ultrasound with a reported gestational age of less than 14 weeks

> ➢ Date of the last menstrual period

> ➢ Prenatal ultrasound with a reported gestational age of 14 weeks or greater

> ➢ Clinical examination after delivery

When multiple estimates of gestational age are ascertained for an individual case, the NBDPN recommends that the value derived using the method highest on this list be used to determine case status. Regardless of which method is used, it is important for birth defects surveillance programs to clearly state the gestational ages of the cases included when reporting surveillance data.

## 3.4.5  Age at Which Defects Are Diagnosed

The age at which a defect is diagnosed is also an important component of the case definition. The frequency of some defects may vary depending on the age of the child at diagnosis. While defects that are visible in the delivery room or symptomatic shortly after birth may be ascertained by most surveillance systems with high sensitivity, some internal defects may not be apparent for weeks or months after birth. Examples include cardiac defects that do not produce overt cyanosis, such as many atrial or ventricular septal defects, many obstructive renal defects, and some instances of intestinal malrotation. In addition, some chromosomal abnormalities may not be diagnosed until a year or more after birth when developmental delay or behavioral symptoms prompt a more in-depth evaluation. The rates of such conditions reported by surveillance systems that ascertain defects only among infants in the newborn nursery may not be comparable with those from systems that ascertain defects among older infants and children.

For the purposes of generating and reporting birth defects surveillance data across multiple states, the NBDPN recommends monitoring defects among live-born infants up to one year of age. Whether an individual program is able to ascertain defects beyond the newborn period will depend on the accessibility of information from sources other than the newborn nursery and the availability of staff and resources to add these additional sources. Programs should regularly state the range of ages at diagnosis included when reporting surveillance data.

As with other case definition criteria, the inclusion of defects that are diagnosed prenatally poses additional issues. The sensitivity and specificity of fetal ultrasound may vary for different defects depending on the gestational age, the skill and experience of the technician, the presence of maternal obesity, and other factors. The sensitivity and specificity of fetal ultrasound also may differ from that of newborn ultrasound and other postnatal diagnostic procedures. In addition, some conditions identified at mid-gestation by prenatal ultrasound may resolve spontaneously before delivery. Examples include renal obstructions, such as pyelectasis and uretero-pelvic junction obstructions, choroid plexus cysts of the brain, and some ventricular septal defects. Even chorionic villus sampling (CVS) may yield placental cells that contain chromosomal mosaicism not actually present in the fetus. For these reasons, many abnormalities diagnosed or suspected prenatally must be evaluated postnatally to determine their true nature. When such postnatal assessment is not possible or the medical records are not available, decisions about whether to include these defects in the surveillance must be made individually based on the certainty and specificity of the prenatal diagnosis for each case. General abstractor's instructions for the inclusion and exclusion of prenatal diagnoses for the 45 defects reported by the NBDPN are provided in Appendix 3.2. When reporting surveillance data, it is important for birth defects surveillance programs to state clearly the ages at which the defects were diagnosed and whether prenatal diagnoses without postnatal confirmation are included.

## 3.4.6  Pregnancies Resulting from Assisted Reproductive Technology

The use of assisted reproductive technology raises unique issues for birth defects surveillance, particularly in pregnancies where the egg from one woman (the biological mother) is used to conceive, but the pregnancy is carried by another woman (the birth mother). In this instance, genetic characteristics of the biological mother will be transmitted to the infant, but the birth mother's environment and lifestyle during pregnancy may also affect the infant. This situation may become quite complex when examining etiologic factors for birth defects. However, for surveillance purposes, the NBDPN recommends that the person listed on the child's birth certificate should be mother of record.

# 3.5 Case Definition and Sensitivity and Specificity

Use of a consistent case definition is critical when evaluating the sensitivity and specificity of surveillance data and the efficiency and utility of surveillance programs.

The *sensitivity* of a surveillance program is defined as the proportion of cases occurring within a population that the program ascertains. Factors that may affect the sensitivity of a birth defects surveillance program include which pregnancy outcomes are ascertained (live births, fetal deaths, elective terminations), the gestational age at which they are ascertained (term infants only, pregnancies $\geq 20$ weeks, all pregnancies), the child's age at the time the defect is diagnosed (prenatally, in the newborn period, before one year, at any age), and the diagnostic setting and methods used for ascertainment. For example, defects that are symptomatic in a live born infant may not be recognized in pregnancies that end in fetal death unless an autopsy is performed. Defects that are not immediately life-threatening, such as many cardiac septal defects, may not be diagnosed until several weeks or months after birth. If managed solely in the outpatient setting, these defects may be missed entirely by hospital-based programs unless surgical correction is required.

The *specificity* of a surveillance program is defined as the proportion of cases within a population that are ascertained by the program and that truly have defects. Factors that affect the sensitivity of a birth defects surveillance program may also affect its specificity. For example, patent ductus arteriosus (PDA) may be entirely normal in a preterm or one-day-old term infant but distinctly abnormal in a three-month-old child. Inclusion of all occurrences of PDA, regardless of gestational or postnatal age, may lead to ascertainment of false positive cases. Variations in the quality of prenatal ultrasound and in the natural course of some prenatal conditions necessitate postnatal confirmation of many diagnoses to avoid including false positive or clinically nonsignificant cases. Such confirmation may not be possible for pregnancies that end in fetal death or elective termination unless fetal autopsies are performed. Similarly, the exact nature of a congenital heart defect may not be finalized until the time of corrective surgery.

# 3.6  References

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## *Appendix 3.1*

## *Birth Defects Included in the Case Definition of the National Birth Defects Prevention Network*

# Appendix 3.1
# Birth Defects Included in the Case Definition of the National Birth Defects Prevention Network

| Birth Defects | ICD-9-CM Codes | CDC/BPA Codes |
|---|---|---|
| **Central Nervous System** | | |
| Anencephalus | 740.0 - 740.1 | 740.00 - 740.10 |
| Spina bifida without anencephalus | 741.0, 741.9 w/o 740.0 -740.10 | 741.00 - 741.99 w/o 740.0 - 740.10 |
| Hydrocephalus without Spina Bifida | 742.3 w/o 741.0, 741.9 | 742.30 - 742.39 w/o 741.00 - 741.99 |
| Encephalocele | 742.0 | 742.00 - 742.09 |
| Microcephalus | 742.1 | 742.10 |
| **Eye** | | |
| Anophthalmia/microphthalmia | 743.0, 743.1 | 743.00 - 743.10 |
| Congenital cataract | 743.30 - 743.34 | 743.32 - 743.326 |
| Aniridia | 743.45 | 743.42 |
| **Ear** | | |
| Anotia/microtia | 744.01, 744.23 | 744.01, 744.21 |
| **Cardiovascular** | | |
| Common truncus | 745.0 | 745.00 - 745.01 |
| Transposition of great arteries | 745.10, .11, .12, .19 | 745.10 - 745.19 |
| Tetralogy of Fallot | 745.2 | 745.20 - 745.21, 746.84 |
| Ventricular septal defect | 745.4 | 745.40 - 745.490 (exclude 745.498) |
| Atrial septal defect | 745.5 | 745.50 - 745.59 (exclude 745.50) |
| Endocardial cushion defect | 745.60, .61, .69 | 745.60 - 745.69 |
| Pulmonary valve atresia and stenosis | 746.01, 746.02 | 746.00 - 746.01 |
| Tricuspid valve atresia and stenosis | 746.1 | 746.10 (exclude 746.105) |
| Ebstein's anomaly | 746.2 | 746.20 |
| Aortic valve stenosis | 746.3 | 746.30 |
| Hypoplastic left heart syndrome | 746.7 | 746.70 |
| Patent ductus arteriosus (Include only if weight=>2500 grams or note if unable to exclude <2500 grams infants.) | 747.0 | 747.00 |
| Coarctation of aorta | 747.10 | 747.10 - 747.19 |
| **Orofacial** | | |
| Cleft palate without cleft lip | 749.0 | 749.00 - 749.09 |
| Cleft lip with and without cleft palate | 749.1, 749.2 | 749.10 - 749.29 |
| Choanal atresia | 748.0 | 748.00 |

| Gastrointestinal | | |
|---|---|---|
| **Birth Defects** | **ICD-9-CM Codes** | **CDC/BPA Codes** |
| Esophageal atresia/tracheoesophageal fistula | 750.3 | 750.30 - 750.35 |
| Rectal and large intestinal atresia/stenosis | 751.2 | 751.20 - 751.24 |
| Pyloric stenosis | 750.5 | 750.51 |
| Hirshsprung's disease (congenital megacolon) | 751.3 | 751.30 - 751.34 |
| Biliary atresia | 751.61 | 751.65 |
| Genitourinary | | |
| Renal agenesis/hypoplasia | 753.0 | 753.00 - 753.01 |
| Bladder exstrophy | 753.5 | 753.50 |
| Obstructive genitourinary defect | 753.2, 753.6 | 753.20-29 - 753.60- 69 |
| Hypospadias and Epispadias | 752.61, 752.62 | 752.600 - 752.627 (excluding 752.621) |
| Musculoskeletal | | |
| Reduction deformity, upper limbs | 755.20 - 755.29 | 755.20 - 755.29 |
| Reduction deformity, lower limbs | 755.30 - 755.39 | 755.30 - 755.39 |
| Gastroschisis | 756.79 | 756.71 |
| Omphalocele | 756.79 | 756.70 |
| Congenital hip dislocation | 754.30, .31, .35 | 754.30 |
| Diaphragmatic hernia | 756.6 | 756.610 - 756.617 |
| Chromosomal | | |
| Trisomy 13 | 758.1 | 758.10 - 758.19 |
| Down syndrome (Trisomy 21) | 758.0 | 758.00 - 758.09 |
| Trisomy 18 | 758.2 | 758.20 - 758.290 |
| Other | | |
| Fetal alcohol syndrome | 760.71 | 760.71 |
| Amniotic bands | No code | 658.80 |

*Appendix 3.2*
*NBDPN Abstractor's Instructions*

# Appendix 3.2
# NBDPN Abstractor's Instructions

# Appendix 3.2
# NBDPN Abstractor's Instructions

## Format for Birth Defect Descriptions

| **Defect Name** | |
|---|---|
| **Description** | Description of the defect. |
| **Inclusions** | Other names or conditions that should be included in the code for the defect. |
| **Exclusions** | Other names or conditions that should not be included in the code for the defect. |
| **ICD-9-CM Codes** | Applicable ICD-9-CM codes for the defect. |
| **CDC/BPA Codes** | Applicable CDC/BPA codes for the defect. |
| **Diagnostic Methods** | Postnatal procedures by which the defect may be accurately and reliably diagnosed. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | Guidance on whether cases with only a prenatal diagnosis should be included in the defect code. |
| **Additional Information** | Tips and useful information about the defect. |

## Anencephalus

| | |
|---|---|
| **Description** | Partial or complete absence of the brain and skull. |
| **Inclusions** | Acrania – Absence of skull bones with some brain tissue present. <br> Absent brain, with or without skull bones present. <br> Anencephalus <br> Anencephaly <br> Craniorachischisis – Anencephaly continuous with an open posterior spinal defect with no meninges covering the neural tissue. |
| **Exclusions** | Encephalocele <br> Iniencephaly <br> Rachischisis – When used alone, this term refers only to the spinal defect and should be coded as spina bifida without anencephalus. |
| **ICD-9-CM Codes** | 740.0 – 740.1 |
| **CDC/BPA Codes** | 740.00 – 740.10 |
| **Diagnostic Methods** | Anencephalus is easily recognized on physical examination at delivery. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | Anencephalus may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. |

**Additional Information:**

Anencephalus is one of a group of defects that result from failure of the neural tube to close.

Maternal serum alphafetoprotein (MSAFP) and/or amniotic fluid alphafetoprotein (AFAFP) and amniotic fluid acetylcholinesterase (ACHE) may be elevated with anencephalus. However, these screening tests alone are not sufficient to diagnose the condition.

In cases where both anencephalus and spina bifida are present but are not continuous (i.e., not craniorachischisis), both anencephalus and spina bifida should be coded.

## Spina Bifida without Anencephalus

| | |
|---|---|
| **Description** | Incomplete closure of the vertebral spine (usually posteriorly) through which spinal cord tissue and/or the membranes covering the spine (meninges) herniate. |
| **Inclusions** | Lipomeningocele<br>Lipomyelomeningocele<br>Meningocele – Herniation of meninges only.<br>Meningomyelocele, Myelomeningocele – Herniation of meninges and spinal cord tissue.<br>Myelocystocele<br>Myelodysplasia<br>Myeloschisis<br>Open spina bifida<br>Rachischisis – Open spina bifida without meninges covering the spinal cord tissue.<br>Spina bifida aperta<br>Spina bifida cystica |
| **Exclusions** | Closed spina bifida<br>Diastematomyelia<br>Diplomyelia<br>Hydromyelia<br>Spina bifida with coexisting anencephalus – Code only as anencephalus.<br>Spina bifida occulta<br>Syringomyelia<br>Tethered spinal cord |
| **ICD-9-CM Codes** | 741.0 or 741.9 without 740.0 – 740.1 |
| **CDC/BPA Codes** | 741.00 – 741.99 without 740.00 – 740.10 |
| **Diagnostic Methods** | The majority of defects result in a direct opening on the infant's back that is easily recognized on physical examination at delivery. However, the exact nature of the defect (meningocele vs. myelomeningocele) may only be distinguished by CT or MRI scan, at surgery, or at autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | Spina bifida may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. In addition, the absence of spina bifida on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery. |

**Additional Information:**
Spina bifida is one of a group of defects that result from failure of the neural tube to close.

Open lesions (spina bifida cystica, spina bifida aperta) are those with no covering or with only meninges covering the neural tissue. They usually leak cerebrospinal fluid. Closed lesions are covered by normal skin.

Closed lesions, or spina bifida occulta, do not produce an opening in the infant's back and may result only in a defect of the vertebral spine without significant herniation of neural tissue or neurologic impairment. When asymptomatic, it may be detected as an incidental finding on an x-ray or other test performed for a different indication.

Hydrocephalus and Arnold-Chiari malformation of the brain frequently, though not always, result from spina bifida. When present, there is no need to code them separately from the spina bifida.

Maternal serum alphafetoprotein (MSAFP) and/or amniotic fluid alphafetoprotein (AFAFP) and amniotic fluid acetylcholinesterase (ACHE) may be elevated in spina bifida. However, these screening tests alone are not sufficient to diagnose the condition.

In cases where both anencephalus and spina bifida are present but are not continuous (i.e., not craniorachischisis), both anencephalus and spina bifida should be coded.

If the defect coding system includes unique codes for different levels of spina bifida (cervical; thoracic; lumbar; sacral) and a defect involves more than one level (cervicothoracic; thoracolumbar; lumbosacral), the highest level at which it occurs should be coded (i.e., cervical; thoracic; lumbar). The highest level of involvement determines the degree of associated neurologic impairment.

## Hydrocephalus without Spina Bifida

| | |
|---|---|
| **Description** | An increase in the amount of cerebrospinal fluid (CSF) within the brain resulting in enlargement of the cerebral ventricles and increased intracranial pressure. |

**Inclusions**

Aqueductal stenosis – Narrowing or incomplete patency of the aqueduct of Sylvius between the third and fourth ventricles. This is the most common type of obstructive hydrocephalus (see below).

Atresia of the foramina of Magendie and Luschka – Incomplete patency of the openings in the roof of the fourth ventricle through which CSF normally flows out of the brain.

Communicating hydrocephalus – Impaired absorption of CSF, leading to an increased amount of CSF within the brain.

Dandy-Walker malformation
Hydranencephaly
Hydrocephalus, type not specified
Obstructive (noncommunicating) hydrocephalus – Obstruction of the flow of CSF within or out of the brain.

**Exclusions**

Hydrocephalus that results from a prior intracranial hemorrhage. This may be seen particularly in preterm infants.

Hydrocephalus that occurs in association with spina bifida. Only the appropriate spina bifida code should be used.

Ventriculomegaly

**ICD-9-CM Codes**          742.3 without 741.0 or 741.9

**CDC/BPA Codes**          742.30 – 742.39 without 741.00 – 741.99

**Diagnostic Methods**

While severe cases may be suspected by physical examination at delivery, hydrocephalus may be conclusively diagnosed only through direct visualization of the brain by cranial ultrasound, CT or MRI scan, surgery, or autopsy. While a child's head circumference may be increased for age, this measurement alone is not sufficient to make the diagnosis.

**Prenatal Diagnoses Not Confirmed Postnatally**

While hydrocephalus may be identified by prenatal ultrasound, it generally should not be included in surveillance data without postnatal confirmation. However, if it is possible to ascertain the degree of certainty of the diagnosis on prenatal ultrasound, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Severe cases may be

included without postnatal confirmation. Live-born children who survive should always have confirmation of the defect postnatally before being included.

**Additional Information:**

Hydrocephalus has a variety of etiologies, including infection, hemorrhage, and tumors, as well as anatomic lesions of the brain such as agenesis of the corpus callosum, encephalocele, cysts, and some bone dysplasias. In many cases, the etiology is not known.

In its true form, Dandy-Walker malformation is a malformation of the cerebellum and not a form of hydrocephalus. However, the term Dandy-Walker variant has been used to denote atresia of the foramina of Magendie and Luschka, dilatation of the cisterna magna (the space between the cerebellum and the brainstem), or cerebellar cysts, all of which have the appearance of increased fluid in the posterior fossa of the brain. It is, somewhat incorrectly, included in the defect codes for hydrocepahlus.

In hydranencephaly, the cerebral hemispheres are largely replaced by fluid-filled sacs within a normal skull. Hydranencephaly is not a true form of hydrocephalus. It is, somewhat incorrectly, included in the defect codes for hydrocepahlus.

Ventriculomegaly refers to enlargement of the cerebral ventricles, as measured by ultrasound (either prenatal or postnatal), CT or MRI scan. The distinction between hydrocephalus and ventriculomegaly has not been clearly defined, and these terms may be used interchangeably. Ventriculomegaly may be described as mild, moderate, or severe. How these designations correlate with the presence of true hydrocephalus, particularly when seen on prenatal ultrasound, also has not been clearly defined.

## Encephalocele

| | |
|---|---|
| **Description** | Herniation of brain tissue and/or meninges through a defect in the skull. The hernia sac is usually covered by skin. |
| **Inclusions** | Cephalocele<br>Cranial meningocele – Herniation of meninges only.<br>Encephalocele<br>Encephalomyelocele - Herniation through a defect in a portion of both the skull and the upper spine.<br><br>Encephalocystomeningocele<br>Hydranencephalocele<br>Meningoencephalocele<br>Ventriculocele |
| **Exclusions** | NA |
| **ICD-9-CM Codes** | 742.0 |
| **CDC/BPA Codes** | 742.00 – 742.09 |
| **Diagnostic Methods** | Most cases of encephalocele are recognizable on physical examination after delivery. However, they may be conclusively diagnosed only through direct visualization of the brain by cranial ultrasound, CT or MRI scan, surgery, or autopsy. This is particularly true for internal herniations through the sphenoid, maxillary, or ethmoid bones, the orbit, or pharynx. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | Encephalocele may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. In addition, the absence of a small encephalocele on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery. |

**Additional Information:**

Encephaloceles are often included as one of a group of defects that result from failure of the neural tube to close.

Maternal serum alphafetoprotein (MSAFP) and/or amniotic fluid alphafetoprotein (AFAFP) and amniotic fluid acetylcholinesterase (ACHE) may be elevated with encephaloceles. However, these screening tests alone are not sufficient to diagnose the condition.

Occipital encephalocele is a component of Meckel-Gruber syndrome.

## Microcephalus

**Description**

A cranial vault that is smaller than normal for age. The size of the cranial vault is an indicator of the size of the underlying brain.

**Inclusions**

Microcephalus
Microcephaly
Primary or True Microcephalus

**Exclusions**

Microcephalus that is secondary to a birth or delivery complication or to a postnatal insult or trauma.

**ICD-9-CM Codes**

742.1

**CDC/BPA Codes**

742.10

**Diagnostic Methods**

Microcephaly is usually easily diagnosed on physical examination by measurement of the occipitofrontal circumference (OFC, head circumference). However, there is difference of opinion as to what the lower limit of a normal head circumference should be (see below). Cranial ultrasound, CT or MRI scans may also reflect the diagnosis and contribute to the diagnosis of any underlying brain abnormalities.

**Prenatal Diagnoses Not Confirmed Postnatally**

While this condition may be identified by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of microcephalus on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

**Additional Information:**

Microcephalus may be defined variously as an OFC less than the 10th, 5th, or 3rd percentile, or less than 2 or 3 standard deviations below the mean for age. There is no single accepted standard. Reference graphs differ in terms of the cut-off values displayed and the reference population used. Reference graphs for postnatal OFC growth usually are displayed separately for males and females.

In addition, it must be recognized that a proportion of normal children will have an OFC below any single cut-off value (i.e., 5% of the population has an OFC below the 5th percentile by definition). For this reason, only children who have been given a clinical diagnosis of microcephalus should be included in birth defects surveillance data. The diagnosis should not be assigned based on the OFC measurement at birth without corroborating evidence from the medical record that the child carries the diagnosis of microcephalus.

Microcephalus itself is not a primary malformation, but a sign that the brain is small. It has a wide variety of causes. It is a component of a number of genetic syndromes. It also may result from a primary brain abnormality or a prenatal, perinatal, or postnatal insult. Examples of the latter include intrauterine infection, such as rubella or cytomegalovirus (CMV); *in utero* exposure to

alcohol and some medications, such as isotretinoin or dilantin; hypoxia during delivery; chronic hypoxia complicating prematurity; postnatal meningitis; head trauma. Only cases of microcephalus that have onset before delivery should be included in surveillance data. Unfortunately, the timing of onset and the etiology often are not known.

## Anophthalmia/Microphthalmia

| | |
|---|---|
| **Description** | Anophthalmia – Total absence of eye tissue or apparent absence of the globe in an otherwise normal orbit.<br><br>Microphthalmia – Reduced volume of the eye. The corneal diameter is usually less than 10 millimeters, or the anteroposterior globe diameter is less than 20 millimeters. |
| **Inclusions** | Anophthalmia<br>Microphthalmia<br>Nanophthalmia – Microphthalmia with normal internal eye (intraocular) structures. This is a distinct genetic condition. |
| **Exclusions** | Small eyes or small palpebral fissures for which the diagnosis of microphthalmia or anophthalmia has not been made.<br><br>Microcornea with otherwise normal eye size. |
| **ICD-9-CM Codes** | 743.0,  743.1 |
| **CDC/BPA Codes** | 743.00 – 743.10 |
| **Diagnostic Methods** | These conditions are usually recognized on physical examination after delivery, especially by an ophthalmologist. However, the anteroposterior diameter of the globe may be measured only by ultrasound, CT or MRI scan, or at autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | While these conditions may be identified by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. In addition, the absence of anophthalmia or microphthalmia on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery. |

**Additional Information:**

Microphthalmia may occur in association with colobomas (gaps) in the uvea, iris, choroid and/or optic nerve (colobomatous microphthalmia).

Anophthalmia and microphthalmia often are accompanied by malformations of the brain and face, and frequently are components of genetic syndromes.

## Congenital Cataract

| | |
|---|---|
| **Description** | An opacity of the lens of the eye that has its origin prenatally. |
| **Inclusions** | Anterior polar cataract<br>Cataract, type not specified<br>Infantile cataract<br>Lamellar cataract<br>Nuclear cataract<br>Posterior lentiglobus/lenticonus cataract<br>Posterior cortical cataract<br>Sectoral cataract<br>Zonular cataract |
| **Exclusions** | Any of the above types of cataract that has its origin after birth<br>Corneal opacities |
| **ICD-9-CM Codes** | 743.30 – 743.34 |
| **CDC/BPA Codes** | 743.320 – 743.326 |
| **Diagnostic Methods** | Some cataracts are readily apparent on physical examination. Others are visible with an ophthalmoscope. However, they may be conclusively diagnosed only through examination by an ophthalmologist. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | While this condition may be identified by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of a cataract on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery. |

**Additional Information:**

Cataracts may be congenital, acquired, or inherited. They may involve all or only part of the lens of either or both eyes. They may be an isolated finding in an otherwise normal eye, or may be part of a more general eye malformation. They may be seen with metabolic disorders, such as galactosemia; genetic syndromes, such as chondrodysplasia punctata; chromosomal abnormalities, such as Trisomy 21; intrauterine infection, such as congenital rubella; or trauma.

In some instances, the severity of the cataract progresses over time. The need for surgical treatment depends on the degree of visual impairment.

When congenital cataract occurs with microphthalmia in the same infant, both conditions should be coded.

## Aniridia

| | |
|---|---|
| **Description** | Hypoplasia of the iris of both eyes. |
| **Inclusions** | Aniridia<br>Hypoplasia of the iris |
| **Exclusions** | Axenfeld-Rieger syndrome<br>Chandler syndrome<br>Coloboma of the iris<br>Iris atrophy<br>Peters anomaly<br>Rieger syndrome |
| **ICD-9-CM Codes** | 743.45 |
| **CDC/BPA Codes** | 743.42 |
| **Diagnostic Methods** | Aniridia may be apparent on physical examination. However, it may be conclusively diagnosed only through examination by an ophthalmologist. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | Aniridia should not be included in surveillance data unless diagnosed postnatally. |

**Additional Information:**

Aniridia is usually associated with other abnormalities of the eye, including a persistent pupillary membrane; displaced lens; glaucoma; corneal and retinal abnormalities; hypoplasia of the optic nerve. While there is often near-total absence of the iris, it is never completely absent. Aniridia may be a component of Peters anomaly (abnormal development of the cornea and anterior chamber of the eye).

Aniridia has been associated with Wilms tumor of the kidney and certain chromosomal abnormalities. It may be inherited or may occur sporadically.

When aniridia occurs with a cataract in the same infant, both conditions should be coded.

## Anotia/Microtia

| | |
|---|---|
| **Description** | Anotia – Total absence of the external ear and canal. |
| | Microtia – Malformation or hypoplasia of the external ear (auricle, pinna). |
| **Inclusions** | Anotia<br>Microtia |
| **Exclusions** | Small ears that retain most of the overall structure of the normal auricle, including lop or cup ear defects. In these, the auditory meatus is usually patent and defects of the ossicular chain of the middle ear are infrequent. However, these defects are sometimes designated as Type I Microtia. |
| | Isolated absence, atresia, stenosis or malformation of the ear canal with a normal external ear. |
| | Congenital absence of the ear not diagnosed as anotia or microtia. |
| **ICD-9-CM Codes** | 744.01, 744.23 |
| **CDC/BPA Codes** | 744.01, 744.21 |
| **Diagnostic Methods** | Anotia and microtia are usually easily recognized on physical examination after delivery. However, abnormalities of the middle and inner ear may be conclusively diagnosed only by CT or MRI scan, surgery, or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | While these conditions may be identified by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. In addition, the absence of anotia or microtia on prenatal ultrasound does not necessarily mean that they will not be diagnosed after delivery. |

**Additional Information:**

The spectrum of severity of microtia may range from a measurably small external ear with minimal structural abnormality to major structural alteration of the external ear with an absent or blind-ending canal. Following is the classification system of Meurman (modified from Marks):

Type I в – Generally small ears that retain most of the overall structure of the normal auricle. These should not be coded as microtia.

Type II в – A moderately severe anomaly with a longitudinal mass of cartilage with some resemblance to a pinna. The rudimentary auricle may be hook-shaped, have an S-shape, or the appearance of a question mark.

Type III в – The ear is a rudiment of soft tissue and the auricle has no resemblance to a normal pinna.

Type IV в – Complete absence of all external ear structures (anotia).

Abnormalities that may be associated with anotia/microtia include anomalies of the middle and/or inner ear, the mandible and face, and hearing loss.

Anotia/microtia may be a component of Goldenhar and other syndromes.

## Common Truncus (Truncus Arteriosus or TA)

| | |
|---|---|
| **Description** | Failure of separation of the aorta and the pulmonary artery, resulting in a single common arterial trunk carrying blood from the heart to both the body and lungs. |
| **Inclusions** | Common truncus<br>Truncus arteriosus (TA) |
| **Exclusions** | Aorto-pulmonary window |
| **ICD-9-CM Codes** | 745.0 |
| **CDC/BPA Codes** | 745.00 – 745.01 |
| **Diagnostic Methods** | While truncus defects may be suspected by clinical presentation, they may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | These conditions may be included as cases when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally. |

**Additional Information:**

A ventricular septal defect is often present in association with truncus defects and should be coded separately.

Truncus arteriosus is one of several abnormalities of the outflow tract of the heart known as conotruncal defects. Some infants with these defects have a deletion on the short arm of chromosome 22 (22q11 deletion). This deletion is diagnosed using fluorescent *in situ* hybridization (FISH) and will not necessarily be detected on a routine karyotype analysis.

## Transposition of the Great Arteries (TGA)

| | |
|---|---|
| **Description** | Transposition of the aorta and the pulmonary artery such that the aorta arises from the right ventricle (instead of the left) and the pulmonary artery arises from the left ventricle (instead of the right). |
| **Inclusions** | Complete transposition (d-TGA without a VSD) <br> Corrected transposition (l-TGA) <br> Incomplete transposition (d-TGA with a VSD) <br> Transposition of the Great Arteries (TGA), not otherwise specified <br> Transposition of the Great Vessels (TGV) |
| **Exclusions** | NA |
| **ICD-9-CM Codes** | 745.10, 745.11, 745.12, 745.19 |
| **CDC/BPA Codes** | 745.10 – 745.19 |
| **Diagnostic Methods** | While transposition defects may be suspected by clinical presentation, they may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | These conditions may be included as cases when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally. |

**Additional Information:**

In order for a child with d-TGA to survive, a communication must be present between the pulmonary and systemic circulations to allow oxygenated blood from the lungs to reach the right ventricle for distribution to the rest of the body through the abnormally placed aorta. In most instances, this communication is through a ventricular septal defect (incomplete TGA). If a VSD is not present, oxygenated blood from the lungs is returned directly to the lungs without being distributed to the rest of the body (complete TGA).

If the defect coding system does not include unique codes to differentiate TGA with and without a VSD (complete vs. incomplete), the VSD should be coded separately when present.

l-TGA (corrected transposition) is a defect in which the ventricle on the right side of the heart has the anatomic appearance of the left ventricle, and the ventricle on the left side of the heart has the anatomic appearance of the right ventricle (ventricular inversion). The pulmonary artery arises from the anatomic left ventricle and the aorta arises from the anatomic right ventricle (hence the designation of transposition). Because blood from the ventricle on the right flows through the pulmonary artery, and that from the ventricle on the left flows through the aorta, circulation is

normal as long as there are no other defects.

Transposition of the great arteries is one of several abnormalities of the outflow tract of the heart known as conotruncal defects. Some infants with these defects have a deletion on the short arm of chromosome 22 (22q11 deletion). This deletion is diagnosed using fluorescent *in situ* hybridization (FISH) and will not necessarily be detected on a routine karyotype analysis.

## Tetralogy of Fallot

| | |
|---|---|
| **Description** | The simultaneous presence of a ventricular septal defect (VSD), pulmonic stenosis, a malpositioned aorta that overrides the ventricular septum, and right ventricular hypertrophy. |
| **Inclusions** | Pentalogy of Fallot – Tetralogy of Fallot with an associated inter-atrial communication, either a patent foramen ovale (PFO) or an atrial septal defect (ASD). Tetralogy of Fallot Tet TOF |
| | Some coding systems may also include Trilogy of Fallot, or Fallot's Triad – the simultaneous presence of an atrial septal defect, pulmonic stenosis, and right ventricular hypertrophy. |
| **Exclusions** | Simultaneous occurrence of a VSD and pulmonary stenosis that has TOF physiology but has not been diagnosed as Tetralogy of Fallot. |
| **ICD-9-CM Codes** | 745.2 |
| **CDC/BPA Codes** | 745.20 – 745.21, 746.84 |
| **Diagnostic Methods** | While Tetralogy of Fallot may be suspected by clinical presentation, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | These conditions may be included as cases when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally. |

**Additional Information:**

Children with Tetralogy of Fallot may experience episodes of cyanosis or hypoxia that result from shunting of unoxygenated blood across the VSD from the right to the left ventricle. Children who have a coexisting VSD and pulmonary stenosis, but do not have Tetralogy of Fallot, may experience similar episodes. Thus, the occurrence of cyanosis or hypoxia does not necessarily mean a child has been diagnosed with Tetralogy of Fallot.

Tetralogy of Fallot is one of several abnormalities of the outflow tract of the heart known as conotruncal defects. Some infants with these defects have a deletion on the short arm of chromosome 22 (22q11 deletion). This deletion is diagnosed using fluorescent *in situ* hybridization (FISH) and will not necessarily be detected on a routine karyotype analysis.

## Ventricular Septal Defect (VSD)

| | |
|---|---|
| **Description** | An opening in the septum that separates the left and right ventricles of the heart. |
| **Inclusions** | Ventricular septal defect<br>VSD |
| **Exclusions** | Ventricular septal defects that occur as part of Tetralogy of Fallot or an endocardial cushion defect. Inflow-type, subtricuspid, and canal-type VSDs are assumed to be part of an endocardial cushion defect and should not be coded separately . |
| **ICD-9-CM Codes** | 745.4 |
| **CDC/BPA Codes** | 745.40 – 745.59, excluding 745.498 |
| **Diagnostic Methods** | Some isolated VSDs may be diagnosed on physical examination and/or EKG without direct imaging of the heart. However, many VSDs may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | While VSDs may be identified by prenatal ultrasound, many close spontaneously before delivery. For this reason, VSDs that are diagnosed prenatally should not be included unless they have been confirmed postnatally. In addition, the absence of a VSD on prenatal ultrasound does not necessarily mean that a VSD will not be diagnosed after delivery, as it is not always possible to accurately visualize the entire ventricular septum by prenatal ultrasound. |

**Additional Information:**

VSDs may be of several types, depending on the location of the opening along the ventricular septum. The most common are:

    Muscular
    Membranous
    Perimembranous

However, in many instances the type of VSD may not be specified in the medical record.

Many muscular, membranous and perimembranous VSDs may close spontaneously in the first weeks or months of life without treatment.

An aneurysm of the ventricular septum indicates a membranous or perimembranous VSD that is in the process of closing.

## Atrial Septal Defect (ASD)

| | |
|---|---|
| **Description** | An opening in the septum that separates the left and right atria of the heart. |
| **Inclusions** | Atrial septal defect, type not specified<br>ASD<br>Secundum ASD (ASD 2 or ASD II)<br><br>ASD vs. PFO – In the first days of life, it may not be possible to distinguish whether the opening in the atrial septum is a true ASD or a patent foramen ovale that has not yet closed (see below). ASD vs. PFO should be included only if the exact nature of the condition was never resolved. |
| **Exclusions** | Atrioventricular septal defects (AVSD) – These are included under endocardial cushion defects (see below).<br><br>Patent foramen ovale (PFO) – A PFO is normal *in utero* and frequently does not close until 24 to 48 hours after birth.<br><br>Primum ASD (1° ASD) – These are included under endocardial cushion defects (see below). |
| **ICD-9-CM Codes** | 745.5 |
| **CDC/BPA Codes** | 745.50 – 745.59, excluding 745.50 |
| **Diagnostic Methods** | Some isolated ASDs may be diagnosed based on physical examination and/or EKG without direct imaging of the heart. However, many ASDs may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | While ASDs may be identified by prenatal ultrasound, they may close spontaneously before delivery. For this reason, ASDs that are diagnosed prenatally should not be included unless they have been confirmed postnatally. In addition, the absence of an ASD on prenatal ultrasound does not necessarily mean that an ASD will not be diagnosed after delivery, as it is not always possible to accurately visualize the entire atrial septum by prenatal ultrasound. |

**Additional Information:**

Secundum ASDs are usually located toward the middle of the atrial septum. Some close spontaneously without treatment.

Primum ASDs are located in the lower portion of the atrial septum, are etiologically related to endocardial cushion (AV canal) defects, and never close spontaneously.

## Endocardial Cushion Defect

| | |
|---|---|
| **Description** | A defect in both the lower portion of the atrial septum and the upper portion of the ventricular septum, producing a large opening (canal) in the central part of the heart. The adjacent parts of the mitral and tricuspid valves may also be abnormal, resulting in a single common atrioventricular valve. In extreme cases, virtually the entire atrial and ventricular septae may be missing. |

**Inclusions**

Atrioventricular septal defect (AVSD)
Common or complete atrioventricular (AV) canal
Common atrioventricular (AV) orifices
Endocardial cushion defect

Primum atrial septal defect (1° ASD) – A defect only in the lower portion of the atrial septum. While this does not also involve a defect in the upper portion of the ventricular septum, it is etiologically related to the more complete form. A cleft mitral valve is often present.

Common atrium – A very large primum ASD.
Incomplete AV canal (incomplete endocardial cushion defect) – Same as a primum ASD.

Inflow-type, subtricuspid, or canal-type ventricular septal defect (VSDAVC) – A defect in the upper (inflow) portion of the ventricular septum. While this does not also involve a defect in the lower portion of the atrial septum, it is etiologically related to the more complete form.

**Exclusions**

Secundum ASDs that coexist with a VSD. In this instance, both the ASD and the VSD should be coded.

**ICD-9-CM Codes**

745.60, 745.61, 745.69

**CDC/BPA Codes**

745.60 – 745.69

**Diagnostic Methods**

While endocardial cushion defects may be suspected by clinical presentation, examination, and EKG changes, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.

**Prenatal Diagnoses Not Confirmed Postnatally**

These conditions may be included as cases when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data, as it may be difficult to

distinguish this condition from other abnormalities of the cardiac septae prenatally. Live-born children who survive should always have confirmation of the defect postnatally.

**Additional Information:**
Endocardial cushion defects are known to be associated with Down syndrome. Approximately 20% of children with Down syndrome have some type of endocardial cushion defect. Conversely, approximately 70% of children with an endocardial cushion defect have Down syndrome.

## Pulmonary Valve Atresia and Stenosis

| | |
|---|---|
| **Description** | Pulmonary valve atresia – Lack of patency, or failure of formation altogether, of the pulmonary valve, resulting in obstruction of blood flow from the right ventricle to the pulmonary artery. |
| | Pulmonary valve stenosis – Obstruction or narrowing of the pulmonary valve, which may impair blood flow from the right ventricle to the pulmonary artery. |
| **Inclusions** | Pulmonary valve atresia<br>Pulmonary valve stenosis<br>Pulmonic stenosis (PS) |
| **Exclusions** | Atresia or stenosis of the main or branch (right or left) pulmonary arteries, not involving the pulmonary valve.<br>Pulmonary stenosis that occurs as part of Tetralogy or Pentalogy of Fallot.<br>Supra-valvular or sub-valvular pulmonic stenosis. |
| **ICD-9-CM Codes** | 746.01,  746.02 |
| **CDC/BPA Codes** | 746.00 – 746.01 |
| **Diagnostic Methods** | While pulmonary valve atresia or stenosis may be suspected by clinical presentation, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | While these conditions may be identified by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. In addition, the absence of pulmonary valve atresia or stenosis on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery. |

**Additional Information:**

Pulmonary valve atresia or stenosis may occur with or without a coexisting ventricular septal defect. When it occurs with a VSD, the child may experience episodes of cyanosis or hypoxia similar to those seen in children with Tetralogy of Fallot. This results from shunting of unoxygenated blood across the VSD from the right to the left ventricle. Thus, the occurrence of cyanosis or hypoxia does not necessarily mean that the child has Tetralogy of Fallot.

## Tricuspid Valve Atresia and Stenosis

| | |
|---|---|
| **Description** | Tricuspid valve atresia – Lack of patency, or failure of formation altogether, of the tricuspid valve, resulting in obstruction of blood flow from the right atrium to the right ventricle.<br><br>Tricuspid valve stenosis – Obstruction or narrowing of the tricuspid valve, which may impair blood flow from the right atrium to the right ventricle. |
| **Inclusions** | Tricuspid atresia<br>Tricuspid stenosis |
| **Exclusions** | Tricuspid regurgitation without specific mention of tricuspid atresia or stenosis. |
| **ICD-9-CM Codes** | 746.1 |
| **CDC/BPA Codes** | 746.10 (excluding 746.105) |
| **Diagnostic Methods** | While tricuspid valve atresia or stenosis may be suspected by clinical presentation, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | While these conditions may be identified by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. In addition, the absence of tricuspid valve atresia or stenosis on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery. |
| **Additional Information** | NA |

## Ebstein's Anomaly

| | |
|---|---|
| **Description** | Downward displacement if the tricuspid valve into the right ventricle. The tricuspid valve is usually hypoplastic and regurgitant. |
| **Inclusions** | Ebstein's anomaly<br>Ebstein malformation |
| **Exclusions** | NA |
| **ICD-9-CM Codes** | 746.2 |
| **CDC/BPA Codes** | 746.20 |
| **Diagnostic Methods** | While Ebstein's anomaly may be suspected by clinical presentation, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | While this condition may be identified by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of Ebstein's anomaly on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery. |

**Additional Information:**

Ebstein's anomaly has been associated with lithium exposure during gestation. However, the magnitude of this association is probably very small.

## Aortic Valve Stenosis

| | |
|---|---|
| **Description** | Obstruction or narrowing of the aortic valve, which may impair blood flow from the left ventricle to the aorta. |
| **Inclusions** | Stenosis of the aortic valve |
| **Exclusions** | Stenosis of the aorta without mention of the aortic valve. Supra-valvular or sub-valvular aortic stenosis. |
| **ICD-9-CM Codes** | 746.3 |
| **CDC/BPA Codes** | 746.30 |
| **Diagnostic Methods** | While aortic valve stenosis may be suspected by clinical presentation, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | While this condition may be identified by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of aortic valve stenosis on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery. |
| **Additional Information** | NA |

## Hypoplastic Left Heart Syndrome (HLHS)

| | |
|---|---|
| **Description** | A condition in which the structures on the left side of the heart and the aorta are extremely small. Classically, this condition includes hypoplasia of the left ventricle, atresia or severe hypoplasia of the mitral and aortic valves, and hypoplasia and coarctation of the aorta. |
| **Inclusions** | Any diagnosis of hypoplastic left heart syndrome, regardless of whether all conditions in the classical definition are present. |
| **Exclusions** | Hypoplasia or diminished size of the left ventricle alone without involvement of other structures on the left side of the heart or the aorta.<br><br>Hypoplastic left heart or small left ventricle that occurs as part of another complex heart defect, such as an endocardial cushion defect (AV canal). |
| **ICD-9-CM Codes** | 746.7 |
| **CDC/BPA Codes** | 746.70 |
| **Diagnostic Methods** | While hypoplastic left heart may be suspected by clinical presentation, examination, and EKG changes, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | These conditions may be included as cases when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data, as it may be difficult to distinguish this condition from other abnormalities of the left ventricle prenatally. Live-born children who survive should always have confirmation of the defect postnatally before being included. |
| **Additional Information** | NA |

## Patent Ductus Arteriosus (PDA)

| | |
|---|---|
| **Description** | Abnormally persistent blood flow through the ductus arteriosus beyond the first few days of life. |
| **Inclusions** | Patent ductus arteriosus in infants with birth weight ≥ 2,500 grams who have not been given prostaglandin (see below). |
| **Exclusions** | Patent ductus arteriosus in infants with birth weight < 2,500 grams. Patent ductus arteriosus in infants ≥ 2,500 grams who have been given prostaglandin (see below). |
| **ICD-9-CM Codes** | 747.0 |
| **CDC/BPA Codes** | 747.00 |
| **Diagnostic Methods** | Some instances of patent ductus arteriosus may be diagnosed on physical examination. However, many PDAs may be diagnosed conclusively only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | Because a patent ductus arteriosus is normal and necessary during fetal life, this condition should not be included in surveillance data unless diagnosed postnatally at an appropriate age. |

**Additional Information:**

In the normal fetal circulation, blood flows from the right ventricle to the pulmonary artery, then crosses the ductus arteriosus to the aorta for distribution to the body and the placenta. This bypasses much of the pulmonary circulation, since fetal blood is oxygenated by the placenta and not the lungs. Over the first hours after a normal full-term birth, smooth muscle in the wall of the ductus contracts and thickens to prevent blood flow through the ductus. Over the subsequent 2 to 3 weeks of life, the ductus is replaced by fibrous tissue and the communication is permanently sealed. Persistence of a patent ductus through which blood may flow beyond that time is abnormal.

In preterm infants, the ability of the ductus to constrict and close after delivery is not fully developed. Patent ductus arteriosus in a preterm infant is more likely to be a consequence of prematurity rather than an inherent abnormality. In these infants, it should not be coded as a defect.

The length of time required for the ductus to close is somewhat variable among term infants, and there is disagreement among specialists about the length of time after which patency is abnormal. Some birth defects surveillance programs only include PDAs that have been present for at least 6 weeks after birth.

Term infants who have additional heart defects may have abnormal patterns of blood flow or abnormal pressures in the pulmonary artery and aorta which prevent the ductus from closing. In these instances, the PDA is not an inherent abnormality but secondary to the additional defects.

In some severe heart defects, such as pulmonary atresia or d-TGA without a VSD, the infant's initial survival may depend on the presence of a patent ductus arteriosus in order for blood to reach the lungs for oxygenation. Prostaglandin (PGE) may be administered intravenously to maintain the patency of the ductus. In these instances, the PDA is an artifact of treatment of the underlying condition and should not be coded as a defect.

Patent ductus arteriosus may be a component of persistent transitional (fetal) circulation, in which the fetal pattern of blood flow through the ductus and bypassing the lungs, persists after birth. This is often a physiologic response to hypoxia from respiratory suppression, as may be seen with meconium aspiration.

## Coarctation of the Aorta

| | |
|---|---|
| **Description** | Narrowing of the descending aorta, which may obstruct blood flow from the heart to the rest of the body. The most common site of coarctation occurs distal to the origin of the left subclavian artery in the region of the ductus arteriosus. |
| **Inclusions** | Coarctation of the aorta, type not specified<br>Preductal, juxtaductal, and postductal coarctations – These terms refer to the exact placement of the segment of coarctation relative to the insertion of the ductus arteriosus. |
| **Exclusions** | NA |
| **ICD-9-CM Codes** | 747.10 |
| **CDC/BPA Codes** | 747.10 – 747.19 |
| **Diagnostic Methods** | While coarctation of the aorta may be suspected by clinical presentation and examination, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | While this condition may be identified by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of coarctation of the aorta on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery. |

**Additional Information:**

Left-sided obstructive lesions of the heart, such as coarctation, have been associated with Turner syndrome (karyotype 45,X and other variants).

## Cleft Palate without Cleft Lip

| | |
|---|---|
| **Description** | An opening in the roof of the mouth resulting from incomplete fusion of the shelves of the palate. The opening may involve the hard palate only, the soft palate only, or both. |
| **Inclusions** | Bifid or cleft uvula<br>Cleft palate, type not specified<br>Cleft hard palate<br>Cleft soft palate<br>Submucous cleft palate – A cleft in the soft palate that is covered by the mucosa or a thin muscle layer. |
| **Exclusions** | Cleft palate that coexists with a cleft lip. These should be coded as cleft lip only (see below). |
| **ICD-9-CM Codes** | 749.0 |
| **CDC/BPA Codes** | 749.00 – 749.09 |
| **Diagnostic Methods** | Cleft palate is usually easily recognized on physical examination by direct visualization of the pharynx after delivery. It may also be seen on CT or MRI scan, at surgery or autopsy. However, submucous cleft palate may be difficult to diagnose by physical examination during the first year of life. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | While this condition may be identified by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of cleft palate on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery. |

**Additional Information:**
Cleft palate may be unilateral, bilateral, or central in location. If the defect coding system includes unique codes for these different types, the location of the cleft should be coded.

Cleft palate sometimes may be described as U-shaped or V-shaped. This distinction is not clinically meaningful and these conditions should not be coded differently.

Bifid uvula is often seen in association with a submucous cleft palate. However, bifid uvula also may occur alone. The presence of submucous cleft palate does not necessarily mean that a bifid uvula is present.

Cleft palate is one component of the Pierre Robin sequence, which also includes micrognathia and glossoptosis (when the tongue falls backward into the posterior pharynx). When diagnosed, Pierre Robin sequence should be coded separately.

## Cleft Lip with and without Cleft Palate

| | |
|---|---|
| **Description** | A defect in the upper lip resulting from incomplete fusion of the parts of the lip. |
| **Inclusions** | Complete cleft lip – The defect extends through the entire lip into the floor of the nose.<br>Incomplete cleft lip – The defect extends through part of the lip but not into the floor of the nose. |
| **Exclusions** | Pseudocleft lip – An abnormal linear thickening, depressed grove, or scar-like pigmentary change on the skin of the lip without an actual cleft.<br><br>Oblique facial clefts<br>Cleft palate without an associated cleft lip |
| **ICD-9-CM Codes** | 749.1,  749.2 |
| **CDC/BPA Codes** | 749.10 – 749.29 |
| **Diagnostic Methods** | Cleft lip is usually easily recognized on physical examination after delivery. It may also be seen on CT or MRI scan, at surgery or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | While this condition may be identified by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of cleft lip on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery. |

**Additional Information:**

Cleft lip may be unilateral, bilateral, or central in location. If the defect coding system includes unique codes for these different types, the location of the cleft should be coded.

Cleft lip may also be seen in association with amniotic bands. In this instance, the amniotic bands should also be coded.

## Choanal Atresia

| | |
|---|---|
| **Description** | Congenital obstruction of the opening of the nasal cavity into the nasopharynx on either side. This prevents communication of the nasal cavity with the pharynx. |
| **Inclusions** | Choanal atresia, type not specified<br>Choanal stenosis<br>Membranous choanal atresia, with or without a bony rim<br>Completely bony choanal atresia |
| **Exclusions** | NA |
| **ICD-9-CM Codes** | 748.0 |
| **CDC/BPA Codes** | 748.00 |
| **Diagnostic Methods** | Bilateral choanal atresia is usually easily recognized at birth from the clinical presentation of obligate mouth-breathing. Unilateral choanal atresia may be suspected by clinical examination. Both conditions may be diagnosed by the inability to pass a feeding tube from the nasal passage(s) into the posterior pharynx. Both conditions may also be seen on CT or MRI scan, at surgery or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | While these conditions may be identified by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. In addition, the absence of choanal atresia on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery. |

**Additional Information:**

Choanal atresia or stenosis may be unilateral or bilateral. If the defect coding system includes unique codes for these different types, the location should be coded.

Choanal atresia is one of the defects reported as part of the CHARGE association, which may also include colobomas, heart defects, retarded growth and development, genital hypoplasia, and ear anomalies and/or deafness.

## Esophageal Atresia/Tracheoesophageal Fistula

| | |
|---|---|
| **Description** | Esophageal atresia – A condition in which the esophagus ends in a blind pouch and fails to connect with the stomach. |
| | Tracheoesophageal fistula – An abnormal communication between the esophagus and the trachea. This is almost always associated with some form of esophageal atresia. |
| **Inclusions** | Esophageal atresia alone |
| | Esophageal atresia with tracheoesohpageal (TE) fistula |
| | Esophageal stenosis, stricture, ring, or web |
| | TE fistula |
| | Tracheoesophageal fistula, all types |
| **Exclusions** | Tracheal atresia |
| | Tracheoesophageal cleft |
| **ICD-9-CM Codes** | 750.3 |
| **CDC/BPA Codes** | 750.30 – 750.35 |
| **Diagnostic Methods** | The diagnosis may be suspected by the clinical presentation of polyhydramnios, vomiting, or respiratory distress. Esophageal atresia may be diagnosed by x-ray documentation of failure of a feeding tube to pass from the pharynx into the stomach. Tracheoesophageal atresia may be conclusively diagnosed only by CT or MRI scan, surgery, or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | These conditions may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally before being included. |

**Additional Information:**

In some instances**,** TE fistula without esophageal atresia may not be diagnosed until weeks, months, or even a year or more after birth if the communication between the esophagus and stomach remains patent.

TE fistula is one of the defects reported as part of the VATER, or VACTERL, association, which may also include vertebral and cardiac defects, anal atresia, renal defects, and limb anomalies.

## Rectal and Large Intestinal Atresia/Stenosis

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|---|---|
| **Description** | Complete or partial occlusion of the lumen of one or more segments of the large intestine and/or rectum. |
| **Inclusions** | Anal atresia or stenosis<br>Colonic atresia or stenosis<br>Imperforate anus<br>Large intestinal atresia or stenosis<br>Rectal atresia or stenosis |
| **Exclusions** | Apple peel intestinal atresia<br>Duodenal atresia or stenosis<br>Ileal atresia or stenosis<br>Jejunal atresia or stenosis<br>Small intestinal atresia or stenosis |
| **ICD-9-CM Codes** | 751.2 |
| **CDC/BPA Codes** | 751.20 – 751.24 |
| **Diagnostic Methods** | Anal atresia (imperforate anus) is usually easily recognized at birth by physical examination. While large intestinal and rectal atresia or stenosis may be suspected by the clinical presentation of failure to pass meconium or stool, they may be conclusively diagnosed only through direct imaging of the bowel by x-ray, barium enema, surgery, or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | While these conditions may be identified by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. In addition, the absence of intestinal, rectal or anal atresia or stenosis on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery. |

**Additional Information:**

These conditions may occur with or without a fistula.

Anal atresia is one of the defects reported as part of the VATER, or VACTERL, association, which may also include vertebral and cardiac defects, TE fistula, renal defects, and limb anomalies.

## Pyloric Stenosis

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|---|---|
| **Description** | Hypertrophy (thickening) of the muscles of the pylorus connecting the stomach to the duodenum, resulting in complete or partial obstruction of the passage of food and gastric contents. |
| **Inclusions** | Infantile (congenital) hypertrophic pyloric stenosis<br>Pyloric stenosis |
| **Exclusions** | Pylorospasm (intermittent spasm of the pyloric muscles) without permanent narrowing of the lumen. |
| **ICD-9-CM Codes** | 750.5 |
| **CDC/BPA Codes** | 750.51 |
| **Diagnostic Methods** | Many instances of pyloric stenosis may be diagnosed by the clinical presentation and physical examination. However, other cases may be diagnosed conclusively only by abdominal ultrasound or contrast x-ray of the stomach. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | In rare cases, pyloric stenosis may develop prenatally and may be identified on prenatal ultrasound. However, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of pyloric stenosis on prenatal ultrasound does not mean that it will not develop after delivery (see below). |

**Additional Information:**

Pyloric stenosis most typically presents with intractable vomiting in a 3- to 4-week-old infant. While it may appear late in gestation, it develops more commonly in the first month or two after birth. As such, it may not be a truly congenital defect.

The etiology of pyloric stenosis remains unclear, but is probably multifactorial with both genetic and environmental influences. Pyloric stenosis has been associated with erythromycin use in newborn infants.

## Hirschsprung Disease (Congenital Megacolon)

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|---|---|
| **Description** | Hirschsprung disease – Absence of the parasympathetic ganglion nerve cells (aganglionosis) of the wall of the colon or rectum, which may result in congenital megacolon.

Megacolon – Enlargement of the diameter of part or all of the colon. |
| **Inclusions** | Aganglionic megacolon
Congenital megacolon
Hirschsprung disease, type not specified
Long-segment Hirschsprung disease (Type II)
Short-segment Hirschsprung disease (Type I)
Total colon (intestinal) aganglionosis |
| **Exclusions** | Psychogenic megacolon |
| **ICD-9-CM Codes** | 751.3 |
| **CDC/BPA Codes** | 751.30 – 751.34 |
| **Diagnostic Methods** | Hirschsprung disease (congenital megacolon) may be suspected by contrast x-ray (barium enema). However, it may be diagnosed conclusively only through direct assessment of the presence or absence of ganglion cells in rectal tissue at biopsy, surgery, or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | While this condition may be suspected by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of congenital megacolon on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery. |

**Additional Information:**

Megacolon may result from any condition that inhibits normal passage of the intestinal contents. Primary underlying conditions include Hirschsprung disease, rectal and large intestinal atresia/stenosis, imperforate anus, and voluntary stool retention (psychogenic megacolon). In Hirschsprung disease, the aganglionic segment of intestine is small and empty, while the normally enervated segment proximal to the affected area is enlarged and filled with fecal matter.

Hirschsprung disease is classified according to the extent of aganglionosis. In 80% of cases, aganglionosis extends from the anal sphincter and rectum to the middle of the sigmoid colon; in 10% to 20% of cases, it extends further to the transverse or right colon; in 3% of cases, aganglionosis involves the entire colon.

Possible complications of Hirschsprung disease/congenital megacolon include bowel perforation, enterocolitis (intestinal inflammation), peritonitis (inflammation of the lining of the abdomen), and septicemia (bloodstream infection).

Approximately 3% of infants with Down syndrome have aganglionosis of the colon. When Down syndrome and Hirschsprung disease/congenital megacolon occur in the same infant, both conditions should be coded.

## Biliary Atresia

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|---|---|
| **Description** | Congenital absence of the lumen of the extrahepatic bile ducts. |
| **Inclusions** | Agenesis, absence, hypoplasia, obstruction or stricture of the bile duct(s) |
| **Exclusions** | Congenital or neonatal hepatitis<br>Intrahepatic biliary atresia (absence or paucity of bile ducts within the liver) not associated with extrahepatic biliary atresia |
| **ICD-9-CM Codes** | 751.61 |
| **CDC/BPA Codes** | 751.65 |
| **Diagnostic Methods** | Biliary atresia may be suspected by the clinical presentation and the presence of elevated direct bilirubin and liver function tests. However, it may be conclusively diagnosed only through direct assessment of the bile ducts by abdominal ultrasound, CT or MRI scan, biliary excretion study (HIDA scan), surgery, or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | While biliary atresia may be suspected by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of biliary atresia on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery. |

**Additional Information:**

The liver contains within its substance intrahepatic bile ducts and passages that join and coalesce to form two main ducts that carry bile out of the liver.

The extrahepatic bile ducts include the hepatic duct (formed by the two main ducts that carry bile out of the liver), the cystic duct (which carries bile out of the gallbladder where it is stored), and the common bile duct (formed by the junction of the hepatic duct and the cystic duct), which carries bile into the duodenum for excretion.

When extrahepatic biliary atresia is present, the intrahepatic bile ducts may also be abnormal or atretic.

Patients with biliary atresia may have jaundice due to direct hyperbilirubinemia, which is not treated with phototherapy. The more common type of neonatal jaundice due to indirect hyperbilirubinemia may be treated with phototherapy and does not indicate the presence of biliary atresia.

## Renal Agenesis/Hypoplasia

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|---|---|
| **Description** | Renal agenesis – Complete absence of the kidney<br>Renal hypoplasia – Incomplete development of the kidney |
| **Inclusions** | Renal agenesis, dysgenesis, aplasia, or hypoplasia<br>Potter syndrome secondary to renal agenesis/hypoplasia |
| **Exclusions** | Cystic renal dysplasia<br>Cystic kidney disease<br>Multicystic kidney<br>Multicystic dysplastic kidney<br>Polycystic kidney<br>Renal cysts<br>Renal dysplasia<br>Small kidney |
| **ICD-9-CM Codes** | 753.0 |
| **CDC/BPA Codes** | 753.00 – 753.01 |
| **Diagnostic Methods** | Bilateral renal agenesis is usually easily recognized on physical examination after delivery. Bilateral renal hypoplasia may or may not be recognized after delivery, depending on the severity and degree of residual kidney function.<br><br>Unilateral renal agenesis or hypoplasia may not be symptomatic at delivery if the contralateral kidney is not impaired.<br><br>Each of these diagnoses may be conclusively diagnosed only through direct assessment by abdominal ultrasound, CT or MRI scan, surgery, or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | Bilateral renal agenesis may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally before being included.<br><br>While bilateral renal hypoplasia and unilateral renal agenesis/hypoplasia may be suspected by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. Lack of visualization of a kidney on prenatal ultrasound does not always indicate that the kidney is truly absent. |

**Additional Information:**
Renal agenesis and hypoplasia may be unilateral or bilateral. If the defect coding system includes unique codes for these different types, the location should be coded.

Bilateral renal agenesis, or any condition that significantly impairs the function of both kidneys *in utero*, may lead to the oligohydramnios sequence (Potter syndrome) due to lack of fetal urine production and the resulting decreased amniotic fluid volume. The sequence includes minor facial dysmorphism (flat face, small chin, large ears), pulmonary hypoplasia, and joint contractures.

Bilateral renal agenesis is incompatible with long-term survival unless a kidney transplant is performed. In contrast, unilateral renal agenesis/hypoplasia may not be diagnosed until weeks, months, or even years after birth if the contralateral kidney function is normal. Some unilateral cases may be diagnosed only as incidental findings during evaluation for other conditions, and some may never be recognized.

## Bladder Exstrophy

| | |
|---|---|
| **Description** | A defect in the lower abdominal wall and anterior wall of the bladder through which the lining of the bladder is exposed to the outside. |
| **Inclusions** | Classic bladder exstrophy<br>Ectopia vesicae<br>Epispadias-exstrophy complex<br>Extroversion of the bladder<br>Variants of bladder exstrophy<br>Vesical exstrophy |
| **Exclusions** | Ambiguous genitalia without mention of bladder exstrophy<br>Cloacal exstrophy<br>Isolated epispadias |
| **ICD-9-CM Codes** | 753.5 |
| **CDC/BPA Codes** | 753.50 |
| **Diagnostic Methods** | Bladder exstrophy is easily recognized on physical examination at delivery. However, the exact nature of the defect and associated anomalies may only be distinguished by abdominal ultrasound, contrast x-ray studies, CT or MRI scan, surgery, or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | These conditions may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data, as it may be difficult to distinguish bladder exstrophy from cloacal exstrophy. Live-born children who survive should always have confirmation of the defect postnatally before being included. |

**Additional Information:**

In the classic form of bladder exstrophy, the entire urinary tract is open anteriorly from the urethral meatus to the umbilicus. The pubic bones are widely separated, as are the abdominal muscles and fascia. There is eversion/exposure of the posterior bladder wall. The genitalia of either gender may be involved and may be bifid or duplicated. The classic form of bladder exstrophy occurs more frequently in males.

Variants of bladder exstrophy occur more rarely and affect females more often then males. Included among these variants are superior vesical fistula, closed exstrophy, duplicate exstrophy, pseudoexstrophy, inferior vesicle. Epispadias is almost uniformly present, but should not be coded separately.

Ambiguous genitalia may be noted in patients with bladder exstrophy if an obvious scrotum and

testes are not present. However, ambiguous genitalia should not be coded as a separate defect in these instances.

Bladder exstrophy should be distinguished from cloacal exstrophy, in which the urinary, intestinal, and genital structures open into a common cavity (the cloaca). The distinction may only be possible with detailed diagnostic studies, surgery, or at autopsy. In cloacal exstrophy, bladder exstrophy and imperforate anus are also present. In bladder exstrophy without cloacal exstrophy, the anus is patent. When both bladder and cloacal exstrophy are present, only cloacal exstrophy should be coded.

## Obstructive Genitourinary Defect

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|---|---|
| **Description** | Partial or complete obstruction of the flow of urine at any level of the genitourinary tract from the kidney to the urethra. |
| **Inclusions** | Atresia, stenosis, stricture or occlusion of one or both ureters, the bladder neck, the urethra or urethral meatus<br>Dilatation of one or both ureters<br>Hydronephrosis<br>Hydroureter<br>Hypoplastic ureter<br>Megaloureter<br>Posterior urethral valves<br>Obstruction of the ureteropelvic junction (UPJ), the ureterovesical (UV) junction, or the vesicourethral (VU) junction<br>Urethral valves, type not specified |
| **Exclusions** | Inhibition of urinary flow at any of the above sites resulting solely from neurologic impairment. |
| **ICD-9-CM Codes** | 753.2,  753.6 |
| **CDC/BPA Codes** | 753.20 – 753.29,  753.60 – 753.69 |
| **Diagnostic Methods** | Genitourinary tract obstruction may be suspected by the clinical presentation. However, the exact nature of the defect and the level of obstruction may only be distinguished by direct visualization. The upper urinary tract (kidneys and ureters) is usually visualized with renal ultrasound, radionuclide scan, or a contrast study such as intravenous pyelography (IVP). The lower urinary tract (bladder and urethra) is usually visualized directly with cystoscopy or urethral endoscopy, or with contrast studies such as voiding cystourethrogram (VCUG) and sometimes IVP. Obstructions also may be diagnosed at surgery or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | While obstructive genitourinary defects may be identified by prenatal ultrasound, many lesions diminish or resolve spontaneously prior to birth. For this reason, they should not be included in surveillance data without postnatal confirmation (see below). In addition, the absence of genitourinary obstruction on prenatal ultrasound does not necessarily mean that an obstructive defect will not be diagnosed after delivery. |

**Additional Information:**

When urine flow is obstructed, the portion of the genitourinary tract proximal to the affected area may become enlarged and dilated with urine. Mild lesions may produce only partial or intermittent urinary obstruction without permanent damage. More severe lesions may substantially or

completely obstruct urine flow, resulting in permanent damage to proximal structures, and sometimes impaired kidney function, if not relieved by surgery.

Increased use of ultrasound screening has led to the recognition of asymptomatic genitourinary tract obstructions in the fetus and newborn, many of which resolve without treatment and would not otherwise have been diagnosed. Inclusion of these lesions in birth defects surveillance data may inflate the apparent frequency of significant obstructive genitourinary defects. If it is possible to correlate the findings on prenatal and/or newborn ultrasound with the clinical course of symptoms and treatment, this should factor into the decision as to which obstructive lesions to include in the surveillance data.

## Hypospadias and Epispadias

| | |
|---|---|
| **Description** | Hypospadias – Displacement of the opening of the urethra (urethral meatus) ventrally and proximally (underneath and closer to the body) in relation to the tip of the glans of the penis. |
| | Epispadias – Displacement of the opening of the urethra (urethral meatus) dorsally and proximally (on the top and closer to the body) in relation to the tip of the glans of the penis. |
| **Inclusions** | First-degree hypospadias – The urethral meatus is located on the glans of the penis. Also called primary, 1°, glandular, or coronal hypospadias. |
| | Second-degree hypospadias – The urethral meatus is located on the shaft of the penis. Also called secondary, 2°, or penile hypospadias. |
| | Third-degree hypospadias – The urethral meatus is located at the base of the penis on the scrotum or perineum. Also called tertiary, 3°, scrotal, penoscrotal, or perineal hypospadias. |
| | Hypospadias, degree not specified<br>Hypospadias of any type with chordee<br>Epispadias |
| **Exclusions** | Chordee alone without associated hypospadias<br>Ambiguous genitalia |
| **ICD-9-CM Codes** | Hypospadias  752.61<br>Epispadias  752.62 |
| **CDC/BPA Codes** | Hypospadias  752.600 – 752.607,  752.620,  752.605 – 752.607<br>Epispadias  752.621 |
| **Diagnostic Methods** | Both hypospadias and epispadias are usually easily recognized on physical examination at delivery. They may also be seen on contrast x-rays of the urinary tract, at surgery or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | While these conditions may be diagnosed by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. In addition, the absence of hypospadias or epispadias on prenatal ultrasound does not necessarily mean that they will not be diagnosed after delivery. |

**Additional Information:**

Chordee indicates a ventral (downward) curve of the penis, which may result from cutaneous or

fibrous restriction. It is present in approximately 35% to 50% of cases of hypospadias.

In mild forms of first-degree hypospadias, the foreskin may appear hooded but there may be no overt clinical symptoms.

In contrast, third-degree hypospadias may be described as ambiguous genitalia. In this instance, it is important to search the medical record for detailed information (including chromosome, molecular, and hormone analyses; genetics and endocrinology consultations; surgery or autopsy reports) that may clarify the anatomy and/or indicate whether an underlying genetic condition or endocrinopathy associated with ambiguous genitalia is present. Ambiguous genitalia should not be coded if hypospadias is the only diagnosis. Hypospadias generally should not be coded if a normal female karyotype (46,XX) is reported.

Epispadias is almost uniformly present with bladder exstrophy. In these cases, only the bladder exstrophy should be coded.

## Reduction Deformity, Upper Limbs

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|---|---|
| **Description** | Complete or partial absence of the upper arm (humerus), lower arm (radius and/or ulna), wrist (carpals), hand (metacarpals), or fingers (phalanges). |
| **Inclusions** | Transverse limb reduction – Complete or partial absence of the distal (furthest from the body) structures of the arm in a transverse (cross-wise) plane at the point where the deficiency begins. Structures proximal to the point where the deficiency begins remain essentially intact. Types of transverse limb reductions include: |

> Acheiria – Absence of a hand
> Adactyly – Absence of digits (fingers), excluding isolated missing thumb (see below)
> Aphalangia – Absence of phalanges. Fingers contain 3 phalanges each. The thumb contains 2 phalanges.
>
> Amelia – Complete absence of the upper limb (humers, radius, ulna, wrist, hand and fingers).
>
> Hemimelia, Meromelia – Partial absence of a limb. This may refer to either transverse or longitudinal reductions.
>
> Oligodactyly – Fewer than 5 digits.
>
> Transverse terminal deficiency – Complete absence of the distal structures of the arm with the proximal structures intact. This term usually refers to reduction defects below the elbow.
>
> Congenital amputation, type not specified.

Longitudinal limb reduction – Partial absence of the arm in parallel with the long axis of the arm. These may involve preaxial (on the thumb side), postaxial (on the fifth finger side), or central parts of the arm. Types of longitudinal limb reductions include:

> Ectrodactyly
> Ectromelia
> Isolated missing thumb
> Lobster claw hand
> Radial aplasia or hypoplasia
>
> Split-hand malformation (split hand/split foot malformation, SHSF) – A central longitudinal limb

reduction in which there is complete or partial absence of one or more of the central rays (second through fourth fingers and their associated metacarpal bones) of the hand.

Ulnar aplasia or hypoplasia

Intercalary limb reduction – Complete or partial absence of the proximal (closest to the body) or middle segments of the arm with all or part of the distal segment present.

Phocomelia is a general term for any type of intercalary limb reduction.

Reduction deformities of the upper limb not elsewhere coded or of unspecified type – Complete or partial absence of the arm that does not fall within the above categories or for which there is no specific description.

| | |
|---|---|
| **Exclusions** | Shortened arms, forearms, hands, or fingers that have all of their component parts, including those that are part of a generalized chondodystrophy, osteodystrophy, or dwarfism. |
| | Hypoplastic nails |
| **ICD-9-CM Codes** | 755.20 – 755.29 |
| **CDC/BPA Codes** | 755.20 – 755.29 |
| **Diagnostic Methods** | Limb reductions are usually easily recognized on physical examination at delivery. However, the exact nature of the defect may only be distinguished by x-ray, surgery, or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | While these conditions may be identified by prenatal ultrasound, they generally should not be included in surveillance data without postnatal confirmation. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Lack of visualization of a bone or limb on prenatal ultrasound does not necessarily mean that the bone or limb truly is not present. Live-born children who survive should always have confirmation of the defect postnatally before being included. |

**Additional Information:**
The terminology for limb reduction deformities is often confusing. Some terms (such as "phocomelia"), have been misused and others (such as "ectrodactyly"), have been used for both longitudinal and transverse defects. If medical record review is available, it is important to look for a complete description of all structures that are present and absent in order to verify the diagnosis.

Preaxial refers to the side of the arm on which the thumb and radius are located.
Postaxial refers to the side of the arm on which the fifth finger and ulna are located.

Transverse limb reductions may be seen in association with amniotic bands. When both are present, both conditions should be coded.

Rudimentary or nubbin fingers may be present at the distal end of a transverse limb reduction. Their presence alone does not change the classification of the defect as transverse.

Joint contractures are commonly seen in association with longitudinal limb deficiencies.

Intercalary reduction deformities (phocomelia) have been associated with the use of thalidomide during early pregnancy. However, thalidomide use may result in a number of other defects, including longitudinal reduction deformities. Intercalary defects also may occur without exposure to thalidomide.

Reduction deformities are one of the defects that may be reported as part of:

   The VATER or VACTERL association, which also may include vertebral, cardiac and renal defects, TE fistula, and anal atresia.

   Poland anomaly, which also includes deficiency of the pectoralis muscle on the same side.

   Moebius anomaly (Oromandibular-Limb Hypogenesis Spectrum), which also may include a small mouth, small chin (micrognathia), small tongue (hypoglossia), sixth and seventh cranial nerve palsies.

## Reduction Deformity, Lower Limbs

| | |
|---|---|
| **Description** | Complete or partial absence of the upper leg (femur), lower leg (tibia and/or fibula), ankle (tarsals), foot (metatarsals), or toes (phalanges). |

**Inclusions**          Transverse limb reduction – Complete or partial absence of the distal (furthest from the body) structures of the leg in a transverse (cross-wise) plane at the point where the deficiency begins. Structures proximal to the point where the deficiency begins remain essentially intact. Types of transverse limb reductions include:

> Adactyly – Absence of digits (toes)
> Aphalangia – Absence of phalanges. The smaller toes contain 3 phalanges each. The big toe contains 2 phalanges.

> Amelia – Complete absence of the lower limb (femur, tibia, fibula, ankle, foot, and toes).

> Hemimelia, Meromelia – Partial absence of a limb. This may refer to either transverse or longitudinal reductions.

> Oligodactyly – Fewer than 5 digits.

> Transverse terminal deficiency – Complete absence of the distal structures of the leg with the proximal structures intact.

> Congenital amputation, type not specified

Longitudinal limb reduction – Partial absence of the leg in parallel with the long axis of the leg. These may involve preaxial (on the big toe side), postaxial (on the fifth toe side), or central parts of the leg. Types of longitudinal limb reductions include:

> Ectrodactyly
> Ectromelia
> Fibular aplasia or hypoplasia

> Split-foot malformation (split hand/split foot malformation, SHSF) – A central longitudinal limb reduction in which there is complete or partial absence of one or more of the central rays (second through fourth toes and their associated metatarsal bones) of the foot.

Tibial aplasia or hypoplasia

Intercalary limb reduction – Complete or partial absence of the proximal (closest to the body) or middle segments of the leg with all or part of the distal segment present.

Phocomelia – A general term for any type of intercalary limb reduction.

Reduction deformities of the lower limb not elsewhere coded or of unspecified type – Complete or partial absence of the leg that does not fall within the above categories or for which there is no specific description.

**Exclusions**

Shortened upper and/or lower legs, feet, or toes that have all of their component parts, including those that are part of a generalized chondodystrophy, osteodystrophy, or dwarfism.

Hypoplastic nails.

**ICD-9-CM Codes**          755.30 – 755.39

**CDC/BPA Codes**          755.30 – 755.39

**Diagnostic Methods**

Limb reductions are usually easily recognized on physical examination at delivery. However, the exact nature of the defect may only be distinguished by x-ray, surgery, or autopsy.

**Prenatal Diagnoses Not Confirmed Postnatally**

While these conditions may be identified by prenatal ultrasound, they generally should not be included in surveillance data without postnatal confirmation. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Lack of visualization of a bone or limb on prenatal ultrasound does not necessarily mean that the bone or limb truly is not present. Live-born children who survive should always have confirmation of the defect postnatally before being included.

**Additional Information:**
The terminology for limb reduction deformities is often confusing. Some terms (such as "phocomelia") have been misused and others (such as "ectrodactyly") have been used for both longitudinal and transverse defects. If medical record review is available, it is important to look for a complete description of all structures that are present and absent in order to verify the diagnosis.

Preaxial refers to the side of the leg on which the big toe and tibia are located.
Postaxial refers to the side of the leg on which the fifth toe and fibula are located.

Transverse limb reductions may be seen in association with amniotic bands. When both are

present, both conditions should be coded.

Rudimentary or nubbin toes may be present at the distal end of a transverse limb reduction. Their presence alone does not change the classification of the defect as transverse.

Joint contractures are commonly seen in association with longitudinal limb deficiencies.

Intercalary reduction deformities (phocomelia) have been associated with the use of thalidomide during early pregnancy. However, thalidomide use may result in a number of other defects, including longitudinal reduction deformities. Intercalary defects also may occur without exposure to thalidomide.

Reduction deformities are one of the defects that may be reported as part of:

The VATER or VACTERL association, which also may include vertebral, cardiac and renal defects, TE fistula, and anal atresia.

Moebius anomaly (Oromandibular-Limb Hypogenesis Spectrum), which also may include a small mouth, small chin (micrognathia), small tongue (hypoglossia), sixth and seventh cranial nerve palsies.

## Gastroschisis

| | |
|---|---|
| **Description** | A congenital opening or fissure in the anterior abdominal wall lateral to the umbilicus through which the small intestine, part of the large intestine, and occasionally the liver and spleen, may herniate. The opening is separated from the umbilicus by a small bridge of skin, and the herniating organs are not covered by a protective membrane. Gastroschisis usually occurs on the right side of the umbilicus, although it may occur on the left. |
| **Inclusions** | Gastroschisis |
| **Exclusions** | Omphalocele |
| **ICD-9-CM Codes** | 756.79 |
| **CDC/BPA Codes** | 756.71 |
| **Diagnostic Methods** | Gastroschisis is usually easily recognized on physical examination after delivery. However, in some instances, it may be conclusively distinguished from omphalocele only at surgery or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | Gastroschisis may be included when only diagnosed prenatally. However, it may be difficult to distinguish gastroschisis from omphalocele on prenatal ultrasound, and the terms sometimes are used interchangeably. If it is possible to ascertain the degree of certainty of the prenatal diagnosis and the location of the umbilical cord insertion relative to the abdominal defect, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally before being included. In addition, the absence of gastroschisis on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery. |

**Additional Information:**

The distinction between gastroschisis and omphalocele is important because they have different etiologies and different implications for treatment and long-term survival.

In gastroschisis, the umbilicus and cord are normal and separated from the abdominal wall defect by a small bridge of skin. The herniating organs are not covered by a protective membrane. However, they may appear matted and covered by a thick fibrous material as a result of prolonged exposure to amniotic fluid *in utero*.

In omphalocele, abdominal organs herniate through the umbilicus into the umbilical cord. There is no bridge of skin between the abdominal wall defect and the umbilicus and cord. While the herniating organs are covered by a protective membrane, this may rupture before, during, or after delivery.

Gastroschisis may be one of the defects reported as part of the Limb-Body Wall complex. This is a disruption complex of the lateral body wall, which may also include limb reductions, neural tube defects, heart defects, and other anomalies.

Maternal serum alphafetoprotein (MSAFP) and/or amniotic fluid alphafetoprotein (AFAFP) may be elevated with gastroschisis. However, these screening tests alone are not sufficient to diagnose the condition.

## Omphalocele

| | |
|---|---|
| **Description** | A defect in the anterior abdominal wall in which the umbilical ring is widened, allowing herniation of abdominal organs, including the small intestine, part of the large intestine, and occasionally the liver and spleen, into the umbilical cord. The herniating organs are covered by a nearly transparent membranous sac. |
| **Inclusions** | Omphalocele |
| **Exclusions** | Gastroschisis<br>Umbilical hernia |
| **ICD-9-CM Codes** | 756.79 |
| **CDC/BPA Codes** | 756.70 |
| **Diagnostic Methods** | Omphalocele is usually easily recognized on physical examination after delivery. However, in some instances, it may be conclusively distinguished from gastroschisis only at surgery or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | Omphalocele may be included when only diagnosed prenatally. However, it may be difficult to distinguish omphalocele from gastroschisis on prenatal ultrasound, and the terms sometimes are used interchangeably. If it is possible to ascertain the degree of certainty of the prenatal diagnosis and the location of the umbilical cord insertion relative to the abdominal defect, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally before being included. In addition, the absence of omphalocele on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery. |

**Additional Information:**

The distinction between omphalocele and gastroschisis is important because they have different etiologies and different implications for treatment and long-term survival.

In omphalocele, abdominal organs herniate through the umbilicus into the umbilical cord. There is no bridge of skin between the abdominal wall defect and the umbilicus and cord. While the herniating organs are covered by a protective membrane, this may rupture before, during, or after delivery.

In gastroschisis, the umbilicus and cord are normal and separated from the abdominal wall defect by a small bridge of skin. The herniating organs are not covered by a protective membrane. However, they may appear matted and covered by a thick fibrous material as a result of prolonged exposure to amniotic fluid *in utero*.

Omphalocele is one of the defects reported as part of the Omphalocele-Exstrophy-Imperforate Anus-Spina Bifida (OEIS) complex.

Maternal serum alphafetoprotein (MSAFP) and/or amniotic fluid alphafetoprotein (AFAFP) may be elevated with omphalocele. However, these screening tests alone are not sufficient to diagnose the condition.

In contrast to omphalocele, umbilical hernias are completely covered by normal skin.

## Congenital Hip Dislocation

| | |
|---|---|
| **Description** | Location of the head of the femur (bone of the upper leg) outside its normal location in the cup-shaped cavity formed by the hip bones (acetabulum). |
| **Inclusions** | Congenital hip dislocation, unilateral or bilateral<br>Developmental dysplasia of the hip<br>Teratologic hip dislocation |
| **Exclusions** | Flexion deformity/contracture of the hip<br>Hip click<br>Predislocation of the hip<br>Preluxation of the hip<br>Subluxation of the hip<br>Unstable hip |
| **ICD-9-CM Codes** | 754.30, 754.31, 754.35 |
| **CDC/BPA Codes** | 754.30 |
| **Diagnostic Methods** | Hip dislocation may be suspected, and sometimes diagnosed, by physical examination. However, ultrasound or x-ray are the definitive diagnostic tests. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | While this condition may be suspected by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of hip dislocation on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery. |

**Additional Information:**

The terminology describing congenital hip dislocation is often confusing. An unstable hip, in which the femoral head may be moved in and out of the acetabulum on physical examination, often resolves spontaneously in young infants. A truly dislocated hip, in which the femoral head remains out of the acetabulum for a prolonged period, may result in acetabular deformity unless treated. Hence, the designation developmental dysplasia of the hip.

The stability of the hip joint may be evaluated on physical examination. In the Barlow test, lateral pressure is applied to the hip with the knees flexed in an attempt to move the head of the femur out of the hip joint (acetabulum) into a dislocated position. In the Ortolani maneuver, a laterally dislocated femoral head is moved back into normal position in the acetabulum by applying pressure medially. The presence of either sign indicates a hip dislocation is present. However, their absence does not always mean that a dislocation is not present. In some instances, the femoral head may be fixed in a dislocated position and it may not be possible to move it in and out of the joint.

Congenital hip dislocation occurs more frequently after footling or breech deliveries and is more common in females than males. It is most often an isolated condition, although hip dysplasia may occur with generalized skeletal abnormalities and in some genetic syndromes. Some instances of congenital hip dislocation are probably familial.

## Diaphragmatic Hernia

| | |
|---|---|
| **Description** | Incomplete formation of the diaphragm through which a portion of the abdominal contents herniate into the thoracic cavity. |
| **Inclusions** | Absence of the diaphragm<br>Bochdalek hernia – Herniation through a defect in the posterolateral portion of the diaphragm.<br><br>Diaphragmatic hernia, type not specified<br>Hemidiaphragm<br><br>Morgagni hernia – Herniation through a defect in the anterior portion of the diaphragm.<br><br>Paraesophageal hernia – Herniation through a defect in the central portion of the diaphragm surrounding the esophagus. |
| **Exclusions** | Eventration of the diaphragm – Weakness in, or absence of, the muscles of the diaphragm which allows upward displacement of a portion of the abdominal contents. However, there is no true herniation of contents through the diaphragm into the thoracic cavity. |
| **ICD-9-CM Codes** | 756.6 |
| **CDC/BPA Codes** | 756.610 – 756.617 |
| **Diagnostic Methods** | While diaphragmatic hernia may be suspected by the clinical presentation of respiratory distress, feeding intolerance, and/or cardiac compromise, it may be conclusively diagnosed only through x-ray, contrast study of the bowel, CT or MRI scan, surgery, or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | Diaphragmatic hernia may be included in surveillance data when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally before being included. |

**Additional Information:**

Children with diaphragmatic hernia often have accompanying abnormalities of the heart, intestine, and lungs, including hypoplastic lungs, which result from the abnormal location of abdominal organs within the thoracic cavity during development.

## Trisomy 13

| | |
|---|---|
| **Description** | The presence of three copies of all or a large part of chromosome 13. |
| **Inclusions** | Patau syndrome<br>Mosaic Patau syndrome<br>Mosaic trisomy 13<br>Translocation Patau syndrome<br>Translocation trisomy 13<br>Trisomy 13, not otherwise specified<br>Trisomy $D_1$, not otherwise specified |
| **Exclusions** | Balanced translocations involving chromosome 13 |
| **ICD-9-CM Codes** | 758.1 |
| **CDC/BPA Codes** | 758.10 – 758.19 |
| **Diagnostic Methods** | Trisomy 13 may be suspected on physical examination. However, it may be diagnosed conclusively only through direct analysis of the infant's chromosomes (karyotype). The chromosomes may be obtained from blood or tissue cells. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | Trisomy 13 may be included when only diagnosed through direct analysis of fetal chromsomes obtained from amniocentesis, chorionic villus sampling (CVS), or percutaneous umbilical blood sampling (PUBS). However, when mosaic trisomy 13 is noted, the defect should be confirmed postnatally on a specimen obtained directly from the infant or fetus after birth (see below). |

**Additional Information:**

When the two copies of chromosome 13 from one parent do not separate during egg or sperm formation, three copies of the entire chromosome 13 will be present in the fetus. In this instance, the karyotype is written as 47,XX,+13 or 47,XY,+13. This is the most common type of trisomy 13 and is associated with advanced maternal age, particularly of 35 years or greater.

Translocation trisomy 13 occurs when two separate copies of chromosome 13 are present, but a third copy of part of chromosome 13 is attached to another chromosome. In this instance, there are 46 total chromosomes present, but 3 copies of part of chromosome 13.

Mosaic trisomy 13 occurs when some, but not all, of the cells in the body contain three copies of all or a large part of chromosome 13. In this instance, the karyotype is written as 46,XY/47,XY,+13, for example. Because the placenta may contain mosaic cell lines not present in the fetus, mosaic trisomy 13 diagnosed through chorionic villus sampling should always be confirmed by direct examination of fetal chromosomes from amniocentesis, PUBS, or preferably postnatal blood or tissue samples.

Approximately 80% of infants with trisomy 13 do not survive beyond the first month of life. Major malformations associated with trisomy 13 may include holoprosencephaly, microcephaly, meningomyelocele, cleft lip and/or palate, microphthalmia, retinal dysplasia, polydactyly, heart defects (most commonly a VSD), omphalocele, and genitourinary defects, among others. Among children who survive the newborn period, severe developmental delay is virtually always present as may be deafness, visual impairment, minor motor seizures, and apneic spells.

Infants with mosaic trisomy 13 may be less severely affected with variable degrees of developmental delay and longer survival. Infants with partial trisomy for the proximal segment of chromosome 13 (13pter→q14) exhibit a nonspecific pattern of abnormalities with near-normal survival. Approximately 25% of infants with partial trisomy for the distal segment of chromosome 13 (13q14→qter) die during early postnatal life.

Children who survive exhibit severe developmental delay and specific abnormalities.

Major malformations that occur with trisomy 13 in the same infant should be coded separately, as their presence may varies among affected individuals.

## Down Syndrome (Trisomy 21)

| | |
|---|---|
| **Description** | The presence of three copies of all or a large part of chromosome 21. |
| **Inclusions** | Down syndrome<br>Mosaic Down syndrome<br>Mosaic trisomy 21<br>Translocation Down syndrome<br>Translocation trisomy 21<br>Trisomy 21, not otherwise specified |
| **Exclusions** | Balanced translocations involving chromosome 21<br>"Downs facies" without associated trisomy 21. |
| **ICD-9-CM Codes** | 758.0 |
| **CDC/BPA Codes** | 758.00 – 758.09 |
| **Diagnostic Methods** | Down syndrome may be suspected on physical examination. However, it may be diagnosed conclusively only through direct analysis of the infant's chromosomes (karyotype). The chromosomes may be obtained from blood or tissue cells. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | Down syndrome may be included when only diagnosed through direct analysis of fetal chromsomes obtained from amniocentesis, chorionic villus sampling (CVS), or percutaneous umbilical blood sampling (PUBS). However, when mosaic trisomy 21 is noted, the defect should be confirmed postnatally on a specimen obtained directly from the infant or fetus after birth (see below). |

**Additional Information:**

When the two copies of chromosome 21 from one parent do not separate during egg or sperm formation, three copies of the entire chromosome 21 will be present in the fetus. In this instance, the karyotype is written as 47,XX,+21 or 47,XY,+21. This is the most common type of trisomy 21 and is associated with advanced maternal age, particularly of 35 years or greater.

Translocation trisomy 21 occurs when two separate copies of chromosome 21 are present, but a third copy of part of chromosome 21 is attached to another chromosome. In this instance, there are 46 total chromosomes present, but 3 copies of part of chromosome 21.

Mosaic trisomy 21 occurs when some, but not all, of the cells in the body contain three copies of all or a large part of chromosome 21. In this instance, the karyotype is written as 46,XY/47,XY,+21, for example. Because the placenta may contain mosaic cell lines not present in the fetus, mosaic trisomy 21 diagnosed through chorionic villus sampling should always be confirmed by direct examination of fetal chromosomes from amniocentesis, PUBS, or preferably postnatal blood or tissue samples.

Infants with Down syndrome have a typical appearance and other characteristics, including decreased muscle tone (hypotonia), a weak startle (Moro) reflex, hyperflexible joints, a flattened facial profile, upslanting eyes, abnormally shaped external ears (auricles), loose skin on the back of the neck, dysplasia of the pelvic bones, incurving of the fifth finger (clinodactyly), and a single transverse crease in the palm of the hand (Simian crease). Developmental delay is virtually always present. Major malformations associated with Down syndrome include heart defects (most notably endocardial cushion defects), gastrointestinal defects, and vertebral abnormalities, among others.

Major malformations that occur with Down syndrome in the same infant should be coded separately, as their presence may varies among affected individuals.

**Mongolism is an outdated term for Down syndrome.**

## Trisomy 18

| | |
|---|---|
| **Description** | The presence of three copies of all or a large part of chromosome 18. |
| **Inclusions** | Edwards syndrome<br>Mosaic Edwards syndrome<br>Mosaic trisomy 18<br>Translocation Edwards syndrome<br>Translocation trisomy 18<br>Trisomy 18, not otherwise specified |
| **Exclusions** | Balanced translocations involving chromosome 18 |
| **ICD-9-CM Codes** | 758.2 |
| **CDC/BPA Codes** | 758.20 – 758.290 |
| **Diagnostic Methods** | Trisomy 18 may be suspected on physical examination. However, it may be diagnosed conclusively only through direct analysis of the infant's chromosomes (karyotype). The chromosomes may be obtained from blood or tissue cells. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | Trisomy 18 may be included when only diagnosed through direct analysis of fetal chromsomes obtained from amniocentesis, chorionic villus sampling (CVS), or percutaneous umbilical blood sampling (PUBS). However, when mosaic trisomy 13 is noted, the defect should be confirmed postnatally on a specimen obtained directly from the infant or fetus after birth (see below). |

**Additional Information:**

When the two copies of chromosome 18 from one parent do not separate during egg or sperm formation, three copies of the entire chromosome 18 will be present in the fetus. In this instance, the karyotype is written as 47,XX,+18 or 47,XY,+18. This is the most common type of trisomy 18 and is associated with advanced maternal age, particularly of 35 years or greater.

Translocation trisomy 18 occurs when two separate copies of chromosome 18 are present, but a third copy of part of chromosome 18 is attached to another chromosome. In this instance, there are 46 total chromosomes present, but 3 copies of part of chromosome 18.

Mosaic trisomy 18 occurs when some, but not all, of the cells in the body contain three copies of all or a large part of chromosome 18. In this instance, the karyotype is written as 46,XY/47,XY,+18, for example. Because the placenta may contain mosaic cell lines not present in the fetus, mosaic trisomy 18 diagnosed through chorionic villus sampling should always be confirmed by direct examination of fetal chromosomes from amniocentesis, PUBS, or preferably postnatal blood or tissue samples.

Most pregnancies affected with trisomy 18 result in spontaneous abortion. Approximately 50% of

live-born infants with trisomy 18 do not survive beyond the first week of life. Only 5% to 10% survive beyond the first year of life. Major malformations associated with trisomy 18 may include microcephaly, micrognathia, cleft lip and/or palate, heart defects, omphalocele, and renal defects, among others. Minor anomalies associated with trisomy 18 may include low-set malformed auricles (external ears), overlapping of the index and fifth fingers over the third and fourth fingers, absent distal crease on the fifth finger, hirsutism (excess hair) of the forehead and back, lateral deviation of the hands, a hypoplastic thumb, a single transverse palmar crease, and rocker-bottom feet, among others. Developmental delay is virtually always present, as may be hypertonicity, a weak cry, growth retardation, hypoplasia of skeletal muscle and subcutaneous fat, and clenched hands.

Infants with mosaic trisomy 18 may be less severely affected, with variable degrees of developmental delay and longer survival. Infants with trisomy of only the short arm of chromosome 18 (partial trisomy 18) exhibit a nonspecific pattern of abnormalities with mild to no developmental delay. Infants with trisomy of the short arm, centromere, and proximal third of the long arm of chromosome 18 exhibit features of trisomy 18 but not the entire spectrum of abnormalities. Infants with trisomy of only one-third to one-half of the long arm of chromosome 18 exhibit features of trisomy 18 but have longer survival and less severe developmental delays.

Major malformations that occur with trisomy 18 in the same infant should be coded separately, as their presence varies among affected individuals.

## Fetal Aclohol Syndrome (FAS)

| | |
|---|---|
| **Description** | A spectrum of abnormalities resulting from exposure to alcohol *in utero*. While the specific abnormalities vary among individuals, the hallmarks include growth deficiency, microcephaly, facial dysmorphisms, and neurodevelopmental abnormalities. |
| **Inclusions** | Fetal alcohol syndrome (FAS) |
| **Exclusions** | Fetal alcohol effects/facies, without diagnosis of FAS |
| **ICD-9-CM Codes** | 760.71 |
| **CDC/BPA Codes** | 760.71 |
| **Diagnostic Methods** | While fetal alcohol syndrome may be suspected from a history of maternal alcohol use during pregnancy, the condition may be conclusively diagnosed only through direct examination of the infant by a physician (usually a dysmorphologist or developmental specialist) familiar with the spectrum of FAS abnormalities. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | While fetal alcohol syndrome may be suspected from a history of maternal alcohol use during pregnancy, the condition should not be included in surveillance data without postnatal confirmation. |

**Additional Information:**

A number of minor malformations may be present with fetal alcohol syndrome, most notably hypoplasia of the maxillary bone (middle) of the face, and a thin upper lip with smooth philtrum (crease). However, these are often subtle in the newborn and may not be recognized until later in childhood. Older children with FAS may manifest poor coordination, irritability, hyperactivity, and neurodevelopmental abnormalities.

Fetal alcohol syndrome is the extreme of a spectrum of effects on growth and development resulting from alcohol exposure *in utero*. At low levels of exposure, the only apparent effect may be a reduction in birth weight. The clinical features and neurodevelopmental abnormalities become increasingly prominent with increasing levels of exposure.

## Amniotic Bands

| | |
|---|---|
| **Description** | Strands of tissue that float in the amniotic fluid as a consequence of tears or ruptures in the amniotic membrane which surrounds the fetus during development. |
| **Inclusions** | Amniotic bands<br>Amniotic band sequence, syndrome, or disruption complex<br>Amniotic rupture sequence<br>Streeter bands<br>Constriction rings – Soft tissue depressions or grooves encircling part of the body, usually a limb. |
| **Exclusions** | NA |
| **ICD-9-CM Codes** | NA |
| **CDC/BPA Codes** | 658.80 |
| **Diagnostic Methods** | Structural defects resulting from amniotic bands usually are readily apparent on physical examination after delivery. However, the fact that they are a consequence of amniotic bands may not be apparent unless a remnant of an amniotic strand is present or amniotic bands were noted on prenatal ultrasound. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | When amniotic bands are seen on prenatal ultrasound, their presence should be correlated with any structural defects noted, and the guidelines for including those defects when only diagnosed prenatally should be followed. Live-born children who survive should always be examined for evidence of amniotic bands postnatally. In addition, the absence of amniotic bands on prenatal ultrasound does not necessarily mean that they are not truly present. |

**Additional Information:**

Amniotic bands may be present in the amniotic sac without impacting the fetus. When noted as an isolated condition without associated structural defects, they should not be coded.

Structural defects that may occur as a result of amniotic bands include:
>  Pseudosyndactyly (digits compressed together by an encircling band)
>  Distal limb amputation, hypoplasia, lymphedema, or deformation
>  Oral clefts
>  Encephalocele
>  Anencephaly

Other disruptive defects of the skull

***Appendix 3.3***

***Examples of Conditions Considered to Be Minor Anomalies***

# Appendix 3.3
# Examples of Conditions Considered to Be Minor Anomalies[2]

**Eye**
- Epicanthal folds
- Iris freckles, Brushfield spots
- Upward or downward palpebral slant

**Ear**
- Darwinian point or tubercle
- Thickened or excessively folded helix
- Lack of helical folding
- Creased, notched, or bifid ear lobe
- Lop, cup-shaped, or retroverted ear
- Preauricular sinus, cyst, pit, or skin tag

**Head, Face and Neck**
- Flat occiput
- Frontal bossing
- Flat brow
- Flat or prominent bridge of nose
- Anteverted nostrils
- Long nasal septum
- Webbed or redundant neck skin

**Hands and Feet**
- Single or horizontal palmar crease
- Clinodactyly
- Tapered fingers
- Overlapping digits
- Webbed or widely spaced 2nd and 3rd toes
- Prominent heel

**Other**
- Sacral dimples
- Nevi
- Cafe-au-lait spots
- Mongolian spots
- Accessory nipples
- Umbilical hernia
- Vaginal tag
- Single umbilical artery

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[2] This is not a comprehensive list. The exact abnormalities considered to be minor defects may vary among experts.

*Appendix 3.4*

*Conditions Related to Prematurity in Infants Born at Less Than 36
Weeks Gestation*

# Appendix 3.4
# Conditions Related to Prematurity in Infants Born at Less Than 36 Weeks Gestation

> ➢  Dolichocephaly

> ➢  Scaphocephaly

> ➢  Blue sclera

> ➢  Fused eyelids

> ➢  Absent or decreased ear cartilage

> ➢  Patent foramen ovale

> ➢  Patent ductus arteriosus

> ➢  Hypoplastic lungs

> ➢  Small or hypoplastic nipples

> ➢  Hypoplastic labia majora

> ➢  Undescended testicles

> ➢  Inguinal hernia

# Chapter 4
# Data Variables

# Table of Contents

# Appendices

# 4.1  Introduction

The potential data sources available to birth defects programs contain a wide variety of information. Each item of information a birth defects program collects requires staff time to locate, abstract, code, and evaluate, as well as computer space to store it. Thus, due to limited resources, a birth defects program must be efficient in the scope of the information it collects and the manner in which the information is collected and stored.

In this chapter we discuss a number of issues relating to the data variables that comprise a birth defects surveillance system. In Section 4.2, for example, we discuss the criteria that should be considered in selecting the variables that will be collected by a surveillance system. In Section 4.3, we present the three possible origins of surveillance data variables; that is, variables may be abstracted, derived or created. Other topics include possible formats for data variables (Section 4.4), logic checks that can be used to ensure data fall within an expected range (Section 4.5), sources for data variables (Section 4.6), and issues concerning a subset of variables related to birth defects risk factors (Section 4.7). In Section 4.8, we introduce two tables that summarize core (Table 4.1) and recommended (Table 4.2) data variables for a birth defects surveillance system. Additional detail on each of these core and recommended variables is provided in Appendices 4.1and 4.2, respectively.

It is our hope that the information in this chapter of *The Surveillance Guidelines* will promote and guide standardization of data elements across birth defects surveillance programs. Using standard data elements is particularly important when aggregating data for regional or national analysis. Standardization allows and supports comparisons and collaborations between states.

Whether a surveillance program is based on active or passive case ascertainment, our recommendation is that vital records information or copies (including birth, death or fetal death certificates as appropriate) be obtained. This allows the collection of some data using sources from which population-based demographic information can also be obtained.

Note that we are indebted to Lynberg and Edmonds (1994) for much of the information in this chapter.

## 4.2  Criteria to be Considered in Selection of Data Variables

A birth defects program should consider a number of different criteria when deciding which variables to collect. These include type of case ascertainment, program objectives, and data characteristics. Each of these criteria is discussed further below. The criteria considered in compiling the lists of core and recommended variables are summarized for each variable under the heading 'Justification' in Appendices 4.1 and 4.2.

## 4.2.1  Type of Case Ascertainment

The case identification methods used by a surveillance program may place constraints on the data variables collected. The available data source(s) for program variables are determined primarily by these methods. For example, birth certificate files usually offer limited data for diagnostic confirmation of the birth defect or a precise description of the defect. An infant's medical record, other than the newborn record, is not likely to include data on the prenatal care received by the mother (see Chapter 6 on Case Ascertainment Methods).

## 4.2.2  Program Objectives

A surveillance program should limit the information collected to those items needed to fulfill its stated objectives. However, it can be difficult to determine what constitutes this essential information. Often individuals, groups, or organizations that utilize surveillance information may request data on variables that are not really needed and will not be used. One guideline a surveillance program might follow is that information should not be collected if it does not serve at least one programmatic objective.

CDC defines **surveillance** as "the ongoing systematic collection, analysis, and interpretation of health data essential to the planning, implementation and evaluation of public health practice, closely integrated with the timely dissemination of these data to those who need to know" (Centers for Disease Control and Prevention, 1988). Under this definition, it is clear there are a number of functions and objectives for which a birth defects program might need to collect data:

➢ *Descriptive epidemiology and monitoring.* Data can be examined to determine and describe the distribution of a disease (condition) within a population along the parameters of place, person, and time. Monitoring offers quantitative estimates of the magnitude of the disease.

➢ *Research.* Data can be used to test hypotheses or in planning research to learn the causes of a disease.

➢ *Service/planning.* Increasingly, surveillance programs are using information on newly identified children with birth defects to refer them for services. These include specialized medical care, educational and early intervention programs, and genetic counseling.  Data can also be applied to evaluate services and prevention measures within a population. Knowledge about the disease or condition and changes in the population can assist in optimizing available resources and services.

➢ *Linkage.* Variables may be used to link to other databases such that data in those databases may be associated at the case level to complement and enrich case-specific data. Linkage is also an essential surveillance management tool needed to identify and consolidate duplicates.

# 4.2.3 Data Characteristics

Among the important data characteristics a surveillance program should consider are availability, consistency, accuracy, uniqueness, definability, collectability, and comparability. We discuss each of these in turn below.

➢ *Availability.* Data must be retrievable from the data sources and be available to the birth defects program. Many data variables are collected and stored at data sources in clinical and administrative databases, facilitating availability and retrievability. In most cases, information should only be collected if it is consistently available. This is particularly true if the information is to be used for statistical analyses or for identifying or contacting case families. If information can be found only in a small portion of the data sources, then staff will spend considerable time looking for unavailable information. The birth defects program may want to either limit collection of such information or work to identify a data source where the same information is consistently available. An exception to this may be where the information is important even if it is only occasionally found in the data sources (e.g., the fact that the infant is in foster care or has been placed for adoption). However, as noted before, this information may be difficult to find and time-consuming to collect.

➢ *Consistency*. It is important that the information assembled within the surveillance system has a consistent meaning from report to report. When obtaining information from a range of data sources, it is essential to have a usable level of consistency from source to source. This is especially important for passive data collection and data mining. Simple issues, such as field content and even field size, can significantly affect the comparability and usefulness of the data. Coding rules and practices are special areas of concern.

➢ *Accuracy.* The information collected should be accurate. If the information is of questionable veracity, then it should not be collected. Second-hand information found in medical records may be incomplete or inaccurate. If information such as medication use and exposures is important, it should be collected from a reliable source, such as through direct contact with the mother, rather than from medical records.

➢ *Uniqueness.* Programs should avoid the collection of redundant information. Information should not have to be recorded in more than one field. For example, if the infant or fetus delivery date and the mother's date of birth are collected, then the mother's age at delivery does not need to be collected.

➢ *Definability.* There should be clear definitions for each of the data variables a birth defects program collects.

➢ *Collectability.* The data variables should lend themselves to easy abstraction. This is a potential problem with complex or subjective information. If it takes an excessive amount of time to track down and collect the information, or if there is a high degree of inter-staff variability in how the information is collected, then the information recorded in the birth defects program's database will be of dubious quality and reliability (Horwitz and Yu, 1984; Demlo et al., 1978). In addition, extensive efforts may be necessary for quality control.

➢ *Comparability.* The birth defects program may want to consider whether other birth defects programs have access to the same sources and types of data. If the program uses a unique data source or collects a unique data variable that other birth defects programs do not, then the program may not be able to compare its data to those of other programs. This may be of limited importance, however, if the data are being collected to meet specific programmatic objectives, where comparison between different states or programs is unimportant.

# 4.3  The Origins of Data Variables

Data variables may be abstracted, derived, or created.

> ➤ *Abstracted data variables.* These are data that are available only from the data sources, and the data sources must supply them.

> ➤ *Derived data variables.* Some data variables are not collected directly from data sources but are rather derived from other information collected from the data sources, e.g., census tract numbers, standardized geographic tables, disease codes.

> ➤ *Created data variables.* Some data variables may need to be created by the birth defects program, e.g., unique case and staff IDs.

Some data variables may fall into more than one of the above categories. For example, if the mother's age at delivery is not available from the data sources, it may be derived using the date of delivery and the mother's date of birth. The origins of each of the core and recommended variables are summarized under the heading 'Source' in Appendices 4.1 and 4.2.

## 4.4  The Formats of Data Variables

Data may be stored in a computer database in a variety of formats, including as a numerical field, a date field, a text field, a checkbox, or a coded data field. Each of these formats is briefly described below. The format for each of the core and recommended variables is also summarized under the heading 'Type' in Appendices 4.2 and 4.2.

> ➤ *Numerical field.* A field that includes only numbers.

> ➤ *Date field.* A field that includes only dates, which are comprised of month, day, and year in a variety of orders and combinations.

> ➤ *Text field.* A field that can contain letters, numbers, and punctuation. Text fields are often of a fixed width. Text fields of infinite width are often called Amemo@fields.

> ➤ *Checkbox.* A field that contains only two options – yes/no, on/off.

> ➤ *Coded data field.* Data may be collected and stored as they appear in the data source, or they may be 'coded'. A code may contain numbers or letters or both. Whether a birth defects program collects and stores data as coded or not depends on the types of data, as well as on potential uses.

If a birth defects program plans to use a field for analysis, then it is important that the field be easily coded or categorized, permitting ready analysis rather than having to sort through a large collection of free-form text. This is because information such as race/ethnicity, diagnoses, and conditions can be described in a number of different ways. For example, a person may be described as 'African-American' or 'black'. A 'cleft lip' may also be described as a 'lip cleft' or a 'harelip'.

Coding eliminates the problem of having to sort through a variety of differing descriptions. It allows for timely and efficient analysis of data and referral of cases. Coding also enables researchers to know that they are talking about the same thing, and it allows for comparability between different birth defects programs using the same or comparable coding systems.

Whenever possible, a birth defects program should use coding systems consistent or compatible with those used by other groups, particularly other birth defects programs, thus allowing for efficient comparison of data. This applies not only to diagnostic codes but also to characteristics such as maternal race and ethnicity.

# 4.5  Data Variable Logic Checks

Errors may occur in the data collection by a birth defects program, either because of errors in data listed in the data source or because of errors in abstraction. A birth defects program should have some method to identify and correct errors (see Chapter 7 on Data Quality Management). One means of identifying and correcting errors is through *logic checks* that ensure data occur within expected ranges.

Many of the core variables in a birth defects surveillance system have a limited number of options or ranges of values. For example, a gestational age of 75 weeks is highly unlikely to occur. And other variables may have certain logical relationships to one another. For example, the mother's date of birth must always be earlier than the infant's date of birth.

Suggested logic checks for each of the core and recommended variables are summarized under the heading 'Checks' in Appendices 4.1 and 4.2.

# 4.6  Data Variable Location

A birth defects surveillance program may have access to a variety of data sources and will collect data on a number of different variables. Clearly, the same variable may be available from several sources. Abstracting data from a variety of sources allows for greater thoroughness in data collection. If a variable is missing in one data source, it may be available in another source.

Staff collecting data should know where a given data variable is likely to be found, as well as the prioritization of sources for those variables retrievable from multiple data sources, since data sources may disagree as to the value for a particular variable. For example, the infant's delivery medical record and the birth certificate might record different values for birth weight. A birth defects program should prioritize the data sources for particular variables. In the above instance, for example, a birth defects program may decide that the birth weight in the medical record takes precedence over the birth weight from a birth certificate.

For each of the core and recommended variables, the data source – as well as the location within the data source where the variable is most likely to be consistently found – are summarized under the heading 'Location' in Appendices 4.1 and 4.2.

# 4.7  Risk Factor Variables

Risk factors in birth defects include:  conditions, illnesses, or complications during pregnancy, labor, or delivery

Selected conditions, such as maternal diabetes and thyroid disease, have been associated with increased risk for certain birth defects (Becerra et al., 1990; Khoury et al., 1989). Information on conditions and complications during pregnancy and delivery may be useful for making syndromic classifications or identifying causality of birth defects, such as diabetic embryopathy.

However, there are a large number of conditions and complications possible during pregnancy and delivery, and birth defects programs could create lists of dozens to hundreds of them. Such long lists would require additional computer storage space and training of field staff regarding where to find the information and how to collect it. Even then, confusion may ensue over which conditions and complications to abstract and subjective differences between staff in their abstraction of this information. Moreover, the information in the data sources commonly available to birth defects programs may not necessarily be consistent or accurate (Olson et al., 1997).

For all of these reasons, birth defects surveillance programs should give careful consideration to the potential thoroughness and usefulness of routine data collection regarding risk factors as relevant to their goals and objectives. In general, programs are more likely to obtain useful information on conditions and complications during pregnancy and delivery through contact with parents, as is done in case-control research studies, than through medical records abstraction.

# 4.8  Data Variable Tables

In the late 1980s, before creation of the National Birth Defects Prevention Network, Larry Edmonds of the Centers for Disease Control and Prevention (CDC) – along with F. John Meaney of Arizona and Susan Panny of Maryland and others – collaborated on development of a set of core data items relevant to birth defects surveillance (Edmonds et al., 1988), based on an earlier list developed by CDC's National Center for Health Statistics. We used the list developed by Edmonds et al. as the foundation for developing the current list of data variables that the NBDPN recommends for birth defects surveillance programs, adding a number of different variables in order to reflect the fact that birth defects surveillance programs have evolved considerably since the 1980s into programs with a variety of objectives and multiple areas of interest.

The data variables in Tables 4.1 and 4.2 (as well as in their corresponding appendices) are categorized as to whether they are infant, maternal, paternal, or contact information variables. For each data variable, we also note in Tables 4.1 and 4.2 the usefulness of that data item relative to a program's specific objectives, which may include descriptive epidemiology and monitoring, research, service and planning, and linkage capability (see Section 4.2.2. for further discussion of program objectives).

To provide a sense of the relative importance of the data variables for a new or expanding surveillance program, we have further distinguished between minimum (or core) variables (Table 4.1 and Appendix 4.1) and recommended variables (Table 4.2 and Appendix 4.2).

> ➢ ***Minimum (core) variables*** are those that are considered necessary to fulfill the most basic programmatic objectives and that also meet most or all of the supplemental criteria discussed earlier in this chapter.

> ➢ ***Recommended variables*** are those that have the potential to enhance surveillance capability or to support broader programmatic objectives.

By glancing down the column for a specific programmatic objective (e.g., 'research'), the reader can determine – based on the relevant check marks – which elements are considered 'core' and which other data elements are 'recommended' to support a given program objective. These data variables can be abstracted using a minimum number of data sources, including maternal records, infant records, and vital records. Birth defects programs that use the passive case ascertainment approach will find the vital record particularly useful as a data source for many of the maternal core data variables.

After reviewing these lists, birth defects surveillance staff may also wish to add further data variables they consider essential for their own specific programmatic purposes.

**Table 4.1**
**Minimum (Core) Data Variables**

| Data Variable | Descriptive Epidemiology and Monitoring | Research | Service/ Planning | Linkage |
|---|---|---|---|---|
| *Infant* | | | | |
| Unique ID | ✔ | ✔ | ✔ | ✔ |
| Date of Pregnancy Outcome | ✔ | ✔ | ✔ | ✔ |
| Sex | ✔ | ✔ | ✔ | ✔ |
| Infant's Name<br>First<br>Middle<br>Last<br>Suffix | ✔<br>✔<br>✔<br>✔ | ✔<br>✔<br>✔<br>✔ | ✔<br>✔<br>✔<br>✔ | ✔<br>✔<br>✔<br>✔ |
| Source of Report | ✔ | ✔ | ✔ | ✔ |
| Medical Record Number(s) | ✔ | ✔ | ✔ | ✔ |
| Vital Record Certificate Number | | | | ✔ |
| Place of Pregnancy Outcome | ✔ | ✔ | ✔ | ✔ |
| Pregnancy Outcome | ✔ | ✔ | ✔ | ✔ |
| Birth Weight | ✔ | ✔ | ✔ | ✔ |
| Plurality | ✔ | ✔ | ✔ | ✔ |
| Gestational Age | ✔ | ✔ | ✔ | ✔ |
| Diagnosis Code | ✔ | ✔ | ✔ | ✔ |
| *Contact Information* | | | | |
| Name of Responsible Party | | | ✔ | |
| Address of Responsible Party | | | ✔ | |
| Telephone Number of Responsible Party | | | ✔ | |
| *Mother* | | | | |
| Mother's Date of Birth | ✔ | ✔ | ✔ | ✔ |
| Mother's Race | ✔ | ✔ | | |
| Mother's Ethnicity | ✔ | | | |
| Mother's Name<br>First<br>Middle<br>Last | ✔<br>✔<br>✔ | ✔<br>✔<br>✔ | ✔<br>✔<br>✔ | ✔<br>✔<br>✔ |
| Mother's Residence At Time of Pregnancy Outcome<br><br>Street address<br>City<br>County<br>State<br>Zip Code | ✔<br>✔<br>✔<br>✔<br>✔ | ✔<br>✔<br>✔<br>✔<br>✔ | | |

**Table 4.2**
**Recommended Data Variables**

| Data Variable | Descriptive Epidemiology and Monitoring | Research | Service/ Planning | Linkage |
|---|:---:|:---:|:---:|:---:|
| *Infant* | | | | |
| Text Description of Birth Defect | ✔ | ✔ | ✔ | |
| Date of Death | ✔ | ✔ | ✔ | ✔ |
| Birth Length | ✔ | ✔ | | |
| Apgar Score | ✔ | ✔ | | |
| Birth Order | ✔ | ✔ | ✔ | |
| Cytogenetic Analyses Performed | ✔ | ✔ | ✔ | |
| Diagnostic Tests and Procedures Performed | ✔ | ✔ | ✔ | |
| Autopsy Performed | ✔ | ✔ | ✔ | |
| Physicians of Record | | ✔ | ✔ | |
| *Mother* | | | | |
| Date of Last Menstrual Period (LMP) | ✔ | ✔ | | |
| Date of Ultrasound | ✔ | ✔ | | |
| Gestational Age at Ultrasound | ✔ | ✔ | | |
| Mother's Medical Record Number(s) | ✔ | ✔ | | ✔ |
| Prenatal Diagnosis | ✔ | ✔ | | |
| Mother's Social Security Number | | ✔ | | ✔ |
| Census Tract of Maternal Residence at Pregnancy Outcome | ✔ | ✔ | | ✔ |
| Mother's Telephone Number | | ✔ | ✔ | |
| Mother's Education | ✔ | ✔ | | |
| Prior Pregnancy History | ✔ | ✔ | | ✔ |
| Prenatal Care | ✔ | ✔ | | |
| *Father* | | | | |
| Father's Date of Birth | ✔ | ✔ | | ✔ |
| Father's Name | | ✔ | ✔ | |
| Father's Education | ✔ | ✔ | | |
| Father's Race | ✔ | ✔ | | |
| Father's Ethnicity | ✔ | ✔ | | |
| Father's Social Security # | | ✔ | | ✔ |

# 4.9 References

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*Appendix 4.1*

*Descriptions of Minimum (Core) Data Variables*

# Appendix 4.1
## Descriptions of Minimum (Core) Data Variables

# Appendix 4.1
## Descriptions of Minimum (Core) Variables

### Format for Variable Descriptions

**Variable Name**      Name of data collection variable

**Definition**      Definition of data collection variable

**Justification**      Reason the birth defects program may want to include variable in its database

**Source**      Where variable comes from – abstracted, derived, created

**Location**      Data sources and location within data sources where variable is most likely to be consistently found

**Type**      How variable should be stored – text, number, date, code (letters and/or numbers), checkbox

**Checks**      Any limits, ranges, or other criteria the variable should meet

**Comments**      Other notes or comments about the variable

**Options**      Recommended options for the variable

# Infant Variables – Core

| | |
|---|---|
| **Variable Name** | *Unique ID* |
| **Definition** | Identification code or number; a code or number that uniquely identifies each case or record |
| **Justification** | With a unique ID code, the birth defects program can refer to a particular case more easily than having to refer to a set of other variables. For example, it is easier to refer to an abstract with ID 1234567 than to an abstract of John Doe, date of birth 04/27/1999, born to mother Jane Doe. |
| | The ID permits easy linkage between multiple data sets as long as each table contains the ID as one of its fields. This is essential for data transfer and processing, so that data for a particular case do not get mixed up with data from other cases. This field permits linking multiple case reports for individual children. |
| **Source** | Created by the registry as cases are added. |
| **Location** | N/A |
| **Type** | Code |
| **Checks** | Every individual in the database should have a *unique* ID. |

# Infant Variables – Core

| | |
|---|---|
| **Variable Name** | ***Date of pregnancy outcome*** |
| **Definition** | Date of delivery or end of the index pregnancy |
| **Justification** | In conjunction with other fields, such as mother's last name, this field helps to identify a case uniquely. It is useful to researchers and social workers in verifying that they are referring to the pregnancy of interest when contacting mothers who may have had other pregnancies. |

The birth defects program may require that, for live births, a diagnosis be made within a certain time period after the date of delivery (e.g., within one year) or by a particular age (e.g., prior to age 6). The date of delivery is necessary in order to determine whether the diagnosis was made within the time limit.

Secular trends have been reported for certain birth defects (Nielsen et al., 2000; O'Leary et al., 1996; Centers for Disease Control and Prevention, 1992). The birth defects program can use the date of delivery in order to produce statistics and reports by delivery year and to examine secular trends in birth defects.

Cluster investigations are based on a defined diagnosis, geographical area, and time period. Knowing the delivery date allows investigators to determine which cases qualify to be included in a particular investigation.

| | |
|---|---|
| **Source** | Abstracted |
| **Location** | • Mother's delivery medical record (labor and delivery record) <br> • Infant's medical record (face sheet, labor and delivery record, birth certificate worksheet) <br> • Vital record |
| **Type** | Date |
| **Checks** | Every record must have a pregnancy outcome date, except in cases of prenatal diagnosis where the pregnancy has not ended yet. The pregnancy outcome date should be after the mother's and father's date of birth, date of last menstrual period, and date of conception; on or after any prenatal diagnostic procedure date or prenatal ultrasound date; and on or before a postnatal procedure date. |
| **Comments** | Date of pregnancy outcome can be: date of birth, date of fetal demise, or date pregnancy ends. |

# Infant Variables – Core

| | |
|---|---|
| **Variable Name** | *Sex* |
| **Definition** | Sex of the infant or fetus |
| **Justification** | Birth defect risk may be associated with sex (Whiteman et al., 2000). The birth defects program can use the sex of the infant or fetus in order to evaluate differences in birth defect rates by sex. |
| **Source** | Abstracted<br>Derived (from the karyotype) |
| **Location** | • Mother's delivery medical record (labor and delivery record)<br>• Infant's medical record (face sheet, labor and delivery record, birth certificate worksheet)<br>• Vital record |
| **Type** | Code |
| **Checks** | Every record should have sex recorded. |
| **Comments** | If a karyotype was performed, the sex should match the karyotype, except in rare cases of such discordances as XY females and XX males. |
| **Options** | • Male<br>• Female<br>• Ambiguous<br>• Unknown |

# Infant Variables – Core

| | |
|---|---|
| **Variable Name** | ***Infant's name*** |
| **Definition** | Name of infant or fetus |
| **Justification** | Field staff can use the infant's name and date of birth or pregnancy outcome date to locate medical records. |
| | The birth defects program will use the name to unduplicate the reported cases and may employ the infant's name in addition to other fields to link to other data sets, such as vital records. |
| | The infant's name is helpful when referring the family to social work, treatment, and prevention agencies. |
| **Source** | Abstracted |
| **Location** | • Infant's medical record (face sheet, birth certificate worksheet) <br> • Vital record |
| **Type** | Text |
| **Checks** | If the infant's last name is the same as the father's or mother's last name, or a combination of the two, the spelling should usually match. |
| **Comments** | This variable may be collected as a single field or multiple fields. Separate fields for first, middle, last name, and suffix are recommended to improve unduplication and record linkage success. Individual fields of up to 25 digits each should be considered to avoid truncated names. This variable should include at least the infant's first and last name and may include the infant's middle name and any suffixes. An infant may be given more than one name or alias, sometimes referred to as "also known as" or AKA. The birth defects program may want to record all of the names, for easier linkage with other databases, to prevent duplication of cases in the database and to remain current with name use. |
| | Fetuses resulting from fetal deaths and elective terminations often do not have names. The birth defects program should consider using the surname of the mother and inserting a standard first name (e.g., fetus) so that name data fields are complete in the database. |

# Infant Variables – Core

| | |
|---|---|
| **Variable Name** | *Source of report* |
| **Definition** | Any data source where information was obtained or where a case report originated. |
| **Justification** | The source of report allows the birth defects program to identify where information in a case abstract comes from. This is important for resolving data edit issues, for confirming the data, and for conducting audits of facility reporting.<br><br>The data source fields permit the birth defects program to evaluate the usefulness of utilizing specific facilities as data sources. |
| **Source** | Abstracted |
| **Location** | Any data source pertinent to program objectives |
| **Type** | Code, with allowance for multiple sources |
| **Checks** | This field should always be filled out and should be a valid code. |
| **Comments** | There can be multiple data sources for a given case. For example, an infant may be identified with a birth defect at the delivery hospital, tertiary care hospital, cytogenetics laboratory, etc. (see also Chapter 6 on Case Ascertainment Methods). |
| **Options** | It is useful to maintain a list of potential data sources and standard codes (hospitals, clinics, laboratories, autopsy, etc.) unique to each program. |

| | |
|---|---|
| **Variable Name** | *Medical record number(s)* |
| **Definition** | Medical record number(s) used by the source from which the information was obtained. |
| **Justification** | A medical record number allows facilities to retrieve records easily. Although it may be possible to locate medical records using the patient's name and date of birth, the birth defects program may have a name different than that recorded at the data source. |
| **Source** | Abstracted |
| **Location** | • Infant's medical record (face sheet) |
| **Type** | Code |
| **Comments** | Medical record numbers are not the same as visit, service, or encounter numbers. Although not standard practice, multiple 'real' medical record numbers may be assigned to the same person, so it is important to identify each number for a given data source. Medical record numbers may also be very long. The birth defects program should make certain the computer program and registry database allow for entry of the entire medical record number. |

# Infant Variables – Core

| | |
|---|---|
| **Variable Name** | ***Vital record certificate number*** |
| **Definition** | Unique number assigned to a certificate by Vital Records. |
| **Justification** | Vital record certificate numbers can be linked to other vital records certificates. Often, vital records will reference or link the death certificate to the birth certificate for infant deaths. |
| | Programs can use an algorithm of data variables to find a potential match to a vital record. This process assists in identifying unique cases, establishing residency, and securing all of the data variables on the vital record. |
| | Birth certificate and fetal death certificate numbers can be the unique ID numbers for a program. Other 'program' numbers can be created using a similar format for cases that do not match to a birth certificate or fetal death certificate. |
| **Source** | Abstracted |
| **Location** | On the certificate of birth, death, or fetal death and in the vital records database |
| **Type** | Number |
| **Comments** | Separate fields for the live birth or fetal death and for the death record number are recommended. |

# Infant Variables – Core

| | |
|---|---|
| **Variable Name** | ***Place of pregnancy outcome*** |
| **Definition** | Location where the delivery or pregnancy outcome occurred |
| **Justification** | Mother and infant records at the delivery facility often provide important information not found in tertiary care facility records (unless the delivery records are copied into the tertiary care records). The birth defects program can use the delivery location (hospital, midwifery, residence, etc.) to identify where delivery records need to be reviewed and abstracted. |
| | The birth defects program may employ the delivery location in addition to other fields to link to other data sets, such as vital records. |
| | The location where the delivery occurred allows the birth defects program to provide facility-specific statistics. |
| **Source** | Abstracted |
| **Location** | • Mother's delivery medical record (face sheet) <br> • Infant's delivery medical record (face sheet) <br> • Vital record |
| **Type** | Code |
| **Checks** | This field should always be filled out and must be a valid code. |
| **Comments** | This includes those situations where delivery occurs outside of health care facilities as well as inside health care facilities. |
| **Options** | It is useful to maintain a list of potential data sources (hospitals, etc.) unique to each program. <br> • Home/residence <br> • Other <br> • Unknown |

# Infant Variables – Core

| | |
|---|---|
| **Variable Name** | ***Pregnancy outcome*** |
| **Definition** | Outcome of the index pregnancy |
| **Justification** | The pregnancy outcome, in conjunction with gestational age fields, may determine whether a record should be included in the birth defects program. |
| | At a minimum birth defect programs should distinguish the outcomes of live birth, fetal death, and induced termination. |
| | Part of the mission of the birth defects program may be to refer families to social services. Since only live births would be referred to many of the services, it is important to know whether a given case is a live birth. Knowing which cases are elective terminations aids in evaluating trends in prenatal diagnosis, as well as evaluating the impact of prevention strategies such as folic acid supplementation and fortification. |
| | Pregnancy outcome can be used to evaluate rates of birth defect by pregnancy outcome. |
| **Source** | Abstracted |
| **Location** | • Mother's delivery medical record (face sheet, discharge summary, labor and delivery record)<br>• Vital records |
| **Type** | Code |
| **Checks** | This field should always be filled out, except in cases of prenatal diagnosis where the pregnancy has not yet ended. |
| **Comments** | See Chapter 3 on Case Definition for definitions of pregnancy outcomes. |

# Infant Variables – Core

| | |
|---|---|
| **Variable Name** | ***Birth weight*** |
| **Definition** | Weight of the infant or fetus at delivery |
| **Justification** | The birth weight may be needed for case definition if inclusion/exclusion criteria for selected birth defects, such as for undescended testes and patent ductus arteriosus, are based on birth weight. |
| | In conjunction with gestational age, length, and head circumference, birth weight can be used to assess prenatal growth retardation, a characteristic of fetal alcohol syndrome. |
| **Source** | Abstracted |
| **Location** | • Mother's delivery medical record (labor and delivery record) <br> • Infant's medical record (admission summary, labor and delivery record, birth certificate worksheet) <br> • Vital records |
| **Type** | Number |
| **Checks** | The birth weight must range between 0 and 10,000 grams or 0 and 50 pounds. |
| **Comments** | The data source may report birth weight in grams or kilograms, pounds and ounces, or pounds with decimals. The birth defects program may decide to record the weight in the units reported or in a uniform fashion, such as always as grams and kilograms. In this latter case, the birth defects program must be able to convert from one type of unit to another while collecting the data. Data fields can have computerized calculation functions. |

# Infant Variables – Core

| | |
|---|---|
| **Variable Name** | ***Plurality*** |
| **Definition** | Number of fetuses or infants. |
| **Justification** | The plurality, in association with other fields such as county of residence and mother's social security number, can be used to avoid duplication of records in the birth defects program. |
| | Knowing that the infant is from a multiple birth alerts the birth defects program that more effort may be needed to link to a particular vital record (Forrester and Canfield, 2000). |
| | The birth defects program can use this data item to evaluate differences in birth defect rates for singletons and multiple births. |
| **Source** | Abstracted |
| **Location** | • Mother's delivery medical record (face sheet, admission summary, discharge summary, prenatal care record, labor and delivery record)<br>• Infant's medical record (face sheet, admission summary, discharge summary, prenatal care record, labor and delivery record, birth certificate worksheet).<br>• Vital records |
| **Type** | Number |
| **Checks** | This field should always be filled out. |
| **Comments** | Because some twin pregnancies are anomalous (for example, conjoined twins or fetus papyraceus), there may not be the expected two vital records for a pregnancy that is identified as a twin pregnancy. |

# Infant Variables – Core

| | |
|---|---|
| **Variable Name** | ***Gestational age*** |
| **Definition** | Gestational age at pregnancy outcome |
| **Justification** | Gestational age can be used to determine whether a pregnancy outcome meets the case definition for the birth defects program. |
| | Certain diagnoses are considered birth defects only when the infant is of a particular gestational age. For example, patent ductus arteriosus is common among premature infants and is often subject to exclusion criteria before being counted as a birth defect. |
| **Source** | Abstracted<br>Derived (see comments) |
| **Location** | • Mother's delivery medical record (labor and delivery record)<br>• Infant's delivery medical record (admission summary, discharge summary, gestational age score record, labor and delivery record, birth certificate worksheet)<br>• Vital records |
| **Type** | Number |
| **Checks** | The gestational age should range between 0 and 52 weeks. |
| **Comments** | The gestational age can be derived via several methods, and conflicting gestational age information may be reported in the medical record (Alexander et al., 1990; Hall, 1990). As a result, the birth defects program will want to have a method for prioritizing gestational age estimates from different sources. |
| **Options** | See Chapter 3 on Case Definition for further information. |

# Infant Variables – Core

| | |
|---|---|
| **Variable Name** | ***Diagnosis code*** |
| **Definition** | Code used for the diagnosis |
| **Justification** | Coding birth defects eliminates the problem of having to sort through a variety of differing descriptions. It allows for timely and efficient analyses of data and identification of cases for research and referral. Coding of birth defects enables birth defects researchers to know that they are talking about the same birth defect, and allows for comparability between different birth defects registries using the same or comparable coding systems (Rasmussen et al., 2001). |
| **Source** | Derived |
| **Location** | N/A |
| **Type** | Code |
| **Checks** | Every case should have at least one diagnosis code (except if the birth defects program includes non-malformed controls, in which case the program may create specific 'disease codes' for use as the data variable in the diagnosis code field).<br><br>A case may have more than one diagnosis code. Every diagnosis description should have a corresponding code and vice versa. |
| **Comments** | The International Classification of Disease (ICD) coding system is the standard used in the health care delivery system. NBDPN currently requires that programs report cases using ICD-9-CM codes. The registry should accommodate a minimum of 15 unique diagnostic codes per case. |
| **Options** | The recommended coding system is the CDC 6-digit code, which is easily converted to ICD-9-CM. See the Chapter 5 on Classification and Coding for further information. |

# Contact Information – Core

| | |
|---|---|
| **Variable Name** | ***Name of responsible party*** |
| **Definition** | Name of parent, custodial parent, or guardian |
| **Justification** | Useful in programs that refer a family to services when contact with a parent may be inappropriate. |
| **Source** | Abstracted |
| **Location** | • Face sheet, signed authorization, social worker's notes, birth certificate worksheet<br>• Immunization registry, metabolic screening database<br>• Vital record |
| **Type** | Text |
| **Comments** | The name may be collected as a single field or multiple fields for first, middle, and last name. Allowing for up to 25 characters for each portion of the name should be considered. |

| | |
|---|---|
| **Variable Name** | ***Address of responsible party*** |
| **Definition** | Address of parent, custodial parent, or guardian |
| **Justification** | Useful in programs that refer a family to services when contact with a parent may be inappropriate. |
| **Source** | Abstracted |
| **Location** | • Face sheet, signed authorization, social worker's notes, birth certificate worksheet<br>• Immunization registry, metabolic screening database<br>• Vital record |
| **Type** | Text |
| **Comments** | Include fields for the street address, city, state, and zip code. Allow at least 25 digits for street address and 20 digits for city name. If the residence address and the mailing address of the responsible party are different, collect the mailing address for this item. |

| | |
|---|---|
| **Variable Name** | ***Telephone number of responsible party*** |
| **Definition** | Telephone number of child's parent, custodial parent, or guardian |
| **Justification** | Useful in programs that refer a family to services when contact with a parent may be inappropriate and when telephone contact may be indicated. |
| **Source** | Abstracted |
| **Location** | • Face sheet, signed authorization, social worker's notes, birth certificate worksheet<br>• Immunization registry, metabolic screening database<br>• Vital record |
| **Type** | Number |
| **Comments** | Include area code with number. |

# Maternal Variables – Core

| | |
|---|---|
| **Variable Name** | ***Mother's date of birth*** |
| **Definition** | Birth mother's date of birth |
| **Justification** | In conjunction with other fields, such as mother's name, the birth defects program field staff can use the mother's date of birth to locate medical records when the mother's medical record number is not known. The birth defects program can use the mother's date of birth and other fields to determine whether a case has been abstracted or added to the registry under a different ID. |

The birth defects program can employ the mother's date of birth in addition to other fields to link to other data sets, such as vital records or Medicaid.

The birth defects program can use the mother's date of birth and infant's date of delivery in order to calculate the mother's age at delivery. The mother's age at delivery can then be used in clinical review.

| | |
|---|---|
| **Source** | Abstracted |
| **Location** | • Mother's delivery medical record (face sheet, prenatal care record)<br>• Infant's medical record (face sheet, prenatal care record, birth certificate worksheet)<br>• Vital records |
| **Type** | Date |
| **Checks** | This date must be earlier than all other dates except possibly the father's date of birth. Medical records may sometimes confuse maternal and paternal information. If the mother's date of birth is the same as the father's date of birth, the birth defects program should double check to make certain that this is true. |
| **Comments** | See also Chapter 6 on Case Ascertainment Methods, the section on Data Sources. |

# Maternal Variables – Core

| | |
|---|---|
| **Variable Name** | *Mother's race* |
| **Definition** | Birth mother's race |
| **Justification** | The birth defects program can use the mother's race in order to present data on birth defect rates by maternal race, one of the most important person variables in descriptive epidemiology. |
| **Source** | Abstracted |
| **Location** | • Mother's delivery medical record (face sheet, prenatal care record)<br>• Infant's medical record (admission summary, prenatal care record, birth certificate worksheet)<br>• Vital records |
| **Type** | Code |
| **Comments** | Racial categories and codes used by birth defects surveillance programs should be compatible with the federal standards in current use for race. |

| | |
|---|---|
| **Variable Name** | *Mother's ethnicity* |
| **Definition** | Birth mother's ethnicity |
| **Justification** | Ethnicity is a designation separate from maternal race. The birth defects program can use the mother's ethnicity in order to evaluate differences in birth defect rates by maternal ethnicity. |
| **Source** | Abstracted |
| **Location** | • Mother's delivery medical record (face sheet, prenatal care record)<br>• Infant's medical record (admission summary, prenatal care record, birth certificate worksheet)<br>• Vital records |
| **Type** | Code |
| **Comments** | It is important to collect ethnicity data that meets the needs of the registry to monitor the health of the ethnic populations within the state. Generally, each state Department of Public Health will have identified the populations of special interest to that state. Ethnicity categories and codes should be compatible with the federal standards in current use for ethnicity. |

# Maternal Variables – Core

| | |
|---|---|
| **Variable Name** | ***Mother's name*** |
| **Definition** | Full name of birth mother |
| **Justification** | In conjunction with other fields, such as mother's date of birth, the birth defects program field staff can use the mother's name to locate medical records when the mother's medical record number is not known. The birth defects program can employ the mother's name in addition to other fields to unduplicate case reports and to link to other data sets, such as vital records or Medicaid.<br><br>The mother's name is needed so that she can be contacted by researchers conducting approved studies and by social workers or others for outreach efforts. |
| **Source** | Abstracted |
| **Location** | • Mother's delivery medical record (face sheet)<br>• Infant's medical record (face sheet, birth certificate worksheet)<br>• Vital records |
| **Type** | Text |
| **Comments** | This variable may be collected as a single field or multiple fields. This variable should include at least the mother's first and last name and may include the mother's middle name and maiden name (name before marriage). A woman may have more than one name or alias (also known as or AKA). Separate fields for first, middle, and last name and for maiden name are recommended. Field lengths of 25 characters or larger for each portion of the name should be considered. The birth defects program should record all of the names, for easier linkage with other databases and to prevent entering duplicate cases in the database. |

# Maternal Variables – Core

| | |
|---|---|
| **Variable Name** | ***Mother's street address of residence at pregnancy outcome*** |
| **Definition** | Street address of birth mother's residence at the time of the outcome of the index pregnancy |
| **Justification** | Street address is necessary for geocoding location of residence and linking with other data systems through geographical information systems (GIS). The street address field may be needed when assigning the county of residence, particularly when a city includes part of more than one adjacent county. |
| **Source** | Abstracted |
| **Location** | • Mother's delivery medical record (face sheet) <br> • Infant's delivery medical record (face sheet, birth certificate worksheet) <br> • Vital records |
| **Type** | Text |
| **Comments** | Include apartment numbers, etc. A field length of up to 40 characters should be considered. <br><br> If there is a difference between residence address and mailing address, choose residence address. Only use P.O. Box if there is no physical address for the mother. |

| | |
|---|---|
| **Variable Name** | ***Mother's city of residence at pregnancy outcome*** |
| **Definition** | City of address of birth mother's residence at the time of the outcome of the index pregnancy |
| **Justification** | The city at delivery field is often needed when assigning the county of residence. |
| **Source** | Abstracted <br> Derived (from zip code or census tract number) |
| **Location** | • Mother's delivery medical record (face sheet) <br> • Infant's delivery medical record (face sheet, birth certificate worksheet) <br> • Vital records |
| **Type** | Text |
| **Comments** | If there is a difference between residence address and mailing address, choose residence address. Allow for up to 25 characters for city name text fields. A separate city code field may be used to correspond with the city name to facilitate statistical analysis. City coding structures should be compatible with Federal Information Processing Standards (FIPS). |

# Maternal Variables – Core

| | |
|---|---|
| **Variable Name** | ***Mother's county of residence at pregnancy outcome*** |
| **Definition** | County code of birth mother's county of residence at the time of the outcome of the index pregnancy |
| **Justification** | The county of residence, in association with other fields such as plurality and mother's social security number, can be used to avoid duplication of records in the registry. |
| **Source** | Abstracted<br>Derived (from street address and city, zip code, or census tract number) |
| **Location** | • Mother's delivery medical record (face sheet)<br>• Infant's delivery medical record (face sheet, birth certificate worksheet)<br>• Vital records |
| **Type** | Code |
| **Comments** | If there is a difference between residence address and mailing address, choose residence address. County coding schemes should be compatible with standard federal FIPS codes. |

| | |
|---|---|
| **Variable Name** | ***Mother's state of residence at pregnancy outcome*** |
| **Definition** | State in which birth mother resided at the time of the outcome of the index pregnancy |
| **Justification** | The state in which the mother resided is needed if the birth defects program's inclusion criteria include only residents of a certain state. The state of residence, along with other address components, is needed so that researchers and social workers can contact the family, provided a more recent address is not known. |
| **Source** | Abstracted<br>Derived (from other residence information such as city, zip code, and census tract number) |
| **Location** | • Mother's delivery medical record (face sheet)<br>• Infant's delivery medical record (face sheet, birth certificate worksheet)<br>• Vital records |
| **Type** | Standard 2-letter state codes used by US Postal Service |
| **Comments** | If there is a difference between residence address and mailing address, choose residence address. Procedures for reporting information for places outside the US need to be contemplated. |

# Maternal Variables – Core

| | |
|---|---|
| **Variable Name** | ***Mother's zip code of residence at time of pregnancy outcome*** |
| **Definition** | Zip code of birth mother's residence at the time of the outcome of the index pregnancy |
| **Justification** | Cluster investigations are based on a defined diagnosis, geographical area, and time period. Knowing the zip code of residence may allow investigators to determine which cases qualify to be included in cluster investigations. |
| | The zip code, along with other address components, is needed so that researchers, social workers, and others can contact the family, provided a more recent address is not known. |
| **Source** | Abstracted |
| | Derived (from other residence information such as street address and city or census tract number) |
| **Location** | • Mother's delivery medical record (face sheet) |
| | • Infant's delivery medical record (face sheet, birth certificate worksheet) |
| | • Vital records |
| **Type** | Number |
| **Comments** | If there is a difference between residence address and mailing address, choose residence address. This code only applies to United States zip codes and may be the 5-digit or the 9-digit code. |

*Appendix 4.2*

*Descriptions of Recommended Data Variables*

# Appendix 4.2
# Descriptions of Recommended Data Variables

# Appendix 4.2
# Descriptions of Recommended Data Variables

## Format for Variable Descriptions

**Variable Name**      Name of data collection variable

**Definition**         Definition of data collection variable

**Justification**      Reason the birth defects program may want to include variable in its database

**Source**             Where variable comes from – abstracted, derived, created

**Location**           Data sources and location within data sources where variable is most likely to be consistently found

**Type**               How variable should be stored – text, number, date, code (letters and/or numbers), checkbox

**Checks**             Any limits, ranges, or other criteria the variable should meet

**Comments**           Other notes or comments about the variable

**Options**            Recommended options for the variable

# Infant Variables - Recommended

| | |
|---|---|
| **Variable Name** | ***Text description of birth defect*** |
| **Definition** | Description of diagnosis |
| **Justification** | A birth defect may be diagnosed based on more than one procedure or examination. Moreover, two procedures or clinicians may provide different details about the birth defect. For example, one procedure may report that the infant had a myelomeningocele, while a second may mention a lumbar spina bifida. These should all be combined into a single description such as lumbar myelomeningocele. Or one procedure may mention the infant had a cleft lip and palate, while a second notes that the cleft lip was only on the left side of the mouth. These should be combined into something like left cleft lip and palate. |
| | The birth defect description recorded in text format in the data makes it easier to assign disease codes when the medical record is no longer available. |
| **Source** | Abstracted |
| **Location** | • Mother's delivery medical record (prenatal care record, labor and delivery record, prenatal diagnostic procedure reports) |
| | • Infant's medical record (face sheet, admission summary, discharge summary, procedure reports, consultation reports, labor and delivery record, birth certificate worksheet) |
| | • Vital records |
| **Type** | Text |
| **Checks** | Every case should have at least one diagnosis description (unless the birth defects program includes non-malformed controls). A case may have more than one diagnosis description. Every diagnosis description should have a corresponding code and vice versa. |
| **Comments** | See Chapter 5 on Classification and Coding. |

# Infant Variables - Recommended

| | |
|---|---|
| **Variable Name** | ***Date of death*** |
| **Definition** | The date when the death occurred |
| **Justification** | The date of death permits the birth defects program to know that most postnatal procedures will not occur after this date, the exceptions being such procedures as autopsies, cytogenetic analyses, and other laboratory analyses. |
| | The delivery date for a live birth along with the date of death can be used to determine length of survival. Researchers can use this to calculate survival rates for specific lengths of time. |
| **Source** | Abstracted |
| **Location** | • Mother's delivery medical record (prenatal care record, labor and delivery record, prenatal diagnostic procedure reports) |
| | • Infant's medical record (face sheet, admission summary, discharge summary, procedure reports, consultation reports, labor and delivery record) |
| | • Vital records |
| **Type** | Date |
| **Checks** | This field should only be filled out if the pregnancy outcome is "live birth". The date of death should be on or after the date of delivery. |

| | |
|---|---|
| **Variable Name** | ***Birth Length*** |
| **Definition** | Length of newborn at birth |
| **Justification** | In conjunction with gestational age, birth weight, and head circumference, length can be used to assess prenatal growth retardation, a characteristic of fetal alcohol syndrome. However, these circumstances account for only a small subset of cases a birth defects program will collect, and it may not be worth collecting the information on all cases. |
| **Source** | Abstracted |
| **Location** | • Infant's medical record (labor and delivery record) |
| | • Vital records in some states |
| **Type** | Number |
| **Checks** | Edit checks for range and for consistency with gestational age are recommended. |
| **Options** | Best collected as centimeters. |

# Infant Variables - Recommended

| | |
|---|---|
| **Variable Name** | ***Apgar score*** |
| **Definition** | Clinical assessment score of newborn at delivery |
| **Justification** | Apgar scores are a gross measure of early neonatal health. If the scores are low, that means that the newborn had cardiorespiratory problems immediately after delivery. These problems may or may not be related to a birth defect in the infant. For example, some postnatal complications that correlate with low Apgar scores (intestinal perforations, intraventricular hemorrhage) overlap with problems caused by birth defects. |
| **Source** | Abstracted |
| **Location** | • Infant's medical record (labor and delivery record), birth certificate work sheet. <br> • Vital records |
| **Type** | Code |
| **Checks** | Values from 0 through 10 or coded unknown/not applicable. |
| **Comments** | Apgar scores at 1 minute, 5 minutes, and 10 minutes are often available. Vital records generally provide 1- and 5-minute scores, with a change to 5- and 10-minute scores for low 5-minute scores being implemented nationwide. |

| | |
|---|---|
| **Variable Name** | ***Birth order*** |
| **Definition** | Order of delivery for multiple births. |
| **Justification** | Birth order is the order in which infants of a multiple gestation pregnancy are delivered. <br><br> In cases of multiple gestation pregnancies, delivery records might not refer to the infants or fetuses by name but by some other designation such as Twin A and Twin B. This might make it difficult to determine which vital records a particular infant or fetus should be linked to. Vital records may record birth order. Thus birth order might be useful for linkage with vital records in cases of multiple gestation pregnancies. However, other variables such as infant or fetus sex and birth weight might prove as useful for linkage. |
| **Source** | Abstracted |
| **Location** | • Infant's medical record (labor and delivery record), birth certificate worksheet. <br> • Vital records |
| **Type** | Number |
| **Checks** | Must be less than or equal to plurality. |

# Infant Variables - Recommended

| | |
|---|---|
| **Variable Name** | ***Cytogenetic analyses performed*** |
| **Definition** | Whether or not a cytogenetics analysis was performed. |
| **Justification** | Certain structural birth defects are associated with chromosomal abnormalities (Boudjemline et al., 2001; Bullen et al., 2001; Torfs and Christianson, 1998). Structural defects in the presence of a chromosomal abnormality are often considered to be secondary to or the result of the chromosomal abnormality. Such cases may not be considered suitable for research into potential environmental causes of structural defects. And analyses of the proportion of structural defects associated with chromosomal abnormalities often are based on the number of cases where the karyotype is known, because some of the cases without a chromosome analysis may be expected to have chromosomal abnormalities. Infants with certain chromosomal abnormalities also have higher mortality and morbidity than infants without chromosomal abnormalities. Thus it may be important to know whether a chromosomal abnormality is present when deciding whether to refer cases for intervention or prevention activities. |

It is also important to know whether a chromosome analysis was performed at all, even if the results of the analysis are not in the medical record. If time and resources are available, and the chromosome analysis results are considered important, the birth defects program may attempt to track down the results of the analysis. Knowledge that a chromosome analysis was not performed (e.g., because the parents refused) or that a chromosome analysis failed will prevent a birth defects program from wasting resources searching for chromosome analysis results that do not exist.

| | |
|---|---|
| **Source** | Abstracted |
| **Location** | • Infant's medical record (face sheet, admission summary, discharge summary, procedure reports, consultation reports, labor and delivery record) |
| **Type** | Number/Code |
| **Checks** | Must be yes, no, or unknown. |

# Infant Variables - Recommended

| | |
|---|---|
| **Variable Name** | ***Diagnostic tests and procedures performed*** |
| **Definition** | Method used to reach diagnosis. |
| **Justification** | Different procedures can be used in the diagnosis of a birth defect. Moreover, procedures differ in their accuracy and reliability in diagnosing certain birth defects. For example, Down syndrome can usually be considered to be more definite when it is based on a chromosome analysis than on a physical examination of the infant/fetus. Thus it is often not enough to know that a birth defect was mentioned in a medical record; it is important to know how the birth defect diagnosis was made. Moreover, the researcher may only be interested in birth defects identified by particular procedures. For example, researchers may only be interested in cases of a heart defect identified through fetal echocardiography.

If a birth defects program has clearly defined case inclusion criteria (e.g., infants and fetuses with certain birth defects are only included if diagnoses were made by certain procedures), then basic research can be conducted. An example would be for a birth defects program to only include cardiac defects diagnosed by echocardiography, cardiac catheterization, prenatal ultrasound, or autopsy. |
| **Source** | Abstracted |
| **Location** | • Infant's medical record (face sheet, admission summary, discharge summary, procedure reports, consultation reports, labor and delivery record) |
| **Type** | Code |
| **Checks** | Any limits, ranges, or other criteria the data variable should meet. |
| **Comments** | Must develop an appropriate coding structure or select a coding standard such as CPT coding to aid in capturing and tabulating the information. |
| **Options** | May collect primary diagnostic method using a specific hierarchy based on diagnostic accuracy or include multiple procedures fields. |

# Infant Variables - Recommended

| | |
|---|---|
| **Variable Name** | ***Autopsy performed*** |
| **Definition** | Indicates whether an autopsy was conducted. |
| **Justification** | The autopsy is considered one of the more definitive procedures for identifying structural birth defects. |
| | However, even if an autopsy is performed, the autopsy information is not always added to the medical record. As long as a birth defects program has clearly defined case inclusion criteria (e.g., infants and fetuses with certain birth defects are only included if diagnoses were made by certain procedures), then basic research can be conducted. An example of such inclusion criteria would be for a birth defects program to only include cardiac defects diagnosed by echocardiography, cardiac catheterization, prenatal ultrasound, or autopsy. |
| **Source** | Abstracted |
| **Location** | • Infant's medical record (face sheet, admission summary, discharge summary, procedure reports, consultation reports, labor and delivery record) <br> • Death certificate, fetal death report. |
| **Type** | Code |
| **Checks** | Not applicable for live births still living. |
| **Options** | Values would include yes, no, or unknown/not applicable. |

# Infant Variables - Recommended

| | |
|---|---|
| **Variable Name** | ***Physicians of record*** |
| **Definition** | Physician(s) identified as being responsible for admission and discharge records. |
| **Justification** | A birth defects program might want to have information on the physicians of record in order to obtain additional information, to determine if all appropriate referrals were made, to alert physician to need for folic acid recommendations, or to obtain permission to contact the family. |
| **Source** | Abstracted |
| **Location** | • Infant's medical record (face sheet, admission summary, discharge summary, procedure reports, consultation reports, labor and delivery record) <br> • Newborn metabolic screening data <br> • Vital record |
| **Type** | Data are stored as text (names and addresses) |
| **Checks** | N/A |
| **Comments** | To be useful, this information should include name and address for the physician. Allow for 40 characters for entry of each name. There may be interest in collecting multiple physicians and their role, as in pediatrician, obstetrician, or family practice physician to clarify appropriate physician depending upon circumstance. |

# Maternal Variables - Recommended

| | |
|---|---|
| **Variable Name** | ***Date of last menstrual period (LMP)*** |
| **Definition** | First day of last menstrual period |
| **Justification** | Date of LMP, along with date of delivery, can be used to calculate gestational age at delivery. Gestational age at delivery can be used for determining if a spontaneous fetal death or pregnancy termination meets the case definition for the registry. |
| **Source** | Abstracted<br>Derived (see comments) |
| **Location** | • Mother's delivery medical record (prenatal care record, labor and delivery record)<br>• Infant's delivery medical record (prenatal care record, labor and delivery record)<br>• Vital records |
| **Type** | Date |
| **Checks** | The LMP date must be before the date of delivery, estimated date of delivery, prenatal ultrasound date, and prenatal and postnatal procedure dates. The LMP date should not be more than one year before the date of delivery, estimated date of delivery, prenatal ultrasound date, and prenatal procedure dates. |
| **Comments** | If the LMP date is recorded in both the prenatal records and the admission interview, use the LMP date in the prenatal records. If more than one LMP date is found in the prenatal records, record the earliest LMP date in this field. |
| **Options** | See Chapter 3 on Case Definition for further information. |

# Maternal Variables - Recommended

| | |
|---|---|
| **Variable Name** | ***Date of ultrasound*** |
| **Definition** | Date of the earliest identified ultrasound used to assess gestational age |
| **Justification** | Date of ultrasound, along with gestational age at time of ultrasound and delivery date, can be used to calculate gestational age at delivery. |
| **Source** | Abstracted |
| **Location** | • Mother's delivery medical record (prenatal care record, labor and delivery record)<br>• Infant's delivery medical record (prenatal care record, labor and delivery record) |
| **Type** | Date |
| **Checks** | The date of ultrasound field should only be filled in when the gestational age at ultrasound is also known. The ultrasound date must be before or on delivery date and postnatal procedure dates and after the LMP date. The ultrasound date should not be more than 10 months before the date of delivery. |
| **Comments** | Only record information present in the medical record. DO NOT calculate gestational ages or dates.<br><br>If multiple ultrasounds were done to determine gestational age, record the date of the earliest ultrasound. |

# Maternal Variables - Recommended

| | |
|---|---|
| **Variable Name** | ***Gestational age at time of ultrasound*** |
| **Definition** | Gestational age (in weeks) at the time of ultrasound, as estimated by the earliest ultrasound performed |
| **Justification** | Gestational age at ultrasound combined with date of pregnancy outcome can be used for determining if a spontaneous fetal death or pregnancy termination meets the case definition for the registry. |
| | Certain diagnoses are considered birth defects only when the infant is of a particular gestational age. For example, patent ductus arteriosus is common among premature infants and is only considered a birth defect if found in infants born at term. |
| **Source** | Abstracted |
| **Location** | • Mother's delivery medical record (prenatal care record, labor and delivery record) |
| | • Infant's delivery medical record (prenatal care record, labor and delivery record) |
| **Type** | Number |
| **Checks** | This field should only be filled in when the date of ultrasound is also known. The gestational age at ultrasound should range between five menstrual weeks and birth. |
| **Comments** | Only record information present in the medical record. Do not calculate gestational ages or dates. |
| | If multiple ultrasounds were done to determine gestational age, record the date of the earliest ultrasound. |

| | |
|---|---|
| **Variable Name** | ***Mother's medical record number(s)*** |
| **Definition** | Birth mother's medical record number(s) |
| **Justification** | A medical record number allows facilities to retrieve records more easily. |
| **Source** | Abstracted |
| **Location** | • Mother's delivery medical record (face sheet) |
| **Type** | Code |
| **Comments** | The mother may have more than one medical record at a given hospital. Medical record numbers may also be very long. Allow for up to 12 alphanumeric characters for this field. The birth defects program should make certain the computer program allows for entry of the entire medical record number. |

# Maternal Variables - Recommended

| | |
|---|---|
| **Variable Name** | ***Prenatal diagnosis*** |
| **Definition** | The diagnosis made before birth by prenatal diagnostic procedures and tests and neither confirmed nor ruled out by postnatal procedures and tests |
| **Justification** | Prenatal diagnostic procedures used to detect structural birth defects may not be considered to support as definitive a diagnosis as postnatal procedures, and prenatal detection of a birth defect is frequently considered to be tentative. Often physicians will attempt to verify or refine the prenatal diagnosis postnatally, such as through physical examinations, x-rays, or ultrasounds of the live birth or through autopsy of fetal deaths and elective terminations. Thus birth defects program staff should determine whether postnatal procedures and tests were performed and the results of such procedures and tests. However, postnatal confirmation or clarification of prenatally detected birth defects may not always be possible. In such cases the diagnoses identified through prenatal diagnostic procedures and tests are the best information available. Thus it may be useful for the birth defects program to indicate those diagnoses based solely on prenatal procedures. |
| **Source** | Abstracted |
| **Location** | • Mother's delivery medical record (prenatal care record, labor and delivery record, prenatal diagnostic procedure reports)<br>• Infant's medical record (face sheet, admission summary, discharge summary, procedure reports, consultation reports, labor and delivery record, birth certificate worksheet)<br>• Vital records |
| **Type** | Checkbox |
| **Comments** | Prenatal cytogenetic tests may also be considered suspect. Depending on the source of the cell sample used, the sample could have been contaminated by maternal cells. Or, as in the case of chorionic villus sampling, any chromosomal abnormalities identified may be limited to the source of the cell sample and may not affect the fetus. However, prenatal cytogenetic tests are usually considered to be of greater validity than prenatal procedures for identifying structural defects. |

# Maternal Variables - Recommended

| | |
|---|---|
| **Variable Name** | ***Mother's Social Security number*** |
| **Definition** | Birth mother's Social Security number |
| **Justification** | The mother's Social Security number, in association with other fields such as plurality and county of residence, can be used to avoid duplication of records in the registry. |
| | The birth defects program can employ the mother's Social Security number to link to other data sets, such as the Medicaid database. |
| **Source** | Abstracted |
| **Location** | • Mother's delivery medical record (face sheet, prenatal care record) <br> • Infant's medical record (face sheet, prenatal care record, birth certificate worksheet) <br> • Vital records |
| **Type** | Number |
| **Checks** | Medical records may sometimes confuse maternal and paternal information. The mother's and father's Social Security numbers should not be the same. |

| | |
|---|---|
| **Variable Name** | ***Census tract of maternal residence at pregnancy outcome*** |
| **Definition** | Census tract number of birth mother's residence at the time of the outcome of the index pregnancy |
| **Justification** | The geographical areas in most cluster investigations to date have been counties, cities, or particular zip codes. However, in the future, cluster and other investigations may focus on geographical areas defined in other ways. Knowing the census tract number at delivery may allow investigators to determine which cases qualify to be included in such future investigations. |
| **Source** | Derived <br> Abstracted (from vital records files) |
| **Location** | • Vital records |
| **Type** | Number |

# Maternal Variables - Recommended

| | |
|---|---|
| **Variable Name** | *Mother's telephone number* |
| **Definition** | Birth mother's most recent telephone number: area code and telephone number |
| **Justification** | The mother's telephone number is needed so that researchers and social workers can contact the family. |
| **Source** | Abstracted |
| **Location** | • Mother's delivery medical record (face sheet) <br> • Infant's medical record (face sheet) |
| **Type** | Number |
| **Comments** | Enter the area code and seven-digit telephone number. If the area code is not known, enter only the seven-digit telephone number. Note that the telephone number found in a tertiary care facility is more likely to be current than the telephone number at the birth hospital. |

| | |
|---|---|
| **Variable Name** | *Mother's education* |
| **Definition** | Birth mother's highest level of education attained |
| **Justification** | Education can be used as an indicator of socioeconomic status (SES). Collecting maternal education would allow the birth defects program to evaluate its relationship to birth defect risk. |
| **Source** | Abstracted |
| **Location** | • Birth certificate worksheet <br> • Birth certificate, fetal death report |
| **Type** | Code |
| **Checks** | Any limits, ranges, or other criteria the data variable should meet. |
| **Comments** | Since maternal education is not reported consistently in medical records, this information can be obtained more easily by linking to vital record certificates. |
| **Options** | Method for storing the information should permit identifying cases with less than high school, high school, some college, and college graduate. |

# Maternal Variables - Recommended

| | |
|---|---|
| **Variable Name** | ***Prior pregnancy history*** |
| **Definition** | Prior live births and fetal deaths to the birth mother |
| **Justification** | Information can be used to identify women with a significant history of fetal loss or infant death. |
| **Source** | Abstracted |
| **Location** | • Mother's delivery medical record (prenatal care record, labor and delivery record, prenatal diagnostic procedure reports) |
| **Type** | Number |
| **Comments** | This information reflects the number of prior live births and fetal deaths the mother has experienced. Vital record would provide prior live births now living and prior live births now deceased. Medical record can provide parity and gravida. |

| | |
|---|---|
| **Variable Name** | ***Prenatal care*** |
| **Definition** | Information on the initiation and extent of prenatal care |
| **Justification** | Data on prenatal care (such as month of prenatal care and number of prenatal visits), may be useful to a birth defects program. Knowing that the mother did or did not have prenatal care may be useful for birth defects program staff in evaluating other fields on the form. E.g., if it is known that the mother did not have prenatal care, there is less likelihood of finding information on prenatal tests or mother's medical history. And prenatal care may be used as an indication of other factors such as socioeconomic status (SES).<br><br>However, birth defects usually occur before pregnancy is recognized and prenatal care can begin. Furthermore, prenatal care may not be consistently or accurately reported in the medical record – the mother may move or change health care providers or the prenatal care visit information may not be counted consistently. There may be differences of opinion as to what qualifies as a prenatal visit. |
| **Source** | Abstracted<br>Derived |
| **Location** | • Mother's delivery medical record (prenatal care record, labor and delivery record, prenatal diagnostic procedure reports) |
| **Type** | Number |
| **Checks** | Range checks and consistency with woman's age |
| **Comments** | The information to be considered for inclusion would be month prenatal care began and number of prenatal visits. |
| **Options** | The prenatal care information can be summarized using the Kotelchuck or possibly the Kessner Index to standardize the information for more meaningful analysis. |

# Paternal Variables - Recommended

| | |
|---|---|
| **Variable Name** | *Father's date of birth* |
| **Definition** | Date of birth for father |
| **Justification** | The birth defects program may employ the father's date of birth in addition to other fields to link to other data sets, such as Medicaid. Paternal age may be associated with risk for certain birth defects (McIntosh et al., 1995; Olshan et al., 1994). The information can be useful in studies of paternal occupational or exposure cohort studies into associations with birth defects in progeny. |
| | The birth defects program can use the father's date of birth and infant's date of delivery in order to calculate the father's age at delivery. The father's age at delivery can then be used in analyzing birth defect rates by paternal age. |
| **Source** | Abstracted |
| **Location** | • Medical record, birth certificate worksheet<br>• Birth/fetal death, death record |
| **Type** | Text |
| **Checks** | Range checks for father's ages under 12. |

| | |
|---|---|
| **Variable Name** | *Father's name* |
| **Definition** | Name of father |
| **Justification** | The birth defects program may employ the father's name in addition to other fields to link to other data sets, such as vital records or Medicaid. |
| | However, information on the birth father is not consistently found in medical records or vital records. |
| **Source** | Abstracted |
| **Location** | • Medical record and birth record worksheet<br>• Birth/fetal death and death record |
| **Type** | Text |
| **Comments** | The name may be a single field or may be stored as separate first, middle, last, and surname suffix fields. Separate fields greatly facilitate record linkage. Providing 25 character fields for first, middle, and last names should be considered. |

# Paternal Variables - Recommended

| | |
|---|---|
| **Variable Name** | ***Father's education*** |
| **Definition** | Father's highest level of education attained |
| **Justification** | Socioeconomic status (SES) can influence risk of having an infant with a birth defect. Collecting paternal education would allow the birth defects program to evaluate its impact on birth defect risk. |
| **Source** | Abstracted |
| **Location** | • Birth certificate worksheet<br>• Birth/fetal death record |
| **Type** | Code |
| **Checks** | Consistency between father's age and education |
| **Comments** | Since paternal education is not reported consistently in medical records, this information can be obtained more easily by linking to vital record certificates. |
| **Options** | Method for storing the information should permit identifying cases with less than high school, high school, some college, and college graduate. |

| | |
|---|---|
| **Variable Name** | ***Father's race*** |
| **Definition** | Race of father |
| **Justification** | The birth defects program can use the birth father's race in order to evaluate differences in birth defect rates and examine program goals and activities by paternal race. |
| **Source** | Abstracted |
| **Location** | • Birth certificate worksheet<br>• Birth/fetal death record |
| **Type** | Code |
| **Comments** | Racial categories and codes used by birth defects surveillance programs should be compatible with the federal standards in current use for race. |

# Paternal Variables - Recommended

| | |
|---|---|
| **Variable Name** | ***Father's ethnicity*** |
| **Definition** | Ethnicity of father |
| **Justification** | Ethnicity is a designation separate from race. The birth defects program can use the father's ethnicity in order to evaluate differences in birth defect rates or outreach effort goals and activities by father's ethnicity. |
| **Source** | Abstracted |
| **Location** | • Birth certificate worksheet<br>• Birth/fetal death record |
| **Type** | Code |
| **Checks** | Should be valid code. |
| **Comments** | Must develop a code structure that meets registry needs and reflects available data on ethnicity. Should be compatible with federal standard for ethnicity classification. |

| | |
|---|---|
| **Variable Name** | ***Father's Social Security number*** |
| **Definition** | Social Security number of the father |
| **Justification** | The birth defects program can employ the father's Social Security number to link to other data sets, such as the Medicaid database. |
| **Source** | Abstracted |
| **Location** | • Medical record, birth certificate worksheet<br>• Birth/fetal death record |
| **Type** | Number |
| **Checks** | Medical records may sometimes confuse maternal and paternal information. The mother's and father's Social Security numbers should not be the same. |

# Chapter 5

# Classification and Coding

# Table Contents

# Appendices

*This document may be viewed or downloaded from the NBDPN website at:
http://www.nbdpn.org/bdsurveillance.html

# 5.1 Introduction

The National Birth Defects Prevention Network (NBDPN) promotes the use of coded information that is comparable across birth defects programs and methods of case ascertainment, especially for conditions that are reported annually to NBDPN. The proper and accurate coding of diagnostic information is an essential aspect of birth defects surveillance.

A disease classification system plays an important role in the ability of surveillance systems to collect, code, retrieve, and translate information regarding diagnoses and procedures. These activities depend on the ability to assign specific codes to medical information, based on a standardized classification scheme. There are two important ways that classification systems and the coding of birth defects within those systems are central to the surveillance process. Classification and coding rely on a standardized set of rules and procedures for case ascertainment based on medical information, as well as on a standardized way of describing and organizing "cases" based on their clinical conditions.

Coded medical information has become an important part of the health care delivery system. Coding rules, guidelines, and standards have evolved for practically every type of health service encounter. Surveillance systems should understand the various factors that affect the quality of the coding of birth defects and should implement procedures to improve the utility of coding.

In this chapter we discuss disease classification systems (Section 5.2), classification issues that affect surveillance systems (Section 5.3), guidelines for effective coding (Section 5.4), quality issues related to coded data (Section 5.5), and tips and hints to assist with the classification and coding aspects of managing a surveillance system (Section 5.6). References cited in this chapter may be found in Section 5.7.

The two appendices to this document may be viewed or downloaded from the NBDPN website at http://www.nbdpn.org/bdsurveillance.html. Appendix 5.1 is the Texas Disease Index and Appendix 5.2 is the listing of CDC 6-digit codes.

## 5.2  Disease Classification Systems

Over time, a number of systems for classifying pathology, diseases, injuries, and clinical procedures have been developed. This has led to a classification system known as the International Classification of Diseases (ICD). At present, the World Health Organization (WHO) and 10 international centers coordinate classification efforts and promote a standardized classification system for organizing coded data for storage, retrieval, and analysis. Using a standardized system, disease information that is collected by various medical professionals can be compared, grouped, and tabulated for statistical purposes. Definitive information about disease classification in the United States is available from the National Center for Health Statistics (NCHS) of the Centers for Disease Control and Prevention (CDC) (see http://www.cdc.gov/nchs/icd9.htm).

The ninth revision of the International Classification of Diseases (ICD-9) was in use from 1979 to 1998. The United States uses the standard ICD version for coding deaths and in 1979 developed a 'clinical modification' for use in hospitals (i.e., ICD-9-CM). The clinical modification of the ICD-9 expanded the general categories, permitted greater detail and description, and included codes for clinical procedures. A large body of ICD-CM coding guidelines and rules was also developed. Since most of the information about birth defects comes from clinical records, the discussion below refers primarily to ICD-9-CM. However, comments regarding structure and organization are applicable to ICD-9 codes as well.

In 1999, the tenth revision (ICD-10) became operational for coding causes of death on death certificates. Although the classification structure is basically unchanged, ICD-10 reflects a significant revision from ICD-9. The codes are alphanumeric instead of numeric (as was the case in ICD-9), there are more general categories, and the codes are described in greater detail than in earlier versions. As of 2002, NCHS is developing the clinical modification ICD-10-CM. (This reference manual will not discuss ICD-10-CM until it is implemented).

Of importance to birth defects surveillance is the fact that, although ICD-9-CM is an acknowledged standard for coding medical information, it is not optimal for the level of detail required for coding many birth defects.

In 1979 the British Paediatric Association (BPA) developed a classification of diseases by modifying ICD-9-CM (British). In 1983, staff in CDC's birth defects branch modified the BPA coding system and developed a classification system specific to birth defects coding. The 6-digit CDC code is a classification system that allows coding of more detailed descriptions of birth defects and related conditions (see Appendix 5.2 for a complete listing of the 6-digit CDC codes).

### 5.2.1  Description and Format

The ICD-9-CM and the 6-digit CDC coding systems are divided into general categories that include body systems, medical conditions, and other health-related issues. The codes are hierarchical and expand to reflect specific conditions within a general category. Each code category is populated with specific diseases and related conditions. In ICD-9-CM the majority of the codes used in birth defects programs is between the code categories 740 and 759, which come under the general heading of 'congenital anomalies'. The ICD-9-CM and the 6-digit CDC coding systems utilize a similar format for categorizing disease. ICD-9-CM utilizes up to five digits, while the CDC coding system utilizes six.

## 5.2.2 ICD-9-CM and the 6-digit CDC Code – A Comparison

In most cases, the first four digits of the 6-digit CDC code are identical to the first four digits of the ICD-9-CM code. This enables birth defects programs to utilize the coded data collected from hospital data sets, while at the same enhancing the level of coding detail for birth defects program use. Since, the 6-digit CDC code usually collapses into the ICD-9-CM at the fourth-digit level, programs that use ICD-9-CM codes have data that are comparable between states.

The most significant difference between ICD-9-CM and the 6-digit CDC code is reflected in the level of detail indicated by the sixth digit. The sixth digit can be used to indicate one of three aspects of the defect:

> *Laterality of the defect*
>> .001  Left side only
>> .002  Right side only
>> .003  Unilateral, unknown which side
>> .004  Bilateral; both sides

> *Greater specificity for a particular defect*
>> .005  Example: 756.615 Diaphragmatic hernia (Bochdalek)
>> .006  Example: 756.616 Diaphragmatic hernia (Morgagni)
>> .007  Example: 756.617 Hemidiaphragm

> *Incomplete confirmation of a defect (includes possible or probable or only diagnosed prenatally)*
>> .008  Example: 745.498 Probable Ventricular Septal Defect (VSD)

## 5.3  Classification Issues That Affect Surveillance Systems

It is important to recognize that there are advantages and disadvantages associated with both the ICD-9-CM and 6-digit CDC coding systems. Programs need to be aware of the ways in which these may affect data quality and other surveillance activities.

➢ *Any coding system is limited to the number of literal descriptions assigned to a code in the system*. In other words, there may be synonyms for one birth defect, or many related birth defects may be assigned to one code. The disease index provided in Appendix 5.1 is a tool that can be used to assist with coding. This alphabetic cross-linked index of birth defects and corresponding 6-digit CDC codes was developed by the Texas Birth Defects Monitoring Program. The cross-linked index is an expansion of the ICD-9-CM Congenital Anomalies category 740-759.9 and includes multiple disease descriptions, synonyms, and other descriptive terms that are used to describe birth defects.

➢ *Classification systems provide a framework for coding but often do not provide compatible definitions of diagnoses*. Clinical case definitions and case definitions used for public health surveillance are not always the same. Surveillance systems must specify how clinical documentation should be used to determine the appropriate disease code. See Chapter 3 on Case Definition for a discussion of ways to determine how birth defects should be coded.

➢ *Diagnostic categories are not consistent in the amount of detail they provide, nor are they always clear.* For instance, the chromosomal anomaly category (758) is very general. The musculoskeletal system (754-756) is not well-defined. Additionally, all birth defects are not identified with an explicit code, so there can be questions about how to code a particular defect or whether it should be coded at all.

➢ *A single ICD-9-CM code may be used to describe several different defects*. This may make it difficult to use the code to recover specific information. For example, codes such as those listed below present challenges because of the potential heterogeneity of the defects included under a given code:

- 742.2  Reduction deformities of brain (includes holoprosencephaly and absent corpus callosum)
- 747.21  Anomalies of aortic arch (includes overriding aorta and double aortic arch)
- 753.0  Renal agenesis and dysgenesis (includes absent kidney and hypoplasia of kidney)
- 756.0  Anomalies of skull and face bones (includes hypertelorism and craniosynostosis)
- 756.79  Other congenital anomalies of abdominal wall (includes gastroschisis and omphalocele)

➢ *ICD-9-CM codes do not reflect the status of the diagnosis*. For example, a condition may be possible or probable. This is problematic when birth defects are reported to the surveillance system in coded format, or when programs use the hospital disease index in case finding.

➢ *How information is coded in an administrative database (e.g., hospital disease index, hospital discharge data, Medicaid data) is determined by the methods used to assign codes and by the objectives of those who maintain the database*. In other words, code use is defined by the "business operations" of the facility or organization doing the coding. For example, the ICD-9-CM classification system is used primarily in hospitals and other care settings to comply with federal financial justification for payment. Coding decisions made by someone with that goal in mind could be different

from those made by someone coding for a surveillance system.

➤ *Professional disease coding training and courses for ICD-9-CM are beneficial in providing a good foundation for training staff regardless of the surveillance approach being used (i.e., active or passive case ascertainment)*. Information on such courses is available from the American Health Information Management Association (http://www.ahima.org).

# 5.4  Guidelines for Effective Coding

As noted earlier, the primary goal in coding information is to provide accurate, consistent, and concise representation of that information. Coded diagnostic information is easier to analyze, compare, retrieve, and store. All of these attributes promote the use and dissemination of information between systems. The use of computer technology and the development of particularly large databases have accelerated the demand for coded information. The standardization of information that is translated into a code or discrete data element is one of the objectives of the Health Insurance Portability and Accountability Act of 1996 (HIPAA). For a thorough discussion of birth defects coding, see Rasmussen and Moore (2001).

Programs should:

➤ **Develop** well-defined surveillance case definitions. This includes identifying the characteristics of eligibility (e.g., demographics, pregnancy outcome, gestational age), and specific birth defects or diagnosis. These issues are discussed in Chapter 3 on Case Definition.

➤ **Understand** that the disease classification system and associated coding guidelines are developed to standardize results and assist in decision-making. The coding rules for ICD-9-CM as used by hospitals are established at the federal level through a set of guidelines administered, maintained, and updated by NCHS. To comply with these federal standards, a hospital coder may be required to use codes that differ from those used by a surveillance system coder. The 6-digit CDC code is supported by a body of guidelines and procedures that specifically address issues in assigning codes to birth defects. Coding rules for the 6-digit CDC code are detailed and have many exceptions. For example, when using the 6-digit CDC code, there may be exceptions in the laterality rule (i.e., does not apply to all diagnoses).

➤ **Adapt** surveillance procedures and the database to disease code changes as they occur. ICD-9-CM codes and code definitions are subject to rule changes, additions, deletions, and edits. ICD-9-CM changes are usually timed to coincide with the beginning of the federal fiscal year. It is essential for programs that use administrative databases to be aware of these code changes.

➤ **Track** disease code changes. Consider adding a date field to each disease code listed in the database. Disease codes are added, deleted, or edited by the authoritative agency, usually on an annual basis. Any code assignment change may affect statistical analysis or other evaluation activities. Tracking disease code changes will be an essential task when ICD-10-CM replaces ICD-9-CM.

➤ **Assign** a disease code to each diagnosis that is reportable to the program. This facilitates building a database of eligible disease codes (conditions), which can be incorporated into abstracting software (e.g., drop-down windows) and used to develop queries and generate lists.

➤ **Identify** the disease classification system that is to be used. Some programs may use more than one disease classification system.

    Examples:

    • An active case ascertainment system might only use the 6-digit CDC code.

- A passive case ascertainment system might only use the ICD-9-CM classification system.

- A passive case ascertainment system might use the 6-digit CDC code if the program receives case reports in a descriptive or literal format and if surveillance staff assign codes.

- A passive case ascertainment system might use ICD-9-CM for case reports that are submitted to the program, but might use the 6-digit CDC code when staff actively review medical charts or for special projects.

➢ **Promote** the use of the 6-digit CDC code where possible. Because the CDC code conveys greater detail, surveillance systems should ideally incorporate this coding system into regular program operations. This may be easier for active ascertainment systems, as passive case ascertainment systems are often limited to the standard classification system in use at hospitals (i.e., ICD-9-CM). However, in order to promote consistency, accuracy, completeness, and comparability across birth defects programs, passive case ascertainment should use the 6-digit CDC code whenever possible.

➢ **Use** the NBDPN Abstractor's Instructions cited in Chapter 3 on Case Definition. This tool should be used as a reference for the birth defects that are central to the NBDPN. It describes the diagnosis and identifies the appropriate disease code.

➢ **Use** technical reference materials. For example, *The International System for Human Cytogenetic Nomanclature (ISCN)* is the definitive guide to understanding the classification system used in cytogenetics (Mitelman, 1995). The text provides information on definitions, on how to read and understand karyotypes, and on other technologies used in laboratory analysis. Surveillance systems can use the *ISCN* as a tool to assist in assigning a disease code to a case with a chromosomal anomaly.

➢ **Use** clinicians for advice on understanding medical conditions and for providing guidance on assignment of disease codes.

➢ **Develop** coding procedures for abstractors, especially as relates to standardized methods for translating medical information into a disease code. Document decision items that result from coding discussions through the use of a decision log or similar record-keeping system. Surveillance systems that are research based may require a different set of procedures than a surveillance system that is focused on providing services. The NBDPN Abstractor's Instructions cited in Chapter 3 on Case Definition provide a good foundation.

# 5.5 Coded Data Quality Issues

Many factors can affect the quality of coded data. As mentioned earlier, any disease coding system has limitations. Additionally, the translation of a medical diagnosis into a disease code requires interpretation and judgment. Programs can improve the quality of coded data by considering the following recommendations.

Programs should:

➢ **Promote** coding to the highest degree of accuracy, completeness, and consistency as required by the surveillance system and as recommended by the NBDPN.

➢ **Develop** methods to identify situations that may result in inconclusive or incomplete diagnoses. This is particularly important for programs that work with or receive diagnosis information in coded format. Programs can use length-of-stay patterns, type of diagnosis, and type of data source (e.g., prenatal diagnosis center) to determine whether follow-up is necessary. For example, a chromosomal anomaly diagnosed during the newborn period may be a 'suspect' condition at discharge, pending receipt of laboratory results.

➢ **Code** all individual defects associated with a chromosomal anomaly, syndrome, or association, unless a coding rule or the NBDPN Abstractor's Instructions cited in Chapter 3 on Case Definition specify otherwise (see next recommendation). Code the major chromosomal anomaly or syndrome as well. Some of the most frequently diagnosed syndromes are listed in the category 759.8 in the 6-digit CDC code (see Appendix 5.2).

- Chromosomal anomalies should be coded to the highest degree of detail that is provided by the karyotype.

- Birth defects that are components of syndromes identified by 759.8x should be coded separately.

➢ **Identify** those birth defects that are exceptions to the 'code all defects' rule outlined above. For some diagnoses, all birth defects related to the condition may not need to be coded. Refer to the NBDPN Abstractor's Instructions cited in Chapter 3 on Case Definition for a listing and description of these conditions. Develop methods to query the database to find potentially "extra" disease codes. This often occurs with passive case ascertainment using multiple data sources. Some sources may report the major birth defect, while others may report each defect within the major diagnosis.

➢ **Code** at the most specific level possible. For example, if the specific heart defect is known, it is essential to list the specific defect rather than a more general description such as 'congenital heart disease'. Passive case ascertainment systems may find it useful to develop data quality audits to identify diagnoses that frequently are assigned general or non-specific codes and that may merit follow-up.

➢ **Develop** computer edit checks to identify problems with code use. For example, some conditions should be combined under a single code. These include spina bifida and hydrocephalus, imperforate anus and anal fistula, esophageal atresia and tracheoesophageal fistula, tetralogy of fallot, and cleft lip and palate. Edit checks can also be developed for gender-specific conditions and for conditions that may also be acquired (e.g., hydrocephalus, skeletal deformations). Edit checks can further be used to identify codes for defects that should not be counted due to gestational age, birth weight, or other established eligibility criteria.

➢ **Develop** methods for identifying general or non-specific codes, miscodes, inappropriate or redundant codes, or unusual combinations of coded data in a case abstract or case record.

➢ **Evaluate** the accuracy and consistency of code assignment. Conduct evaluations to determine the level of agreement in code assignment among program staff, as well as between staff and acute care coders in hospitals. This is particularly effective in identifying differences that result due to federal ICD-9-CM coding guidelines. Identify problem areas and implement quality control procedures as necessary.

➢ **Develop** coding procedures documentation especially regarding decision items, discussion points, or code assignments. Identify implementation dates.

# 5.6  Tips and Hints

➢ *Coded data can be used to enhance surveillance capability, as they are easily manipulated and queried in a database.* For example:

  • Birth defects case records that have multiple disease codes can be identified and investigated further to determine whether an underlying condition or syndrome is present.

  • Birth defects codes that are included or excluded due to specific criteria can be identified and flagged.

➢ *Administrative databases, especially hospital discharge data, use the ICD-9-CM coding system.* Discharge data can be used for specific screening purposes. For example:

  • Maternal pregnancy disease codes may identify potential birth defects cases, especially if the pregnancy results in a fetal demise.

  • Possible cases of birth defects can be queried using disease codes for prematurity, low birth weight, stillbirths, etc.

➢ *Some programs may find it helpful to retain the complete descriptive text of the birth defect.* As previously stated, disease coding systems have limitations. While birth defects are translated to the most accurate disease code, the code may not be precise enough in describing the birth defect.

➢ *Patterns of disease code assignment for particular birth defects may vary between hospital disease coders.* During case finding and abstracting and when reviewing medical records, it is helpful to be observant of coding patterns and inclinations. In many instances, disease codes are listed in the medical records, which helps with these informal assessments.

# 5.7 References

American Health Information Management Association. The lead organization in the development of professional practice standards for health information management, including training in ICD-9-CM. http://www.ahima.org

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Centers for Disease Control and Prevention, Metropolitan Atlanta Congenital Defects Program. *1998 Procedures Manual, Appendices A and B, CDC 6-digit code list*. January 1998. For copies send request by e-mail to macdp@cdc.gov or fax 404-498-3040.

Mitelman FS, ed. *International System for Human Cytogenetic Nomenclature.* Farmington, CT: Karger Publishers, Inc.; 1995.

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**Appendix 5.1**
**Texas Disease Index**

The Texas Birth Defects Monitoring Division (TBDMD) created the Texas Disease Index to be used in conjunction with the six-digit codes for reportable birth defects developed by the National Center for Birth Defects and Developmental Disabilities of the Centers for Disease Control and Prevention (CDC). The six-digit birth defect codes, commonly called the BPA code, were developed based on the British Pediatric Association (BPA) Classification of Diseases (1979) and the World Health Organization's International Classification of Diseases, 9th Revision, Clinical Modification (ICD-9-CM) (1979).

The Texas Disease Index was developed for use by the TBDMD, which utilizes active case ascertainment. In addition to being useful to other surveillance programs that carry out active case ascertainment, it is also a valuable resource for systems that have passive case ascertainment based on reporting by standard ICD-9-CM codes.

It should be noted that the TBDMD made some modifications to the BPA code list. Therefore the Texas Disease Index may deviate slightly from the six-digit CDC code list used by other active case ascertainment surveillance programs, which is included as an appendix to these guidelines by reference to the website. Most of the modifications relate to birth defects that were not listed explicitly in the original BPA codes. These additional birth defects have been reviewed by various TBDMD staff, including two clinical geneticists, and appropriate BPA codes have been assigned to them.

Note that for ease of use a diagnosis may be listed in more than one format in this index. For example, 'absent eye' may be found under 'absent, eye' or 'eye, absent.

The TBDMD revises this index periodically, indicating the revision date on each page. New revisions will be made available through the surveillance guidelines and standards webpage.

This document may be viewed or downloaded at the NBDPN website at:
http://www.nbdpn.org/bdsurveillance.html

**References**

British Paediatric Association (BPA). *Classification of Diseases.* London, England: British Paediatric Association; 1979.

National Center for Health Statistics (NCHS). *International Classification of Diseases, 9th Revision, Clinical Modification.* Washington, DC: National Center for Health Statistics and the Health Care Financing Agency; 1998.

**-A-**

Aarskog syndrome - 759.800
Abdominal
      cyst NOS - 759.990
      mass NOS - 759.990
Abdominal wall
      benign neoplasm - # 216.500
      other and unspecified anomalies - 756.790
Abduction
      foot - L 754.690
      hip - x
Aberrant
      innominate artery - L 747.640
      subclavian artery - L 747.640
Ablepharon - L * 743.630
Absent - see also agenesis, atresia
      adrenal gland - L 759.100
      alimentary tract, NOS (complete or partial) - 751.800
      anus
            with fistula - 751.230
            without fistula - 751.240
      aorta - 747.200
      aortic valve - 746.480
      appendix - 751.200
      arm - L 755.200
      auditory canal (without hypoplastic pinna) - L 744.000
      auricle - L 744.010
      bladder - 753.800
      brain - 740.000
      breast
            nipple absent - L 757.600
            nipple present - L 757.610
      broad ligament - L 752.100
      bronchus - L 748.350
      carotid artery - L 747.640
      cervix (genital) - 752.400
      clitoris - * 752.450
      colon - 751.200
      diaphragm - L 756.600
      digestive system, NOS (complete or partial) - 751.800
      digit, NOS - L 755.440
      duodenum - 751.100
      ear - L 744.010
      ear canal (without hypoplastic pinna) - L 744.000
      external genitalia
            female - * 752.440
            male - 752.880
      eye - L 743.000
      eyebrow - L 744.880
      eyelash - L * 743.630
      eyelid - L * 743.630
      face - L 744.880
      fallopian tube - L 752.100
      femur (total or partial)
            only - L 755.320

with absent tibia and fibula (total or partial)- L 755.310
with absent tibia, fibula, and foot - L 755.300
fibula
only (total or partial) - L 755.366
with absent femur (total or partial) and tibia - L 755.310
with absent femur (total or partial), tibia, and foot - L 755.300
with absent tibia - L 755.320
with absent tibia and foot - L 755.330
finger
fifth (with or without fourth) - L 755.270
first (thumb) - L 755.260
first (thumb) and absent radius (total or partial) - L 755.260
NOS - L 755.240
third (with or without second, fourth) - L 755.250
with absent forearm long bone - L 755.265
fontanelle - # 754.040
foot
only - L 755.340
with absent femur (total or partial), tibia, and fibula - L 755.300
with absent lower leg - L 755.330
with absent tibia and fibula (total or partial) - L 755.330
forearm
long bone with absent fingers - L 755.265
only - L 755.220
with absent hand - L 755.230
with absent upper arm - L 755.210
foreskin - 752.860
genitalia (sex unknown) - * 752.790
hand
only - L 755.240
with absent forearm - L 755.230
with absent humerus (total or partial), radius, and ulna - L 755.200
with absent radius and ulna (total or partial) - L 755.230
head - 740.080
humerus (total or partial)
only - L 755.220
with absent radius, and ulna - L 755.210
with absent radius, ulna, and hand - L 755.200
ileum - 751.120
intestine
large - 751.200
small - 751.190
small, with fistula - 751.195
iris - L 743.420
jejunum - 751.110
kidney
bilateral - 753.000
NOS - 753.009
unilateral - L 753.010
lacrimal apparatus - L 743.640
leg - L 755.300
lens - L 743.300
limb, NOS - L 755.400
liver, total or partial - 751.600
long bone leg with absent toe - L 755.360
lower leg
only - L 755.320

with absent foot - L 755.330
with absent thigh - L 755.310
lung - L 748.500
meatus (external auditory, ear) - L 744.000
mitral valve - 746.505
muscle - L 756.810
nail - L 757.500
nares - 748.100
nasal septum - # 748.180
neck - # 744.900
nipple
only - L 757.630
with absent breast - L 757.600
nose - 748.100
olfactory nerve - 742.270
ovary - L 752.000
palate
hard - 749.030
NOS - 749.090
soft - 749.070
pancreas - 751.700
patella - L 755.647
penis - 752.850
phalange (isolated)
finger - L 755.240
toe - L 755.340
pinna (ear) - L 744.010
pulmonary arteriovenous - L 747.340
pulmonary artery - L 747.300
pulmonary valve - 746.000
punctum lacrimale - L 743.640
radius
only (total or partial) - L 755.260
with absent humerus (total or partial) and ulna - L 755.210
with absent humerus (total or partial), ulna, and hand - L 755.200
with absent thumb - L 755.260
with absent ulna - L 755.220
rectum
with fistula - 751.210
without fistula - 751.220
renal artery - L 747.610
respiratory organ NOS - 748.900
rib - L 756.300
right superior vena cava - x
septum between aorta and pulmonary artery - 745.000
septum pellucidum - 742.210
skin - 757.395
spleen - 759.000
sternocleidomastoid muscule - L 754.100
sternum - 756.350
stomach
with absent GI tract - 750.780
with rest of GI tract intact - 750.700
superior vena cava, right - x
tarsal bones - L 755.340
tendon - L 756.820
testicle - L 752.800

thigh and lower leg - L 755.310
thymus - * 759.240
tibia
  only (total or partial) - L 755.365
  with absent femur (total or partial) and fibula (total or partial)- L 755.310
  with absent femur (total or partial), fibula, and foot - L 755.300
  with absent fibula - L 755.320
  with absent fibula (total or partial) and foot - L 755.330
  with absent first toe (with or without second toe) - L 755.365
toe
  fifth (with or without fourth) - L 755.366
  first toe (with or without second toe) - L 755.365
  first toe (with or without second toe) and tibia (total or partial) - L 755.365
  NOS - L 755.340
  third (with or without second, fourth)- L 755.350
  with absent long bone leg - L 755.360
tongue - 750.100
ulna
  only (total or partial) - L 755.270
  with absent humerus (total or partial) and radius - L 755.210
  with absent humerus (total or partial), radius, and hand - L 755.200
  with absent radius - L 755.220
upper arm
  only - L 755.220
  with absent forearm - L 755.210
ureter - L 753.400
urethra - 753.800
uterus - 752.300
uvula - 749.080
vagina (complete or partial) - 752.410
vena cava (except left superior) - 747.480
vulva - * 752.440
Acardiac twins - 759.480
Accessory - see also extra
  adrenal gland - L 759.120
  auricle - L # 744.100
  carpal bone - L 755.525
  breast (with accessory nipple) - L 757.620
  digit - see polydactyly
  finger - see polydactyly
  kidney - L 753.300
  lung lobe - L 748.620
  nipple
    only - L # 757.650
    with accessory breast - L 757.620
  nose - 748.110
  ovary - L 752.020
  pancreas - 751.710
  spleen - 759.040
  toe - see polydactyly
  ureter - L 753.410
Achalasia of cardia - 750.720
Achilles tendon, short - L 754.720
Achondrogenesis
  type I - 756.480
  type II - 756.480
Achondroplastic dwarfism - 756.430

Acne, neonatal - x
Acrania - 740.010
Acrocallosal syndrome - 759.890
Acrocephalosyndactyly
       NOS - 756.050
       other specified - 756.057
       type I - 756.055
       type II - 756.055
       type III - 756.056
Acrocephaly - 754.080
Acrodactylia
       finger - L # 755.500
       toe - L # 755.600
Acyanotic congenital heart disease - 746.920
Adams-Oliver syndrome - 759.840
Adduction foot - L 754.590
Adductus
       metatarsus - L # 754.520
Adhesion of omentum and peritoneum - 751.420
Adrenal gland
       absent - L 759.100
       accessory - L 759.120
       dysgenesis - L 759.180
       ectopic - L 759.130
       enlarged - L 759.180
       fused  - L 759.180
       hyperplasia, congenital
              classical (salt) water - # 255.200
              classical (simple virilizer) - # 255.210
              NOS - # 255.290
              other than 21-OHP deficiency - # 255.240
       hypoplasia - L 759.110
       other specified anomalies - L 759.180
       unspecified anomalies - L 759.190
Adrenogenital syndrome - # 255.290
Aganglionosis of intestine
       beyond the rectum - 751.310
       involving no more than the anal sphincter and the rectum - 751.320
       total - 751.300
Agenesis - see also absent
       bile duct - 751.650
       cervix (genital) - 752.400
       gallbladder - 751.630
       hepatic duct - 751.650
       kidney
              bilateral - 753.000
              NOS - 753.009
              unilateral - L 753.010
       liver, total or partial - 751.600
       lung - L 748.500
       nose - 748.100
       ovary - L 752.000
       pancreas - 751.700
       uterus - 752.300
       vagina (complete or partial) - 752.410
       vertebrae
              cervical - 756.146

lumbar - 756.166
                    sacral - 756.170
                    thoracic - 756.156
Aglossia - 750.100
Agnathia - * 524.000
Agnathia formation complex - 759.800
Agyria - 742.240
Aicardi syndrome - 759.890
Alae nasae hypoplasia - # 748.180
Alagille syndrome - 759.870
Albers-Schonberg syndrome - 756.540
Albinism - # 270.200
Albright-McCune-Sternberg syndrome - 756.510
Alimentary tract
          absent (complete or partial) - 751.800
          duplication - 751.810
          ectopic - 751.820
          obstruction, NOS - 752.900
          other specified anomalies - 751.880
          unspecified anomalies - 751.900
Almond shaped eye - L # 743.800
Alopecia - 757.400
Alport syndrome - 759.870
Ambiguous genitalia - * 752.790
Amelia
          arm - L 755.200
          leg - L 755.300
          limb, NOS - L 755.400
Amniotic
          bands - # 658.800
          cyst - # 658.800
Amputation, NOS
          arm - L 755.285
          leg - L 755.385
          limb, NOS - L 755.420
Amsterdam dwarf - 759.820
Amyelia - 742.500
Amyoplasia congenita - 756.840
Amyotrophia congenital - 756.840
Anasarca - # 778.000
Androgen insensitivity syndrome - 257.800
Anencephaly - 740.020
          other - 740.080
Aneurysm
          aorta - 747.270
          arteriovenous (brain) - L 747.800
          atrial septum - x
          pulmonary artery - 747.330
          sinus of Valsalva - 747.240
          tricuspid valve - 746.100
Angelman syndrome - 759.890
Angulation of tibia - L * 755.630
Aniridia - L 743.420
Anisocoria - L 743.440
Ankle
          anomalies - L 755.620
          other specified deformities - L 754.780

Ankyloblepharon - L * 743.630
Ankyloglossia - # 750.000
Annular pancreas - 751.720
Anomalous portal vein termination - 747.440
Anomalous pulmonary venous return
        partial - 747.430
        total - 747.420
        total/partial not specified - 747.480
Anonychia - L 757.500
Anophthlmos - L 743.000
Anotia - L 744.010
Anovaginal fistula - 752.420
Anterior
        frenulum (tongue, lingual) - # 750.000
        segment of eye
                other specified colobomas - L 743.480
                other specified anomalies - L 743.480
                unspecified anomalies - L 743.490
        urethral valve - 753.620
Anteversion of femur - L 755.650
Antimongolian syndrome - 758.300
Antimongoloid slant to eyes - L # 743.800
Anus/anal
        absent
                with fistula - 751.230
                without fistula - 751.240
        atresia
                with fistula - 751.230
                without fistula - 751.240
        benign neoplasm - # 216.500
        displaced - 751.530
        duplication - 751.500
        dysgenesis with fistula - 751.230
        dysgenesis without fistula - 751.240
        ectopic - 751.530
        fissure - x
        fistula - 751.540
        imperforate
                with fistula - 751.230
                without fistula - 751.240
        stenosis
                with fistula - 751.230
                without fistula - 751.240
Aorta/Aortic
        artery
                absent - 747.200
                absent septum between pulmonary artery and - 745.000
                aneurysm - 747.270
                atresia - 747.200
                coarctation
                        distal - 747.110
                        juxtaductal - 747.190
                        postductal - 747.110
                        preductal - 747.100
                        proximal - 747.100
                        unspecified - 747.190
                collateral vessel involving - 747.280

dextroposition - 747.260
dilatation - 747.270
double arch - 747.250
enlarged - 747.270
hypoplasia - 747.210
interrupted arch - 747.215
large - 747.270
malaligned - 747.260
narrow - 747.210
other specified anomalies - 747.280
overriding - 747.260
pseudocoarctation - 747.280
right arch - 747.230
small - 747.210
supra-aortic stenosis - 747.220
supravalvular - 747.220
unspecified - 747.290
NOS
septal defect - 745.010
stenosis - 746.300
subvalvular stenosis - 746.300
valve
abnormal - 746.490
absent - 746.480
atresia - 746.480
bicuspid - * 746.400
dysmorphic - 746.480
dysplastic - 746.480
hypoplastic - 746.480
incompetence - * 746.400
insufficiency - * 746.400
other specified - 746.480
quadricuspid - 746.480
regurgitation - * 746.400
small - 746.300
stenosis - 746.300
thickened - 746.480
unspecified - 746.490
Aortic annulus - see aortic valve
Aortopulmonary window - 745.010
Apert syndrome - 756.055
Aphakia - L 743.300
Aplasia - see also absent, agenesis
cutis
not involving scalp - 757.395
scalp - 757.800
eye - L 743.100
penis - 752.850
red cell - # 284.000
scrotum - L * 752.810
testicle - L * 752.810
Appendix
absent - 751.200
atresia - 751.200
duplication - 751.500
stenosis - 751.200
testicle - L 752.870

transposition - 751.510
Aqueductal stenosis (without spina bifida) - 742.300
Aqueduct of Sylvius anomalies without spina bifida - 742.300
Arachnodactyly
     finger - L # 755.500
     toe - L # 755.600
Arachnoid cyst - x
Arm
     absent - L 755.200
     amelia - L 755.200
     amputation, NOS - L 755.285
     benign neoplasm - L # 216.600
     hyperextensibility - L 755.580
     hypomelia - L 755.585
     hypoplasia - L 755.585
     intercalary reduction defect - L 755.210
     long - x
     longitudinal reduction defect
          NOS - L 755.265
          postaxial - L 755.270
          preaxial - L 755.260
     other anomalies (whole) - L 755.560
     other specified anomalies - L 755.580
     other specified reduction defect - L 755.280
     phocomelia - L 755.210
     positional deformity - L 755.580
     short - L 755.580
     transverse reduction defect, NOS - L 755.285
     unspecified anomalies - L 755.590
     unspecified reduction defect - L 755.290
Arnold-Chiari malformation
     with spina bifida - 741.010
     without spina bifida - 742.480
Arrhinencephaly - 742.270
Arrhythmias, cardiac, NOS - 427.900
Arteriovenous malformation
     brain - L 747.800
     peripheral - L 747.620
Arthrogryposis multiplex congenita - L 755.800
Ascites, congenital - # 778.000
Asphyxiating thoracic dystrophy - 756.400
Asplenia - * 759.000
Association - see syndrome
Astragaloscaphoid synostosis - L 755.620
Asymmetry
     brain - x
     calvarium - 754.055
     chest - 754.820
     crying facies - L 351.000
     ears - x
     eyes - x
     face - 754.000
     gluteal cleft - x
     head - 754.055
     jaw - * 756.080
     mouth - L 744.880
     nipples - # 757.680

nose - # 748.180
skull - 754.055
Atelomyelia - 742.510
Atresia
    anus
        with fistula - 751.230
        without fistula - 751.240
    aorta - 747.200
    aortic valve - 746.480
    appendix - 751.200
    bile duct - 751.650
    biliary - 751.650
    bladder neck - 753.610
        other and unspecified - 753.690
    cervix (genital) - 752.400
    choanal - L 748.000
    colon - 751.200
    duodenum - 751.100
    esophageal
        without tracheoesophageal fistula - 750.300
        with tracheoesophageal fistula - 750.310
    hepatic duct - 751.650
    ileum - 751.120
    intestine
        large - 751.200
        small - 751.190
        small, with fistula - 751.195
    jejunum - 751.110
    lung - L 748.500
    meatus (urethral, urinary) - 753.630
    mitral valve - 746.505
    nares - L 748.000
    piriform aperature - L 748.000
    pulmonary
        artery
            without septal defect - L 747.300
            with septal defect - L 747.310
        NOS (heart) - 746.995
        valve - 746.000
        vein - 747.480
    pyloric - 751.100
    rectum
        with fistula - 751.210
        without fistula - 751.220
    trachea - 748.330
    tricuspid valve - 746.100
    ureter - L 753.210
    urethra
        anterior - 753.620
        other and unspecified - 753.690
    urinary meatus - 753.630
    vagina (complete or partial) - 752.410
    vas deferens - L 752.830
Atrioventricular canal
    common - * 745.630
    common, with VSD - * 745.620
    complete - * 745.630

complete, with VSD - * 745.620
Atriventricular septal defect - see atriventricular canal
Atrioventricular valve
       left - see mitral valve
       right - see tricuspid valve
       single - 746.900
            insufficiency - 746.900
            regurgitation - 746.900
Atrium/atrial
       common - 745.610
       dilatation - x
       enlarged - x
       hypoplastic - 746.887
       inversion - 746.880
       other defects - 746.887
       septal defect
            aneurysm - x
            fenestrated - 745.510
            fossa ovalis - 745.510
            NOS - * 745.590
            ostium primum - * 745.600
            ostium secundum - 745.510
            other specified - 745.580
            primum - * 745.600
            secundum - 745.510
            vs PFO - * 745.590
       single - 745.610
Atrophy
       cerebellar - 742.230
       cerebral - 742.480
       cortical (brain) - 742.480
       muscle (specified muscle) – L 756.880
       optic nerve - L 743.520
       testicle - L * 752.810
       umbilicus - # 759.900
       vermian - 742.230
Auditory canal
       absent - L 744.000
       benign neoplasm - L # 216.200
       small - L 744.000
       stenosis - L 744.000
Auditory meatal stenosis - L 744.000
Auricle - see pinna
Auricular
       pit (ear) - L # 744.410
       septal defect (heart) - * 745.590
Autosome (chromosome)
       deletion - see deletion
       marker - 758.580
       mosaic - see mosaic
       other specified anomalies - 758.580
       translocation - see translocation
       trisomy - see trisomy
       unspecified anomalies - 758.590


**-B-**

Balantic hypospadias - 752.605
Balantic hypospadias with chordee  - 752.625
Baller-Gerold syndrome - 759.840
Band
       amniotic - # 658.800
       heart, anomalous - 746.910
       intestine - 751.420
       Ladd's - 751.420
       omentum - 751.420
       peritoneum - 751.420
Barrel chest - 754.820
Bart syndrome - 757.330
Basilar craniosynostosis - 756.030
Bat ear - L # 744.220
Bathocephaly - * 756.080
Beaded hair - 757.410
Beals syndrome - 759.860
Beckwith syndrome - 759.870
Beckwith-Wiedemann syndrome - 759.870
Beemer Langer syndrome - 759.860
Bell shaped chest - 754.820
Bell's palsy - L # 351.000
Benign external hydrocephaly - x
Bent nose - # 754.020
Bicornate uterus - L 752.380
Bicuspid
       aortic valve - 746.400
       pulmonary valve - 746.080
       tricuspid valve - 746.100
Bifid - see also cleft, accessory
       nose - 748.120
       rib - L 756.310
       scrotum - 752.820
       sternum - 756.380
       thumb - L 755.010
       uvula - 749.080
       vertebrae
              cervical - 756.140
              lumbar - 756.160
              NOS - 756.180
              sacral - 756.170
              thoracic - 756.150
       xyphoid process - 756.380
Bilateral superior vena cava - 747.410
Bile duct
       agenesis - 751.650
       atresia - 751.650
       other anomalies - 761.670
Biliary
       atresia - 751.650
       dysgenesis - 751.670
       obstruction - x
Biliary tract anomalies, NOS - 751.680
Bilirubin excretion disorders - # 277.400
Bilobar right lung - 748.625
Biparietal narrowing - * 756.080
Birthmark, NOS - # 757.385

Bitemporal narrowing - * 756.080
Bladder
      absent - 753.800
      cystocele - 753.820
      diverticulum - 753.820
      ectopic - 753.810
      enlarged - x
      exstrophy - 753.500
      extroversion - 753.500
      hernia - 753.820
      hypertrophy - x
      hypoplasia - 753.880
      hypoplastic - 753.880
      neck
            atresia - 753.610
            other and unspecified atresia and stenosis - 73.690
            stenosis - 753.610
      neurogenic - x
      other specified anomalies - 753.880
      outlet obstruction - 753.690
      prolase (mucosa) - 753.830
      small - x
      thickened - x
      trabeculated - x
      unspecified anomalies - 753.920
Blepharophimosis - L 743.635
Blepharophimosis syndrome - 759.800
Blepharoptosis - L 743.600
Block, heart - 746.870
Bloom syndrome - 759.890
Blue
      baby - 746.930
      Mongolian spot - x
      nevus - see skin-benign neoplasm
      sclera - L * 743.450
Blueberry muffin spots - x
BOR syndrome - 759.800
Body stalk anomaly - 756.790
Bone
      unspecified anomalies - 756.920
Bonneville-Ullrich syndrome, NOS - 758.690
Bourneville's disease - 759.500
Bowed/bowing
      femur - L 754.400
      legs, NOS - 754.420
      lip - L 744.880
      lower leg - L 754.410
      fibula - L 754.410
      tibia - L 754.410
      ulna without Madelung deformity - L 755.530
Box shaped head - 754.080
Brachial plexus palsy - L # 767.600
Brachiocephalic trunk, common - L 747.640
Brachycephaly - 754.080
Brachydactyly
      finger - L # 755.500
      toe - L # 755.600

Bradycardia - x
Brain
     absent - 740.000
     asymmetry - x
     atrophy - 742.480
     enlarged - * 742.400
     other specified anomalies - 742.480
     small - 742.486
     unspecified - 742.900
Brainstem
     anomalies - 742.480
     hypoplastic - 742.280
     reduction defect - 742.280
     small - 742.280
Branch pulmonary artery stenosis - L * 747.325
Branchial arch syndrome - 759.800
Branchial cleft
     cyst - L 744.400
     fistula - L 744.400
     other anomalies - L 744.480
     pit - L 744.400
     remnant - L 744.400
     sinus - L 744.400
Breast
     absent
          nipple absent - L 757.600
          nipple present - L 757.610
     accessory (with accessory nipple) - L 757.620
     benign neoplasm - # 216.500
     ectopic (with nipple) - L 757.620
     hypertrophy - x
     hypoplastic (with hypoplastic nipple) - L 757.610
     other specified anomalies - # 757.680
     small - x
Broad
     face - 744.910
     hand - L 755.510
     neck - # 744.500
Broad ligament
     absent - L 752.100
     other and unspecified anomalies - L 752.190
Bronchiectasis - L 748.610
Bronchoesophageal fistula - 750.330
Bronchogenic cyst - L 748.350
Bronchomalacia - x
Bronchopulmonary dysplasia - x
Bronchopulmonary fistula - L 748.350
Bronchus
     absent - L 748.350
     other anomalies - L 748.350
     other specified anomalies - L 748.380
     stenosis - L 748.340
     unspecified anomalies - 748.390
Brown syndrome - # 378.000
Brushfield spots - L # 743.800
Bulging eye - L # 743.800
Bullosa

       epidermolysis - 757.330

       ichthyosis - 757.115

Bullous type ichthyosis congenita - 757.115

Buphthalmos - L 743.200

Buried penis - 752.860

Butterfly vertebra

       cervical - 756.140

       lumbar - 756.160

       NOS - 756.180

       sacral - 756.170

       thoracic - 756.150


**-C-**

Café au lait spots - # 757.390

Caffey syndrome - 756.530

Calcaneovalgus - L 754.600

Calcaneovarus - L 754.510

Calvarium - see aso skull

       absent - 740.020

       asymmetry - 754.055

Camptodactyly

       finger - L # 755.500

       toe - L # 755.600

Camptomelic dysplasia - 756.480

Camurati-Engelmann syndrome - 756.550

Canal of Nuck cyst - 752.470

Cardiomegaly - * 746.860

Cardiomyopathy - * 746.860

Cardiomyopathy, hypertrophic - * 746.860

Cardiospasm - 750.720

Cardio-splenic syndrome - 759.890

Cardiovascular system, other specified anomalies - L 747.880

Carotid artery

       absent - L 747.640

Carpal bone

       accessory - L 755.525

Carpenter syndrome - 759.840

Carp shaped mouth - L 744.880

Cartilage (ear)

       absent - L * 744.230

       decreased - L * 744.230

       unspecified anomalies - 756.930

Cat eye syndrome - 758.580

Cataract

       anterior polar - L 743.325

       NOS - L 743.320

       other specified - L 743.326

Cauda equina anomalies, other - 742.530

Caudal dysplasia - 759.840

Caudal regression syndrome - 759.840

Cauliflower ear - L * 744.230

Cavum septum pellucidum - x

Cebocephaly - 759.800

Cecum

       duplication - 751.500

       malrotation - 751.400

Central nervous system (CNS) hemorrhage - x
Cephalohematoma - x
Cephalopagus conjoined twins - 759.410
Cerebellar atrophy - 742.230
Cerebellum anomalies - 742.230
Cerebral/cerebrum
       atrophy - 742.480
       cortical dysplasia - 742.480
       cyst - 742.420
       lipidoses - # 330.100
       reduction deformities - 742.200
Cerebral vessels, other anomalies - L 747.810
Cerebro-oculo-facial-skeletal syndrome - 759.890
Cervical rib - L # 756.200
Cervix (genital)
       absent - 752.400
       agenesis - 752.400
       atresia - 752.400
       doubling - * 752.480
       other specified anomalies - * 752.480
       unspecified anomalies - 752.490
Chalasia - x
CHARGE association - 759.890

Chediak-Higashi syndrome - 757.300
Cheek
       hypoplastic -L 744.880
       skin tag - L # 744.110
Chest
       asymmetry - 754.820
       barrel - 754.820
       bell shaped - 754.820
       benign neoplasm - # 216.500
       deformed - 754.820
       funnel - 754.810
       narrow - 754.820
       other anomalies - 754.820
       pigeon - 754.800
       shield - 754.825
       small - 754.820
Chin
       cleft - x
       dimple - x
       pointed - * 756.080
       receding - 524.000
       small - 524.000
Choanal
       atresia - L 748.000
       stenosis - L 748.000
Choledochal cyst - 751.660
Chondroectodermal dysplasia - 756.520
Chondrodysplasia - 756.410
       other specified - 756.480
       punctata - 756.575
       with hemangioma - 756.420
Chondrodystrophy
       other specified - 756.480

unspecified - 756.490
Chordee (penile)
        with hypospadias
                    coronal - 752.625
                    first degree - 752.625
                    glandular - 752.625
                    NOS - 752.620
                    penile - 752.626
                    perineal - 752.627
                    scrotal - 752.627
                    second degree - 752.626
                    third degree - 752.627
        without hypospadias - 752.621
Choroid (eye)
        coloboma - L 743.535
        specified anomalies - L 743.530
Choroid plexus cyst
        bilateral - * 742.485
        multiple - * 742.485
        unilateral - x
Chorioretinitis - # 363.200
Chromosome
        autosome - see autosome
        NOS
                    additional , NOS - 758.910
                    deletion, NOS - 758.920
                    duplication, NOS - 758.930
                    mosaicism, NOS - 758.900
        unspecified anomaly - 758.990
        sex - see sex chromosome
Chylothorax - # 457.800
Circulatory system, unspecified anomalies - 747.900
Cisterna magna, enlarged - 742.380
Clavicle anomalies - L 755.550
Claw
        foot - L 755.350
        hand - L 755.250
Cleft
        alveolar ridge/alveolus - 749.100
        branchial - L 744.400
        chin - x
        ear - L * 744.230
        face/facial - L 744.880
        foot - L 755.350
        gingiva - 749.100
        gum - 749.100
        hand - L 755.250
        laryngotracheoesophageal - 748.385
        larynx - 748.385
        lip
                    lateral - 744.800
                    with any cleft palate - L 749.200
                            central - 749.220
                            midline - 749.220
                    without cleft palate - L 749.100
                            central - 749.120
                            midline - 749.120

mandible - * 756.080
mitral valve - 746.505
mouth, lateral - 744.800
nose - 748.120
palate
    with cleft lip - see cleft lip with any cleft palate
    without cleft lip
        hard palate (alone) - L 749.000
            central - 749.020
            midline - 749.020
        NOS (hard/soft not specified) - 749.090
        soft and hard palate - 749.090
        soft palate (alone) - L 749.040
            central - 749.060
            midline - 749.060
        submucosal
            hard - 749.020
            NOS (hard/soft not specified) - 749.090
            soft - 749.060
tongue - 750.140
tricuspid valve - 746.100
uvula - 749.080
vertebrae
    cervical - 756.140
    lumbar - 756.160
    NOS - 756.180
    sacral - 756.170
    thoracic - 756.150
Cleidocranial dysostosis - 755.555
Clenched hand or fist - L # 755.500
Clenched toes - L # 755.600
Click, hip - x
Clifford's syndrome - x
Clinodactyly
    finger - L # 755.500
    toe - L # 755.600
Clitoris
    absent - * 752.450
    enlarged - * 752.450
    hypertrophy - * 752.450
    other anomaly - * 752.450
    prominent - * 752.450
    prominent prepuce - x
Clitoromegaly - * 752.450
Cloaca
    exstrophy - 751.550 and 756.790
    persistent - 751.550
Close set eyes - * 756.080
Cloudy cornea - L 743.400
Cloverleaf head shape - 756.000
Club/clubbed
    fingers - L 754.840
    foot, NOS - L 754.730
    hand - L 754.840
    nail - L 757.540
Coarctation of aorta
    distal - 747.110

juxtaductal - 747.190
postductal - 747.110
preductal - 747.100
proximal - 747.100
unspecified - 747.190
Cockayne syndrome - 759.820
Coffin-Siris syndrome - 759.800
COFS syndrome - 759.890
Collateral vessel
involving aorta - 747.280
involving pulmonary artery (and not aorta) - L 747.380
not involving aorta or pulmonary artery - L 747.880
Collodian baby - 757.110
Coloboma
anterior segment
other - L 743.480
unspecified - L 743.490
choroid - L 743.535
eyelid - L 743.636
iris - L 743.430
lens - L 743.340
NOS - L 743.490
optic disc/nerve - L 743.520
retina - L 743.535
Colon
absent - 751.200
atresia - 751.200
hypoplastic - 751.520
malrotation - 751.400
short - 751.520
small - 751.520
stenosis - 751.200
transposition - 751.510
Colpocephaly - 742.280
Common
atrioventricular canal - * 745.630
atrioventricular canal with VSD - * 745.620
atrium - 745.610
brachiocephalic trunk - L 747.640
ventricle  (heart) - 745.300
Complete
atrioventricular canal - * 745.630
atrioventricular canal with VSD - * 745.620
mirror reversal of abdominal organs with normal thoracic organs - 759.330
mirror reversal of all organs - 759.300
mirror reversal of thoracic organs with normal abdominal organs - 759.320
Complex - see syndrome
Concealed penis - 752.860
Conduction defects (heart) - 746.880
Cone shaped head - 754.080
Congenital anomaly, NOS - 759.990
Congenital contractural arachnodactyly syndrome - 759.860
Congenital encephalopathy - x
Congenital heart disease
acyanotic - 746.920
cyanotic - 746.930
NOS - 746.990

Conjoined twins
 cephalopagus - 759.410
 craniopagus (head-joined twins) - 759.410
 dicephalus (two heads) - 759.400
 ischiopagus - 759.480
 other specified - 759.480
 pelvis-joined twins - 759.480
 pygophagus (buttock-joined twins) - 759.440
 thoracopagus (thorax-joined twins) - 759.420
 unspecified - 759.490
 xiphopagus (xiphoid-joined twins) - 759.430
Conjunctivitis - x
Connective tissue
 other specified anomalies - L 756.880
 unspecified anomalies - 756.940
Conradi syndrome - 756.575
Constriction band syndrome - # 658.800
Contracture
 joint (flexion, individual) - L 755.800
 sternocleidomastoid muscle - L 754.100
Cor biloculare - 745.700
Cornea
 cloudy - L 743.400
 enlarged - L 743.220
 leukoma - L 743.400
 opacity - L 743.400
 other specified - L 743.410
Cornelia de Lange syndrome - 759.820
Coronal suture
 closed - L 756.010
 craniosynostosis - L 756.010
 fused - L 756.010
Coronary artery anomalies - 746.885
Coronary sinus anomalies - 746.885
Corpus callosum
 anomalies - 742.210
 cyst - 742.420
Cortex/cortical
 anomalies - 742.200
 atrophy - 742.480
 dysplasia (cerebral) - 742.480
 hyperostosis, infantile - 756.530
Cor triatriatum - 746.820
Cor triloculare biatriatum - 745.300
Costello syndrome - 759.800
Coxa
 valga - L 755.660
 vara - L 755.660
Cranial nerve defects - 742.480
Craniofacial
 abnormality NOS - 756.090
 craniofacial disproportion - 756.090
 dysostosis - 756.040
 other syndromes - 756.046
Craniorachischisis - 740.100
Cranioschisis - 740.020
Craniosynostosis

basilar - 756.030
coronal - L 756.010
lambdoidal - L 756.020
metopic - 756.006
NOS - 756.000
other - 756.030
sagittal - 756.005
squamosal - 756.000
Craniotabes - x
Cranium, square - 754.080
Crease
      ear - L 744.280
      infraorbital - L # 743.800
      palm or hand - see palmar crease
Crepitus hip - x
Cri du chat syndrome - 758.310
Cross fused renal ectopia - 753.320
Crossed eyes - # 368.000
Crouzon's disease - 756.040
Cryptophthalmos - L 743.000
Cryptorchidism
      bilateral - * 752.514
      left - L * 752.501
      NOS - * 752.520
      right - L * 752.502
      unilateral - L * 752.500
Cubitus valgus - L 755.540
Curvature of spine (postural), NOS - 754.220
Curved sternum - 754.820
Cutis aplasia
      not involving scalp - 757.395
      scalp - 757.800
Cutis laxa hyperelastica - 757.370
Cutis marmorata - x
Cyanotic congenital heart disease - 746.930
Cyclops - 759.800
Cyst/cystic
      abdominal NOS - 759.990
      adenomatoid malformation lung - L 748.480
      amniotic - # 658.800
      arachnoid - x
      branchial cleft - L 744.400
      bronchogenic - L 748.350
      canal of Nuck - 752.470
      cerebral - 742.420
      choledochal - 751.660
      choroid plexus
            bilateral - * 742.485
            multiple - * 742.485
            unilateral - x
      corpus callosum - 742.420
      dysplasia kidney - L 753.160
      duplication - 751.500
      embryonal (vagina) - # 752.460
      embryonic remnants (male) - L 752.870
      enterogenous - 751.500
      ependymal - 742.420

epoophoron - L 752.110
fimbrial - L 752.120
Gartner's duct - L 752.110
glioependymal - 742.420
gum - x
hydatid of Morgagni - L 752.870
hygroma - 228.100
intracranial - 742.420
kidney (single) - L 753.100
lacrimal apparatus/duct - L 743.660
liver - 751.610
lung
       multiple - L 748.410
       other specified - L 748.480
       single - L 748.400
mediastinum - 748.810
mesenteric remnant - L 752.110
ovarian
       multiple - L 752.085
       single - L 752.080
pancreatic - 751.740
parovarian - L 752.120
periventricular - 742.420
porencephalic - * 742.410
posterior fossa - 742.230
preauricular - L # 744.410
renal (single) - L 753.100
skin - # 757.390
spleen - 759.080
subependymal - 742.420
thyroglossal - 759.220
tongue - x
urachus - # 753.700
vagina
       embryonal - # 752.460
       other - 752.470
ventricular (brain) - * 742.485
vulva - 752.470
Wharton duct - x
Wolffian duct - L 752.870
Cystic fibrosis, no mention of meconium ileus - # 277.000
Cystic fibrosis, with mention of meconium ileus - # 277.010
Cystic kidney NOS - L 753.180
Cystocele bladder - 753.820
Cytomegalovirus (CMV), congenital (in utero infection) - # 771.100

**-D-**

Dacryocystocele - L 743.660
Dacryostenosis - L # 743.650
Dandy-Walker syndrome - * 742.310
Deafness, congenital - L * 744.090
Defect
   Gerbode - 745.420
Deletion (chromosome)
   4 - 758.320
   5 - 758.310

13 (long arm, q) - 758.330
                17 (long arm, q) - 758.340
                17 (short arm, p) - 758.350
                18 (long arm, q) - 758.340
                18 (short arm, p) - 758.350
                21 (partial or total) - 758.300
                B, NOS - 758.310
                B, NOS - 758.320
                D, NOS (long arm, q) - 758.330
                E (long arm, q) - 758.340
                E (short arm, p) - 758.350
                G, NOS (partial or total) - 758.300
                NOS (unspecified chromosome) - 758.920
                other specified (autosomal) - 758.380
                unspecified (autosomal) - 758.390
                X (partial) - 758.610
Depressions in skull - # 754.040
Dermal
        sinus of head - L 744.480
        sinus spine - # 685.100
Dermoid cyst
        epibulbar - L 743.810
        eye - L 743.810
Deviation nasal septum - # 754.020
Dextrocardia
        with complete situs inversus - 759.300
        with situs solitis - 746.800
        without situs inversus - 746.800
Dextroposition
        aorta - 747.260
        heart - see dextrocardia
Diamond-Blackfan syndrome (anemia) - # 284.000
Diaphragm/diaphragmatic
        absent - L 756.600
        elevated - x
        eventration - L 756.620
        hernia
                Bochdalek - L 756.615
                Morgagni - L 756.616
                NOS - L 756.610
                Posterolateral - L 756.615
        other specified anomalies - L 756.680
        paralysis - L 756.680
        unspecified anomalies - L 756.690
Diaphyseal dysplasia, progressive - 756.550
Diastasis recti - x
Diastematomyelia - 742.520
Diastrophic dwarfism - 756.445
Didelphys uterus - 752.200
Diencephalic syndrome - 253.820
DiGeorge syndrome - 279.110
Digestive system, NOS
        absent (complete or partial) - 751.800
        duplication - 751.810
        ectopic - 751.820
        fistula
                with urinary tract - 753.860

with uterus - 752.320
  obstruction, NOS - 752.900
  other specified anomalies - 751.880
  unspecified anomalies - 751.900
Digit, NOS
  absent - L 755.440
  accessory - see polydactyly
  extra - see polydactyly
  overlapping - L 755.880
Digitalized great toe - L # 755.600
Digitalized thumb - L # 755.500
Dilatation/dilated/dilation - see also large
  aorta - 747.270
  atrium - x
  esophagus - 750.400
  pulmonary artery - 747.330
  pulmonary valve - 746.080
  renal collecting system
    central - L 753.380
    lower - L 753.480
    upper - L 753.480
  renal pelvis - L 753.380
  tricuspid valve - 746.100
  ureter - L 753.220
  vena cava - 747.480
  ventricle (brain) - 742.390
  ventricle (heart) - x
Dimple in chin - x
Disappearing penis syndrome - 752.860
Disease - see syndrome
Dislocatable hip - L 754.310
Dislocation
  elbow - L 754.830
  hip - L 754.300
  knee - L 754.440
  shoulder - x
  tongue - 750.130
Displaced anus - 751.530
Displacement
  cardiac through esophageal hiatus - 750.600
  esophagus - 750.410
  stomach - 750.730
  tongue - 750.130
  uterus - 752.310
Distal arthrogryposis syndrome - L 755.800
Diverticulum
  bladder - 753.820
  esophagus - 750.420
  Meckel's - # 751.010
  stomach - 750.740
  urethral - 753.880
Divisum, pancreas - 751.780
Dolichocephaly - * 754.030
Dorsiflexion of foot - L 754.780
Double - see also duplication
  aortic arch - 747.250
  collecting system (renal) - L 753.410

inlet left ventricle - 745.300
inlet right ventricle - 745.300
kidney (and renal pelvis) - L 753.310
meatus (urethral, urinary) - 753.840
ossification center in the manibrium - 756.380
outlet left ventricle - 745.180
outlet right ventricle - 745.180
ureter - L 753.410
urethra - 753.840
urethral orifice - 753.840
Double orifice mitral valve - 746.505
Doubling
       cervix - * 752.480
       uterus - 752.200
       vagina - * 752.480
Down syndrome
       facies - 744.910
       karyotype trisomy 21 - 758.000
       karyotype trisomy G, NOS - 758.010
       mosaic - 758.040
       NOS - 758.090
       translocation trisomy (duplication of a 21) - 758.020
       translocation trisomy (duplication of a G, NOS) - 758.030
Downturned mouth - L 744.880
Duane syndrome - # 378.000
Duct
       bile
              agenesis - 751.650
              atresia - 751.650
       hepatic
              agenesis - 751.650
              atresia - 751.650
       omphalomesenteric - 751.000
       vitelline - 751.000
Duodenum
       absent - 751.100
       atresia - 751.100
       stenosis - 751.100
       web - 751.560
Du Pan syndrome - 759.840
Duplex renal collecting system - L 753.410
Duplication - see also double/doubling
       alimentary tract, NOS - 751.810
       chromosome - see also trisomy
           NOS - 758.930
       collecting system (renal) - L 753.410
       digestive system, NOS - 751.810
       esophagus - 750.430
       gallbladder - 751.640
       intestine - 751.500
       nail - L 757.580
       pylorus - 751.500
       renal collecting system - L 753.410
       stomach - 750.750
Dwarf/dwarfism
       Amsterdam - 759.820
       achondroplastic - 756.430

diastrophic - 756.445
hypochondrodysplastic - 756.480
metatrophic - 756.446
NOS - 756.490
thanatophoric - 756.447
Dysautonomia, familial - 742.810
Dysgenesis
adrenal gland - L 759.180
biliary - 751.670
Dysostosis
cleidocranial - 755.555
craniofacial - 756.040
mandibulofacial - 756.045
metaphyseal - 756.450
radioulnar - L 755.536
spondylocostal - 756.480
Dysmorphic
aortic valve - 746.480
mitral valve - 746.505
pulmonary valve - 746.080
Dysplasia - see also hypoplasia
aortic valve - 746.480
bronchopulmonary - x
caudal - 759.840
chondroectodermal - 756.520
cortical (cerebral) - 742.480
dyssegmental - 756.480
ears - L * 744.230
ectodermal
NOS - 757.340
other specified - 757.346
X-linked type - 757.345
eye - L 743.100
fronto-nasal - 756.046
hip
bilateral - 755.667
NOS - 755.665
unilateral - L 755.666
kidney
bilateral - 753.000
NOS - 753.009
unilateral - L 753.010
kyphomelic - 756.480
mitral valve - 746.505
multiple epiphyseal - 756.570
nail - L 757.580
oculoauriculovertebral - 756.060
pulmonary valve (not hypoplasia) - 746.080
polystotic fibrous - 756.510
progessive diaphyseal - 756.550
pulmonary valve - 746.080
rib - L 756.340
Septo-optic - 742.880
spondyloepiphyseal - 756.460
spondylometaphyseal - 756.480
spondylothoracic - 756.480
Streeter syndrome/dysplasia - # 658.800

thoracic-pelvic-phalangeal- 756.400
            tricuspid valve - 746.100
Dyssegmental dysplasia - 756.480
Dystrophy/dystrophic
            asphyxiating thoracic - 756.400
            myotonic - 759.890
            nail - L 757.580


**-E-**

Eagle-Barrett's syndrome - 756.720
Ear
            absent - L 744.010
            absent carilage - L * 744.230
            anomaly NOS - L 744.300
            appendage (not preauricular) - L # 744.120
            asymmetry - x
            bat - L # 744.220
            benign neoplasm - L # 216.200
            cauliflower - L * 744.230
            cleft - L * 744.230
            crease - L 744.280
            decreased cartilage - L * 744.230
            deformity NOS - L 744.300
            dysplastic - L * 744.230
            elfin - L * 744.230
            hypoplastic (not microtia) - L * 744.230
            inner ear anomalies - L 744.030
            large - L 744.200
            lobule (not preauricular) - L # 744.120
            lop - L * 744.230
            low set - L # 744.245
            malformed - L * 744.230
            middle ear anomalies - L 744.020
            misplaced - L 744.240
            other misshapen - L * 744.230
            other specified - L 744.280
            papilloma - L # 744.120
            pit (not preauricular) - L 744.280
            pit (preauricular) - L # 744.210
            pixie-like - L * 744.230
            pointed - L * 744.230
            posteriorly rotated - L # 744.246
            rotated - L # 744.246
            small (not microtia) - L * 744.230
            tag (not preauricular) - L # 744.120
            unspecified anomalies - L 744.300
            unspecified, with hearing impairment - L * 744.090
Ear canal - see auditory canal
Ebstein's anomaly - 746.200
Echogenic kidney - x
Ectodermal dysplasia
            NOS - 757.340
            other specified - 757.346
            X-linked type - 757.345
Ectopia (ectopic) cordia - 746.880
Ectopia vesicae - 753.500

Ectopic - see also displacement
    adrenal gland - L 759.130
    alimentary tract, NOS - 751.820
    anus - 751.530
    bladder - 753.810
    breast (with accessory nipple) - L 757.620
    digestive system, NOS - 751.820
    heart - 746.880
    kidney - L 753.330
    lung tissues - L 748.600
    nipple
        only - L # 757.650
        with accessory breast - L 757.620
    pancreas - 751.730
    pupil - L 743.440
    spleen - 759.050
    testicle - L 752.530
    ureter - L 753.420
    urethra - 753.850
    urethral orifice - 753.850
Ectrodactyly
    foot - L 755.350
    hand - L 755.250
    NOS - L 755.440
Ectrodactyly-Ectodermal dysplasia-Clefting syndrome - 759.840
Ectropion - L 743.610
Edema
    hereditary, of legs - 757.000
    not of legs - x
Edwards syndrome
    karyotype normal (Edwards phenotype) - 758.295
    karyotype trisomy 18 - 758.200
    karyotype trisomy E, NOS - 758.210
    mosaic - 758.240
    NOS - 758.290
    translocation trisomy 18 (duplication or an 18) - 758.220
    translocation trisomy 18 (duplication or an E, NOS) - 758.230
EEC syndrome - 759.840
Ehlers-Danlos syndrome - 756.850
Eisenmenger's syndrome - 745.410
Elbow
    anomalies - L 755.540
    dislocation - L 754.830
    hyperextension - L 755.540
    webbed - L 755.800
Elevated diaphragm - x
Elfin ear - L * 744.230
Ellis-van Creveld syndrome - 756.525
Elongated - see long
Embryonic remnants (male) cyst - L 752.870
Embryopathia, NEC - 759.910
Emphysema, lobar - L 748.880
Encephalocele
    frontal - 742.085
    frontonasal - 742.085
    occipital - 742.000
    occipitocervical - 742.000

other specified site - 742.080
            parietal - 742.086
            posterior - 742.000
            sphenoid - 752.080
            unspecified site - 742.090
Encephalocutaneous angiomatosis - 759.610
Encephalopathy, congenital - x
Enchondromatosis - 756.410
Endocardial cushion defect
        NOS - 745.690
        other - 745.680
Endocardial fibroelastosis - 425.300
Endocrine gland
        other specified anomalies - 759.280
        unspecified anomalies - 759.290
Endothelial vessel - L 747.880
Engelmann syndrome - 756.550
Enlarged - see large
Enophalmia - L # 743.800
Enophthalmos - L # 743.800
Enterogenous - 751.500
Entropion - L 743.620
Ependymal cysts - 742.420
Epiblepharon - L * 743.630
Epicanthal folds - L # 743.800
Epidermal nevus syndrome - 757.300
Epidermolysis bullosa - 757.330
Epigastric hernia - 756.795
Epiglottis
        anomalies - 748.300
        hypoplastic - 748.300
Epiloia - 759.500
Epiphyseal dysplasia, multiple - 756.570
Epispadias - 752.610
Epoophoron cyst - L 752.110
Epstein's pearls - x
Epulis - x
Equinovalgus - L 754.680
Equinovarus - L 754.500
Equinus foot - L 754.730
Erb's palsy - L # 767.600
Escobar syndrome - 759.840
Esophagus/esophageal
        atresia
                without tracheoesophageal fistula - 750.300
                with tracheoesophageal fistula - 750.310
        dilatation - 750.400
        displacement - 750.410
        diverticulum - 750.420
        duplication - 750.430
        fistula - 750.480
        giant - 750.400
        other specified anomalies - 750.480
        pouch - 750.420
        short - x
        stenosis - 750.340
        unspecified anomalies - 750.910

web - 750.350
Esotropia - # 368.000
Ethmocephaly - 759.800
Eustacian tube
        absent - L 744.250
        anomaly - L 744.250
Eustacian valve - x
Eventration of diaphragm - L 756.620
Eversion/everted eyelid - L 743.610
Eversion foot - L 754.680
Exencephaly - 740.020
Exomphalos - 756.700
Exophthalmos - L # 743.800
Exostosis - 756.470
Exotropia - # 378.000
Exstrophy
        bladder - 753.500
        cloaca - 751.550 and 756.790
        lung - L 748.690
External auditory meatal stenosis - L 744.000
External genitalia, absent
        female - * 752.440
        male - 752.880
Extra - see also accessory
        chromosome - see trisomy
        digit - see polydactyly
        finger - see polydactyly
        renal pelvis - L 753.380
        rib
                in cervical region - L # 756.200
                other - L 756.330
        toe - see polydactyly
Extremity - see limb
Extroversion bladder - 753.500
Eye/eyes
        absent - L 743.000
        agenesis - L 743.000
        almond shaped - L # 743.800
        aplasia - L 743.100
        asymmetry - x
        bulging - L # 743.800
        close set - * 756.080
        crossed - # 368.000
        deep set - L # 743.800
        dysplasia - L 743.100
        enlarged - L 743.210
        flat - L # 743.800
        fused
                closed - L * 743.630
                together - 759.800
        Harlequin deformity - L 743.670
        hypoplasia - L 743.100
        mesodermal dysgenesis - L 743.900
        other specified - L # 743.800
        prominent - L # 743.800
        protruding - L # 743.800
        rudimentary - L 743.100

slant (upward, downward) - L # 743.800
small - L 743.100
sunken - L # 743.800
sun-setting - x
unspecified - L 743.900
wide set - 756.085
Eyebrow
absent - L 744.880
Eyelash
absent - L * 743.630
long - L * 743.630
Eyelid
absent - L * 743.630
benign neoplasm - L # 216.100
coloboma - L 743.636
eversion/everted - L 743.610
fused - L * 743.630
other specified - L * 743.630
weak - L * 743.630

**-F-**

Face/facial
absent - L 744.880
anomaly NOS - 744.910
asymmetry - 754.000
asymmetry crying - L 351.000
benign neoplasm - # 216.300
broad - 744.910
cleft - L 744.880
flat profile - 744.910
microsomia - L 756.065
other specified anomalies - L 744.880
other specified bone anomalies - * 756.080
palsy - L # 351.000
skin tag - L # 744.110
small - 744.910
teratoma - 238.010
triangular - 744.910
unspecified bone anomalies - 756.090
Facies - see also features
compression - 754.010
Down syndrome - 744.910
flat - 744.910
Potter's - 754.010
Facio-auricular-digital syndrome - 759.800
Facio-auriculo-vertebral syndrome - 756.060
Fallopian tube
absent - L 752.100
hypoplastic - L 752.190
other and unspecified anomalies - L 752.190
Fallot's pentalogy - 745.210
Fallot's tetralogy - 745.200
Familial dysautonomia - 742.810
Fascia
other specified anomalies - L 756.880
Features

abnormal - 744.910
Down syndrome - 744.910
dysmorphic - 744.910
Trisomy 21 - 744.910
Female genitalia (external)
    benign neoplasm - # 221.000
    other specified anomalies - * 752.480
    unspecified anomalies - 752.490
Femoral fibular hypoplasia – unusual facies syndrome - 759.840
Femoral hypoplasia – unusual facies syndrome - 759.840
Femur
    absent
        only - L 755.320
        with absent tibia and fibular (total or partial)- L 755.310
        with absent tibia, fibula, and foot - L 755.300
    anteversion - L 755.650
    bowed - L 754.400
    hypoplastic - L 755.650
    other specified anomalies - L 755.650
    short - L 755.650
    torsion - L 755.650
Femur-fibula-ulna syndrome - 759.840
Fenestrated ASD - 745.510
Fetal
    Accutane (Isoretinoin) syndrome - 760.760
    akinesia deformation sequence - 759.840
    alcohol
        effect - 760.720
        syndrome - 760.710
    Dilantin syndrome - 760.750
    hydantoin syndrome - 760.750
FG syndrome - 759.800
Fibroelastosis, endocardial - 425.300
Fibromatosis colli - L 754.100
Fibrosis
    liver - 751.610
    myocardial - 425.300
Fibula
    absent
        only (total or partial) - L 755.366
        with absent femur (total or partial) and tibia (total or partial)- L 755.310
        with absent femur (total or partial), tibia, and foot - L 755.300
        with absent tibia - L 755.320
        with absent tibia and foot - L 755.330
    bowed - L 754.410
    hypoplastic - L * 755.630
    other specified anomalies - L * 755.630
    short - L * 755.630
Fibular hemimelia - L 755.366
Fibular ray defect, NOS - L 755.366
Filum terminale, fat - x
Fimbrial cyst - L 752.120
Finger
    absent
        fifth (with or without fourth) - L 755.270
        first (thumb) - L 755.260
        first (thumb) with absent radius (total or partial) - L 755.260

NOS - L 755.240
third (with or without second, fourth) - L 755.250
acrodactylia - L # 755.500
anomalies - L # 755.500
arachnodactyly - L # 755.500
bifid (thumb) - L 755.010
brachydactyly - L # 755.500
camptodactyly - L # 755.500
clinodactyly - L # 755.500
club - L 754.840
cortical (thumb) - x
digitalized (thumb) - L # 755.500
flexion deformity - L # 755.500
fused - L 755.100
hyperextension - L # 755.500
hypoplastic
all other - L 755.585
thumb (isolated) - L 755.260
incurving - L # 755.500
long - L # 755.500
nubbin - L 755.240
overlapping - L # 755.500
rudimentary - L 755.240
short - L # 755.500
small, all other - L # 755.500
small, thumb - L # 755.500
symbrachydactyl - L #  755.500 and L 755.190-755.199 (depending on the laterality)
symphalangism - L # 755.500
syndactyly, unspecified
bilateral - 755.192
NOS - 755.193
unilateral - 755.191
triphalangeal (thumb) - L # 755.500
webbed - L 755.110
Fissure
anal - x
rectal - * 751.580
thin palpebral - L 743.635
Fistula
anal - 751.540
anourethral - 753.860
anovaginal - 752.420
anovesical - 753.860
branchial cleft - L 744.400
bronchoesophageal - 750.330
bronchopulmonary - L 748.350
digestive tract with uterus - 752.320
digestive-urinary tract - 753.860
esophageal - 750.480
Fourchette - * 752.480
hepatic artery-portal vein - 747.450
lip - 750.260
portal vein-hepatic artery - 747.450
rectal - 751.540
rectourethral - 753.860
rectovaginal - 752.420
rectovesical - 753.860

tracheoesophageal
       H type - 750.325
       with esophageal atresia - 750.310
       without esophageal atresia - 750.320
  urethral, NOS - 753.870
  urethrorectal - 753.860
  urinary tract with uterus - 752.320
  uterointestinal - 752.320
  uterovesical - 752.320
  uterus with digestive or urinary tract - 752.320
  vesicovaginal - 752.420

Flat
  eye - L # 743.800
  facial profile - 744.910
  facies - 744.910
  foot - L 754.610
  hand - L 754.880
  head - 754.080
  midface 744.910
  occiput - * 756.080
  side of head - L * 754.050

Flexed wrist - L 755.520
Flexion deformity finger - L # 755.500
Flexion deformity toe - L # 755.600
Fontanelle
  absent - # 754.040
  large - # 754.040
  small - # 754.040
  three - # 754.040

Foot
  abduction - L 754.690
  absent
       only - L 755.340
       with absent femur (total or partial), tibia, and fibula - L 755.300
       with absent lower leg - L 755.330
       with absent tibia and fibula (total or partial) - L 755.330
  adduction - L 754.590
  anomalies - L 755.610
  broad - L 755.610
  claw - L 755.350
  cleft - L 755.350
  clubbed - L 754.730
  deformities, NOS - L 754.735
  dorsiflexion - L 754.780
  ectrodactyly - L 755.350
  equinus - L 754.730
  eversion - L 754.680
  flat - L 754.610
  hyperextended - L 754.780
  hypoplasia - L 755.685
  inversion - L 754.590
  large - L 755.610
  lobster-claw - L 755.350
  long - L 755.610
  oligodactyly - L 755.340
  other specified anomalies - L 754.780
  plantar crease, deep - L 755.610

plantar furrow - L 755.610
positional defect, NOS - L 754.780
rocker-bottom - L # 755.616
short - L 755.610
small - L 755.610
split - L 755.350
turns
        inward - L 754.590
        outward - L 754.690
        upward - L 754.780
vertical talus - L # 755.616
Foramina of Magendie and Luschka atresia - * 742.310
Forearm
    absent
        only - L 755.220
        with absent hand - L 755.230
        with absent upper arm - L 755.210
    anomalies - L 755.530
    hemimelia - L 755.230
    short - L 755.530
Forehead
    hirsute - # 744.910
    other anomalies - * 756.080
Forelock, white - # 757.390
Foreskin
    absent - 752.860
    hooded - 752.860
    incomplete - x
    redundant - x
Fossa ovalis atrial septal defect - 745.510
Fourchette fistula - * 752.480
Fragile X syndrome - 758.880
Fragilitas ossium - 756.506
Franceschetti syndrome - 756.045
Frasier syndrome - 759.800
Freeman Sheldon syndrome - 759.800
Frenulum (tongue, lingual)
    anterior - # 750.000
    short - # 750.000
    thick - x
Frenulum (upper lip)
    anomalies - 750.270
    thick - x
Frontal bossing - 754.080
Frontal lobe anomalies - 742.200
Fronto-nasal dysplasia - 756.046
Fryn syndrome - 759.840
Fukuyama congenital muscular dystrophy - 759.890
Funnel chest - 754.810
Fused/fusion
    adrenal glands - L 759.180
    eyes
        closed - L * 743.630
        together - 759.800
    eyelid - L * 743.630
    fingers - L 755.100
    kidney - 753.320

      legs - 759.840
      lung lobes - L 748.580
      ossicles (ear)- L 744.020
      penoscrotal - 752.880
      radius and ulna - L 755.536
      rib - L 756.320
      sacroiliac joint - L 755.670
      scrotum - x
      suture
            basilar - 756.030
            coronal - L 756.010
            lambdoidal - L 756.020
            metopic - 756.006
            NOS - 756.000
            other - 756.030
            sagittal - 756.005
      thalami - 742.260
      toes - L 755.120
      ulna and radius - L 755.536
      vertebrae
            cervical - 756.140
            lumbar - 756.160
            NOS - 756.180
            sacral - 756.170
            thoracic - 756.150
      vulva - * 752.440

**-G-**

Galactokinase deficiency - # 271.110
Galactosemia
      classic - # 271.100
      NOS - # 271.190
Gallbladder
      agenesis - 751.630
      duplication - 751.640
      hypoplasia - 751.630
      other anomalies - 751.640
      small - x
Gangliosidosis - # 330.100
Gartner's duct cyst - L 752.110
Gardner syndrome - 759.630
Gastric volvulus - x
Gastroesophageal reflux (GER) - x
Gastroschisis - 756.710
Gaucher disease Type II - 759.870
Genitalia absent (sex unknown) - * 752.790
Genital organs, unspecified anomalies - 752.900
Genu
      recurvatum - L 754.430
      valgum - L 755.645
      varum - L 755.646
Gerbode defect - 745.420
Giant
      esophagus - 750.400
      kidney - L 753.340
Gingiva, cleft - 749.100

Glabella, prominent - # 748.180
Glaucoma - L 743.200
Glioependymal cysts - 742.420
Glossoptosis - 750.130
Glottic web - 748.205
Gluteal cleft, asymmetric - x
Glycogen storage disease - # 271.000
Goiter, congenital - 759.210
Goldenhar syndrome - 756.060
Goltz syndrome - 757.300
Gonadal dysgenesis, pure - 752.720
Gracile rib - L 756.340
Great veins
      other specified anomalies - 747.480
      unspecified anomalies - 747.490
Gum
      cleft - 749.100
      hypertrophy - 750.280
      hypoplastic - x
      other anomalies - 750.280
      prominent gum - 750.280

**-H-**

Hair
      beaded - 757.410
      Taenzer's - 757.430
      twisted - 757.420
      other specified anomalies - 757.480
      unspecified anomalies - 757.910
      whorl anomalies - # 757.390
Hairline
      low anterior - # 744.910
      low NOS - # 744.900
      low posterior - # 744.900
Hairy nevus - *216.920
Hallermann-Streiff syndrome - 756.046
Hallux
      valgus - L 755.605
      varus - L 755.606
Hamartoma
      other specified - 759.680
      unspecified - 759.690
Hammer toe - L # 755.600
Hand
      abnormal position
            with mention of forearm/wrist bone abnormality - L 754.840
            without mention of forearm/wrist bone abnormality- L 755.520
      absent
            only - L 755.240
            with absent forearm - L 755.230
            with absent humerus (total or partial), radius, and ulna - L 755.200
            with absent radius and ulna (total or partial) - L 755.230
      anomalies - L 755.510
      broad - L 755.510
      claw - L 755.250
      cleft - L 755.250

clenched - L # 755.500
club - L 754.840
ectrodactyly - L 755.250
finger-like (thumb) - L # 755.500
flat - L 754.880
hyperflexion - x
hypoplasia - L 755.585
large - L 755.510
lobster-claw - L 755.250
long - L 755.510
narrow - x
oligodactyly - L 755.240
other specified anomalies - L 754.880
short - L 755.510
small - L 755.510
spade-like - L 754.850
split - L 755.250
ulnar deviation - L 755.520
Harelip - see cleft lip
Harlequin deformity of eye - L 743.670
Harlequin fetus - 757.100
Head
abnormal shape NOS - 754.090
absent - 740.080
asymmetric - 754.055
box shaped - 754.080
cloverleaf shape - 756.000
cone shaped - 754.080
elongated - * 754.030
enlarged - * 742.400
flat - 754.080
flat side of - L * 754.050
misshapen - 754.090
small - 742.100
square - 754.080
teratoma - 238.010
tower - 754.080
triangular shape - 754.070
Heart
band, anomalous - 746.910
block - 746.870
conduction defects - 746.880
disease
acyanotic - 746.920
cyanotic - 746.930
NOS - 746.990
displacement through esophageal hiatus - 750.600
enlarged - * 746.860
"hole in the heart" - 745.900
hypoplastic left - 746.700
hypoplastic NOS - 746.880
hypoplastic right - 746.882
large - * 746.860
murmur - x
other specified - 746.880
tumor - 746.880
Heel, prominent - L 755.610

Hemangioendothelioma liver - L  * 228.040
Hemangioma
        intra-abdominal - L * 228.040
        intracranial - * 228.020
        other sites - L * 228.090
        retinal - L * 228.030
        skin and subcutaneous - * 228.010
        unspecified site - * 228.000
        with chondrodysplasia - 756.420
Hemianencephaly - 740.030
Hemiazygos vein anomalies - L 747.650
Hemicephaly - 740.030
Hemidiaphragm - L 756.617
Hemifacial microsomia - L 756.065
Hemihypertrophy - 759.890
Hemimelia, fibular - L 755.366
Hemimelia forearm - L 755.230
Hemimelia tibia - L 755.365
Hemipelvis - L 755.670
Hemivertebra
        cervical - 756.145
        lumbar - 756.165
        NOS - 756.185
        sacral - * 756.170
        thoracic - 756.155
Hemophilia (all types) - # 286.000
Hemorrhage, central nervous system (CNS) - x
Hepatic artery-portal vein fistula - 747.450
Hepatic duct
        agenesis - 751.650
        atresia - 751.650
        other anomalies - 751.670
Hepatic vein
        stenosis - L 747.650
Hepatitis, neonatal
        NOS - # 774.490
        other specified - # 774.480
Hepatomegaly - # 751.620
Hepatosplenomegaly - # 751.620 and # 759.020
Hereditary
        edema of legs - 757.000
        trophedema - 757.000
Hermaphroditism, true - 752.700
Hernia
        bladder - 753.820
        diaphragmatic
                Bochdalek - L 756.615
                Morgagni - L 756.616
                NOS - L 756.610
                Posterolateral - L 756.615
        epigastric - 756.795
        hiatal/hiatus - 750.600
        inguinal
                incarcerated - L * 550.100
                with mention of gangrene - L * 550.000
                with obstruction - L * 550.100
                without obstruction without mention of gangrene - L * 550.900

paraesophageal - 750.600

umbilical - # 553.100

Herpes simplex, congenital (in utero infection) - # 771.220

Heterotaxy syndrome - * 759.390

Heterotopia pancreas - 751.780

Hiatal/hiatus

hernia - 750.600

High arched palate - # 750.240

Hip

abduction - x

anomalies - L 755.660

Barlow positive - L 754.310

benign neoplasm - L # 216.700

click - x

crepitus - x

dislocatable - L 754.310

dislocation - L 754.300

dysplasia

bilateral - 755.667

NOS - 755.665

unilateral - L 755.666

hyperextended - L 755.660

hypoplasic

bilateral - 755.667

NOS - 755.665

unilateral - L 755.666

laxity - x

loose - x

Ortolani positive - L 754.310

positive Barlow - L 754.310

positive Ortolani - L 754.310

predislocation - L 754.310

preluxation - L 754.310

subluxable - L 754.310

subluxation - L 754.310

unstable - L 754.310

webbed - L 755.800

Hirschsprung's disease

NOS - 751.330

long-segment - 751.310

short-segment - 751.320

Hirsutism

forehead - # 744.910

other - # 757.450

"Hole in the heart" - 745.900

Holoprosencephaly - 742.260

Holt-Oram syndrome - 759.840

Honeycomb lung - L 748.420

Hooded foreskin - 752.860

Horner syndrome - L 744.880

Horseshoe kidney - 753.320

Humerus

absent (total or partial)

only - L 755.220

with absent radius and ulna - L 755.210

with absent radius, ulna, and hand - L 755.200

hypoplastic - L 755.540

        other specified anomalies - L 755.540
        short - L 755.540
Hurler syndrome - 277.510
Hyaline membrane disease - x
Hydatid of Morgagni cyst - L 752.870
Hydranencephaly - 742.320
Hydrocele, congenital - L # 778.600
Hydrocephaly, without spina bifida
        benign external - x
        communicating - 742.380
        ex-vacuo - x
        non-communicating - 742.380
        other - 742.380
        secondary to intraventricular hemorrhage (IVH) or CNS bleed - x
        unspecified, NOS - 742.390
        with spina bifida - see spina bifida
Hydrocolpos - * 752.430
Hydrocytoma - see skin-benign neoplasm
Hydrometrocolpos - * 752.430
Hydromyelia - 742.540
Hydronephrosis - L 753.200
Hydrophthalmos - L 743.200
Hydrops fetalis - # 778.000
Hydrorachis - 742.540
Hydroureter - L 753.220
Hydroureteronephrosis - L 753.200 and L 753.220
Hymen
        imperforate - * 752.430
        tag - * 752.480
Hyperconvex nail - L 757.580
Hyperextended/hyperextensibility
        arm - L 755.580
        elbow - L 755.540
        finger - L # 755.500
        foot - L 754.780
        hip - L 755.660
        joints - L 755.880
        knee - L 755.640
        leg - L 755.680
        thigh - x
        toe - L # 755.600
Hyperflexion hand - x
Hyperostosis, infantile cortical - 756.530
Hyperpigmentation of skin - # 757.390
Hyperplasia/hyperplastic
        adrenal, congenital
            classical (salt) water - # 255.200
            classical (simple virilizer) - # 255.210
            NOS - # 255.290
            other than 21-OHP deficiency - # 255.240
        kidney - L 753.340
        lung - x
        primary vitreous, persistent - L 743.500
        pulmonary - x
        spleen - # 759.020
Hypertelorism - 756.085
Hypertelorism-hypospadias syndrome - 759.800

Hypertension, primary pulmonary artery - L * 747.680
Hypertrichosis - # 757.450
Hypertrophic/hypertrophy
    bladder - x
    breast - x
    cardiomyopathy - * 746.860
    clitoris - * 752.450
    gum - 750.280
    kidney - L 753.340
    nail - L 757.510
    pyloric stenosis - 750.510
    thymus - * 759.240
    urethra - x
    ventricle/ventricular (heart) - L * 746.886
    ventricular septum - * 746.860
Hypochondrodysplasia - 756.480
Hypochondrogenesis - 756.480
Hypoglossia - 750.110
Hypoglossia-hypodactylia syndrome - 759.840
Hypoglycemia, idiopathic - # 251.200
Hypognathia - 524.000
Hypomelia
    arm - L 755.585
Hypoparathyroidism, congenital - # 252.100
Hypophosphatasia, congenital - # 275.330
Hypophosphatemic rickets - # 275.330
Hypopigmentation of skin - # 757.390
Hypopituitarism, congenital - #253.280
Hypoplasia/hypoplastic
    adrenal gland - L 759.110
    alae nasae - # 748.180
    aorta - 747.210
    aortic valve - 746.480
    arm - L 755.585
    atrium - 746.887
    bladder - 753.880
    brainstem - 742.280
    breast (with hypoplastic nipple) - L 757.610
    cheek - L 744.880
    colon - 751.520
    ear (not microtia) - L * 744.230
    epiglottis - 748.300
    eye - L 743.100
    fallopian tube - L 752.190
    femur - L 755.650
    fibula - L * 755.630
    finger
        all other - L 755.585
        thumb (isolated) - L 755.260
    foot - L 755.685
    gallbladder - 751.630
    gum - x
    hand - L 755.585
    heart, NOS - 746.880
    hip
        bilateral - 755.667
        NOS - 755.665

unilateral - L 755.666
humerus - L 755.540
innominate vein - L 747.650
jugular vein - L 747.650
kidney
       bilateral - 753.000
       NOS - 753.009
       unilateral - L 753.010
labia (majora or minora) - * 752.440
larynx - 748.300
left heart syndrome - 746.700
left ventricle - 746.881
leg - L 755.685
lip - # 744.830
lung - L * 748.510
malar - * 756.080
mandible - 524.000
maxillary - * 756.080
mid-facial - * 756.080
mitral valve - 746.505
muscle - L 756.810
nail - L 757.585
nasal bridge - # 748.180
nipple
       only - L * 757.640
       with hypoplastic breast - L 757.610
nose - 748.100
olfactory nerve - 742.270
ovary - L 752.080
pancreas - 751.700
penis - 752.865
pontine - 742.280
pulmonary
       artery - L 747.380
       lung - L * 748.510
       NOS (heart) - 746.995
       valve - 746.000
radius - L 755.530
rib - L 756.340
right heart - 746.882
right ventricle - 746.882
scrotum - L * 752.810
septum pellucidum - 742.210
spleen - 759.010
sternocleidomastoid muscule - L 754.100
supraorbital ridges - * 756.080
testicle - L * 752.810
thalamus - 742.280
thymus - * 759.240
tibia - L * 755.630
toe
       all other - L 755.685
       first - L 755.365
tricuspid valve - 746.100
ulna - L 755.530
umbilical artery - # 747.500
ureter - L 753.210

ventricle (heart) NOS - 746.883

vertebrae

  cervical - 756.140

  lumbar - 756.160

  NOS - 756.180

  sacral - 756.170

  thoracic - 756.150

Hypospadias

 with chordee

  balantic - 752.625

  coronal - 752.625

  first degree - 752.625

  glandular - 752.625

  NOS - 752.620

  penile - 752.626

  perineal - 752.627

  scrotal - 752.627

  second degree - 752.626

  subcoronal - 752.625

  third degree - 752.627

 without chordee

  balantic - 752.605

  coronal - 752.605

  first degree - 752.605

  glandular - 752.605

  mild - 752.605

  NOS - 752.600

  penile - 752.606

  perineal - 752.607

  scrotal - 752.607

  second degree - 752.606

  subcoronal - 752.605

  third degree - 752.607

Hypotelorism - * 756.080

Hypothalamus anomalies - 742.220

Hypothyroidism

 congenital - # 243.990

 secondary/tertiary - # 244.800


**-I-**

Ichthyosiform erythroderma - 757.197

Ichthyosis congenita

 bullous type - 757.115

 other - 757.190

 unspecified - 757.190

 X-linked - 757.196

Ichthyosis vulgaris - 757.195

Icterus - x

Ileum

 absent - 751.120

 atresia - 751.120

 stenosis - 751.120

Ilium anomalies - L 755.670

Immotile cilia syndrome - 759.340

Imperforate

 anus

with fistula - 751.230
                    without fistula - 751.240
          hymen - * 752.430
          meatus (urethral, urinary) - 753.630
Incontinentia pigmenti - 757.350
Incurving
          finger - L # 755.500
          toe - L # 755.600
Indeterminate sex NOS - * 752.790
Infantile cortical hyperostosis - 756.530
Infantile myofibromatosis - 759.680
Infantile spasms, congenital - # 345.600
Infantile spinal muscular atrophy - 335.000
Infection, congenital (in utero infection)
          cytomegalovirus (CMV) - # 771.100
          herpes simplex - # 771.220
          human immunodeficiency virus (HIV) - x
          other specified - # 771.280
          parvovirus - 771.280
          rubella - 771.000
          syphilis - # 090.000
          TORCH, unspecified - # 771.090
          toxoplasmosis - # 771.200
          varicella - # 052.000
Inferior vermis anomalies - 742.230
Infraorbital crease - L # 743.800
Inguinal hernia
          incarcerated - L * 550.100
          with mention of gangrene - L * 550.000
          with obstruction - L * 550.100
          without obstruction without mention of gangrene - L * 550.900
Iniencephaly
          closed - 740.200
          open - 740.210
          unspecified - 740.290
Innominate artery, aberrant - L 747.640
Innominate vein anomalies - L 747.650
Insufficiency
          aortic valve - * 746.400
          mitral valve - * 746.600
          pulmonary valve - * 746.020
          single atrioventricular valve - 746.900
          tricuspid valve - * 746.105
          truncal valve - 746.900
Integument
          other specified anomalies - 757.800
          unspecified anomalies - 757.990
Intercalary reduction defect
          arm - L 755.210
          leg - L 755.310
          limb, NOS - L 755.410
Interrupted aortic arch - 747.215
Interrupted inferior vena cava - 747.480
Intestine
          adhesion - 751.420
          aganglionosis
                    beyond the rectum - 751.310

involving no more than the anal sphincter and the rectum - 751.320

             total - 751.300

      band - 751.420

      duplication - 751.500

      large

             absent - 751.200

             atresia - 751.200

             malrotation - 751.400

             stenosis - 751.200

      obstruction - x

      other specified anomalies - * 751.580

      small

             absent - 751.190

             absent, with fistula - 751.195

             atresia - 751.190

             atresia, with fistula - 751.195

             malrotation - 751.495

             short - 751.190

             stenosis - 751.190

             stenosis , with fistula - 751.195

      transposition - 751.510

      unspecified anomalies - 751.590

Intracranial cyst - 742.420

Intussusception - x

Inversion

      atrium (heart) - 746.880

      foot - L 754.590

      ventricular - 745.120

Inverted nipples - x

Iris

      absent - L 743.420

      coloboma - L 743.430

      other specified - L 743.440

Ischiopagus conjoined twins - 759.480

Ischium anomalies - L 755.670

Ivemark syndrome - 759.005


**-J-**

Jackson-Weiss syndrome - 756.046

Jacobsen syndrome - 757.300

Jadassohn-Lewandasky syndrome - 759.890

Jarcho Levin syndrome - 756.480

Jaw

      asymmetry - * 756.080

      size abnormalities - 524.000

      shape abnormalities - * 756.080

Jaw-winking syndrome - 742.800

Jejunal/jejunum

      absent - 751.110

      asymmetric - * 756.080

      atresia - 751.110

      stenosis - 751.110

      web - * 751.580

Jeune syndrome - 756.400

Johansen-Blizzard syndrome - 759.870

Joints, hyperextended - L 755.880

Jugular vein
        hypoplastic - L 747.650

**-K-**

Kabuki syndrome - 759.800
Kalischer's disease - 759.610
Kartagener syndrome (triad) - 759.340
Kast syndrome - 756.420
Kawasaki disease - x
Keratitis-ichthyosis-deafness syndrome - 757.190
Keratoglobus - L 743.220
KID syndrome - 757.190
Kidney - see also renal
        absent
                bilateral - 753.000
                NOS - 753.009
                unilateral - L 753.010
        accessory - L 753.300
        agenesis
                bilateral - 753.000
                NOS - 753.009
                unilateral - L 753.010
        cyst (single) - L 753.100
        cystic dysplasia - L 753.160
        cystic NOS - L 753.180
        double (and pelvis) - L 753.310
        dysplasia
                bilateral - 753.000
                NOS - 753.009
                unilateral - L 753.010
        echogenic - x
        ectopic - L 753.330
        fused - 753.320
        giant - L 753.340
        horseshoe - 753.320
        hyperplastic - L 753.340
        hypertrophy - L 753.340
        hypoplasia
                bilateral - 753.000
                NOS - 753.009
                unilateral - L 753.010
        large - L 753.340
        lobulated - 753.320
        malrotated - L 753.330
        medullary cystic disease
                adult type - 753.150
                juvenile type - 753.140
        medullary sponge kidney - 753.150
        multicystic (dysplasia) - L 753.160
        other specified anomalies - L 753.380
        other specified cystic disease - L 753.180
        pelvic - L 753.330
        polycystic
                adult type - 753.120
                autosomal dominant - 753.120
                autosomal recessive - 753.110

infantile type - 753.110
NOS - 753.130
small
bilateral - 753.000
NOS - 753.009
unilateral - L 753.010
triple (and pelvis) - L 753.310
unspecified anomalies - L 753.900
Kinky hair syndrome - 759.870
Klinefelter syndrome
karyotype 47,XXY - 758.700
karyotype 48,XXXY - 758.710
karyotype 48,XXYY - 758.710
karyotype 49,XXXXY - 758.710
NOS - 758.790
other karyotype with additional X chromosomes - 758.710
Klippel-Feil syndrome - 756.110
Klippel-Trenaunay-Weber syndrome - 759.840
Knee
anomalies - L 755.640
dislocation - L 754.440
hyperextended - L 755.640
laxity - L 754.440
subluxation - L 754.440
valgus - L 755.645
webbed - L 755.640
Koilonychia, congenital - 757.520
Kyphomelic dysplasia - 756.480
Kyphoscoliosis - 756.120
Kyphosis - 756.120


**-L-**

Labia (minora or majora)
enlarged - * 752.440
hypoplastic - * 752.440
prominent - * 752.440
Lacrimal aparatus/duct
absent - L 743.640
cyst - L 743.660
obstruction - L # 743.650
other specified - L 743.660
stenosis - L # 743.650
Ladd's bands - 751.420
Lagophthalmos - x
Lambdoidal suture
closed - L 756.020
craniosynostosis - L 756.020
fused - L 756.020
Lanugo, persistent or excessive - # 757.450
Large - see also dilatation
adrenal gland - L 759.180
aorta - 747.270
atrium - x
bladder - x
clitoris - * 752.450
cornea - L 743.220

eye - L 743.210
fontanelle - # 754.040
foot - L 755.610
hand - L 755.510
heart - * 746.860
kidney - L 753.340
labia (minora or majora) - * 752.440
lips - # 744.820
liver - # 751.620
mouth - 744.800
nail - L 757.510
penis - 752.880
pulmonary artery - 747.330
pulmonary valve - 746.080
renal pelvis - L 753.380
septum pellucidum - x
spleen - # 759.020
testicle - 752.820
thymus - * 759.240
tongue - 750.120
tricuspid valve - 746.100
urethra - x
vena cava - 747.480
ventricle (brain) - 742.390
ventricle (heart) - x
uvula - x
Larsen's syndrome - 755.810
Laryngotracheoesophageal  cleft - 748.385
Laryngomalacia - x
Laryngotracheomalacia - x
Larynx/laryngeal
        anomalies of  (and supporting cartilage) - 748.300
        cleft - 748.385
        hypoplastic - 748.300
        other specified anomalies - L 748.380
        stenosis
            NOS - 748.300
            subglottic - * 748.310
        stridor - * 748.360
        subglottic stenosis - * 748.310
        unspecified anomalies - 748.390
        web
            glottic - 748.205
            NOS - 748.209
            subglottic - 748.206
Laurence-Moon-Biedl syndrome - 759.820
Laxity
        hip - x
        knee - L 754.440
Left
        atrioventricular valve - see mitral valve
        semilunar valve - see aortic valve
        superior vena cava - 747.410
Left-sided liver - # 751.620
Leg
        absent - L 755.300
        amelia - L 755.300

amputation, NOS - L 755.385
benign neoplasm - L # 216.700
bowed lower leg - L 754.410
bowed, NOS - 754.420
deformity, NOS - L 754.490
edema, hereditary - 757.000
fused - 759.840
hyperextended - L 755.680
hypoplasia - L 755.685
intercalary reduction defect, NOS - L 755.310
longitudinal reduction defect
     NOS - L 755.360
       postaxial - L 755.366
       preaxial - L 755.365
lymphedema - 757.000
other specified anomalies - L 755.680
other specified reduction defect - L 755.380
phocomelia, NOS - L 755.310
positional deformity - L 755.680
short - L 755.680
short lower leg - L * 755.630
single (fused legs, not one absent) - 759.840
transverse reduction defect, NOS - L 755.385
unspecified anomalies - L 755.690
unspecified reduction defect - L 755.390

Lens
    absent - L 743.300
    coloboma - L 743.340
    displaced - L 743.330
    other specified - L 743.380
    spherical - L 743.310
    unspecified - L 743.390

Lenticonus - L 743.380
Leprechaunism - 759.870
Lethal multiple pterygium syndrome - 759.840
Leukoma cornea - L 743.400
Leukonychia, congenital - 757.530
Levocardia
    only - x
    with situs inversus - 759.310

Limb, NOS
    absent - L 755.400
    amelia - L 755.400
    amputation - L 755.420
    intercalary reduction defect - L 755.410
    phocomelia - L 755.410
    upper - see arm
    longitudinal reduction defect
       NOS - L 755.430
        postaxial - L 755.430
        preaxial - L 755.430
    lower - see leg
    other specified anomalies - L 755.880
    other specified reduction defect - 755.480
    transverse reduction defect, NOS - L 755.420
    unspecified anomalies - L 755.900
    unspecified reduction defect - L 755.490

Limb-body wall complex - 759.840
Lip
       benign neoplasm - # 216.000
       bowed - L 744.880
       cleft
              lateral - 744.800
              with any cleft palate - L 749.200
                     central - 749.220
                     midline - 749.220
              without cleft palate - L 749.100
                     central - 749.120
                     midline - 749.120
       fistula - 750.260
       hpoplastic - # 744.830
       large - # 744.820
       notched - 750.270
       other anomalies - 750.270
       pit - 750.260
       small - # 744.830
       smooth - 750.270
       thin - # 744.830
Lipochondrodystrophy - 277.510
Lipoma
      intra-abdominal organs - L # 214.300
      intrathoracic organs - L # 214.200
      lumbar - # 214.810
      other specified sites - L # 214.800
      paraspinal - # 214.810
      sacral - # 214.810
      skin and cutaneous tissue
          face - # 214.000
          other - # 214.100
      spermatic cord - # 214.400
      unspecified site - # 214.900
Lipomeningocele - see spina bifida
Lipomyelomeningocele - see spina bifida
Lissencephaly - 742.240
Liver
      absent, total or partial - 751.600
      agenesis, total or partial - 751.600
      cyst - 751.610
      cystic disease - 751.610
      fibrocystic disease - 751.610
      enlarged - # 751.620
      fibrosis - 751.610
      hemangioendothelioma liver - L   * 228.040
      left-sided - # 751.620
      other anomalies - # 751.620
      transverse - # 751.620
Lobster-claw
      foot - L 755.350
      hand - L 755.250
Lobulated kidney - 753.320
Lobulated spleen - 759.030
Lop ear - L * 744.230
Long
      arm - x

finger - L # 755.500
foot - L 755.610
hand - L 755.510
head - * 754.030
neck - # 744.900
philtrum - 750.270
skull - * 754.030
sternum - 756.380
toe - L # 755.600
Long QT syndrome - 746.880
Longitudinal reduction defect
    arm
        NOS - L 755.265
        postaxial - L 755.270
        preaxial - L 755.260
    leg
        NOS - L 755.360
        postaxial - L 755.366
        preaxial - L 755.365
    limb, NOS
        NOS - L 755.430
        postaxial - L 755.430
        preaxial - L 755.430
Loose hip - x
Lordosis (postural) - 754.210
Loss of chromosomal material - see deletion (chromosome)
Lowe syndrome - 759.870
Lower leg
    absent
        only - L 755.320
        with absent foot - L 755.330
        with absent thigh - L 755.310
    anomalies - L * 755.630
    bowed - L 754.410
    short - L * 755.630
Lower limb - see leg
Low-lying umbilicus - # 759.900
Low set ears - L # 744.245
Lung
    absent - L 748.500
    accessory lobe - L 748.620
    agenesis - L 748.500
    atresia - L 748.500
    bilobar right - 748.625
    cyst
        multiple - L 748.410
        other specified - L 748.480
        single - L 748.400
    cystic adenomatoid malformation - L 748.480
    ectopic tissues - L 748.600
    emphysema, lobar - L 748.880
    exstrophy - L 748.690
    four or more lobes (right) - L 748.620
    fused lobes - L 748.580
    honeycomb - L 748.420
    hyperplasia - x
    hypoplasia - L * 748.510

incomplete separation of lobes - L 748.580
lobar emphysema - L 748.880
lymphangiectasia - L 748.880
one lobe (left or right) - L 748.580
other specified anomalies - L 748.690
other specified dysplasia - L 748.580
polycystic - L 748.410
right lung with left lung bronchial pattern - 748.625
sequestration - L 748.520
small - L * 748.510
three lobes (right) - x
three or more lobes (left) - L 748.620
two lobes (left) - x
unspecified anomalies - L 748.690
unspecified dysplasia - L 748.590
Lutembacher's syndrome - 745.520
Lymphangiectasis of lung - L 748.880
Lymphangioma (any site) - 228.100
Lymphatics, other specified disorders - # 457.800
Lymphedema
of legs - 757.000
not of legs - x

## -M-

Macrocephaly - * 742.400
Macrocheilia - # 744.820
Macrocolon, not aganglionic - 751.340
Macrocornea - L 743.220
Macrogenitalia (male) - 752.880
Macroglossia - 750.120
Macrognathia - 524.000
Macrostomia - 744.800
Macrotia - L 744.200
Madelung deformity - L * 755.526
Maffucci syndrome - 756.420
Malaligned aorta - 747.260
Malar hypoplasia - * 756.080
Male genitalia (external)
benign neoplasm - # 222.000
other specified anomalies - 752.880
Malrotation
bowel - 751.490
cecum - 751.400
colon - 751.400
kidney - L 753.330
large bowel - 751.400
large intestine - 751.400
midgut - 751.495
other - 751.490
small bowel - 751.495
small intestine alone - 751.495
unspecified - 751.490
Mandible
cleft - * 756.080
hypoplasia - 524.000
Mandibulofacial dysostosis - 756.045

Manibrium, double ossification center - 756.380
Marble bones - 756.540
Marcus Gunn syndrome - 742.800
Marfan syndrome - 759.860
Maxilla
       hypoplasia - * 756.080
       prominent - * 756.080
Meatus/meatal (external auditory, ear)
       absent - L 744.000
       stenosis - L 744.000
       stricture - L 744.000
Meatus/meatal (urethral, urinary)
       atresia - 753.630
       double - 753.840
       imperforate - 753.630
       obstruction - 753.630
       stenosis - 753.630
Meckel-Gruber syndrome - 759.890
Meckel's diverticulum - # 751.010
Meconium
       peritonitis - # 777.600
       plug syndrome - # 777.100
       stained nails - x
       stained skin - x
Mediastinum cyst - 748.810
Medullary cystic disease kidney
       adult type - 753.150
       juvenile type - 753.140
Medullary sponge kidney - 753.150
Megalencephaly - * 742.400
Megalocolon - 751.340
Megalocornea - L 743.220
Megalogastria - 750.710
Megaloureter - L 753.220
Megameatus - 753.880
Megaurethra - 753.880
Melnick-Fraser syndrome - 759.800
Membranous labyrinth (ear) anomalies - L 744.030
Meningocele - see spina bifida
       cervical - 741.085
       occipital - 742.000
       thoracic - 741.086
       lumbar - 741.087
       sacral - 741.087
Meningomyelocele - see spina bifida
       with unspecified hydrocephalus
            cervical - 741.030
            cervicothoracic - 741.030
            lumbar - 741.050
            lumbosacral - 741.050
            sacral - 741.060
            sacrococcygeal - 741.060
            thoracic - 741.040
            thoracolumbar - 741.040
Menkes syndrome - 759.870
Mermaid syndrome - 759.840
Mesentary anomalies - 751.410

Mesenteric remnant cyst - L 752.110
Mesocardia - 746.880
Mesodermal dysgenesis eye - L 743.900
Metaphyseal dysostosis - 756.450
Metatarsus
      adductus - L # 754.520
      varus - L # 754.520
Metatrophic dwarfism - 756.446
Metopic suture
      closed - 756.006
      craniosynostosis - 756.006
      fused - 756.006
Microcephalus - 742.100
Microcheilia - # 744.830
Microcolon - 751.520
Microcoria - L 743.440
Microcornea - L 743.410
Microgastria - 750.700
Microgenitalia (male) - 752.880
Microglossia - 750.110
Micrognathia - 524.000
Microgyria - 742.250
Micromelia
      arm - L 755.580
      leg - L 755.680
Micropenis - 752.865
Microphthalmos - L 743.100
Microsomia
      facial - L 756.065
      hemifacial - L 756.065
Microstomia - 744.810
Microtia - L 744.210
      (hypoplastic pinna and absence or stricture of external auditory meatus)
Midface
      flat - 744.910
      hypoplasia - * 756.080
Midgut malrotation - 751.495
Miller-Dieker syndrome - 759.800
Milroy's disease - 757.000
Misshapen
      rib - L 756.310
      skull - 754.090
      speen - 759.030
      sternum - 756.360
Mitral valve
      abnormal - 746.505
      absent - 746.505
      anomaly - 746.505
      atresia - 746.505
      cleft - 746.505
      double orifice - 746.505
      dysmorphic - 746.505
      dysplastic - 746.505
      hypoplasia - 746.505
      insufficiency - * 746.600
      parachute - 746.505
      prolapse - 746.505

       redundant - x
       regurgitation - * 746.600
       small - 746.505
       stenosis - 746.500
       thickened - 746.500
Moebius syndrome - 352.600
Mohr syndrome - 759.800
Mongolian blue spot - x
Mongoloid slant to eyes - L # 743.800
Monilethix - 757.410
Monodactyly
       hand - L 755.250
       foot - L 755.350
Monorchidism - L 752.800
Monosomy G mosaicism - 758.360
Mosaic
       45,X/46,XX (excludes Turner phenotype) - 758.800
       46,XY/47,XXY (excludes Klinefelter phenotype) - 758.820
       49,XXXXY (excludes Klinefelter phenotype) - 758.830
       Down syndrome - 758.040
       Edwards syndrome - 758.240
       Monosomy G - 758.360
       NOS - 758.900
       Patau syndrome - 758.140
       Turner syndrome - 758.610
       XO/XX (excludes Turner phenotype) - 758.810
       XO/XY (excludes Turner phenotype) - 758.800
       XXXXY (excludes Klinefelter phenotype) - 758.830
       XY/XXY (excludes Klinefelter phenotype) - 758.820
       XYY male - 758.840
Mouth
       abnormal shape - L 744.880
       asymmetry - L 744.880
       carp shape - L 744.880
       downturned - L 744.880
       large - 744.800
       lateral cleft - 744.800
       other specified anomalies - 750.280
       small - 744.810
       unspecified anomalies - 750.900
Mucocele - x
Multicystic (dysplasia)
       kidney - L 753.160
       pancreas - 751.780
       renal - L 753.160
Multiple congenital anomalies - 759.700
Multiple epiphyseal dysplasia - 756.570
Multiple pterygium syndrome - 759.840
Muscle
       absent - L 756.810
          pectoralis major - L 756.810
       atrophy, infantile spinal - 335.000
       atrophy (specified muscle) - L 756.880
       hypoplastic - L 756.810
       other specified anomalies - L 756.880
       sternocleidomastoid - see sternocleidomastoid muscle
       unspecified anomalies - 756.900

Muscle-eye-brain disease - 759.890
Muscular dystrophy, Fukuyama congenital - 759.890
Musculoskeletal system, NOS
       unspecified anomalies - 756.990
Myelocele - see spina bifida
Myelodysplasia - 742.510
Myelomeningocele - see spina bifida
       with unspecified hydrocephalus
              cervical - 741.030
              cervicothoracic - 741.030
              lumbar - 741.050
              lumbosacral - 741.050
              sacral - 741.060
              sacrococcygeal - 741.060
              thoracic - 741.040
              thoracolumbar - 741.040
Myocardial fibrosis - 425.300
Myocardium anomalies - * 746.860
Myofibroma (cardiac) - 425.300
Myofibromatosis, infantile - 759.680
Myopathy, congenital, NOS - L 756.880
Myopia - x
Myotonic dystrophy - 759.890

**-N-**

Nager syndrome - 756.046
Nail
       absent - L 757.500
       club - L 757.540
       duplication - L 757.580
       dysplastic - L 757.580
       dystrophic - L 757.580
       enlarged - L 757.510
       hyperconvex - L 757.580
       hypertrophic - L 757.510
       hypoplastic - L 757.585
       meconium stained - x
       narrow - L 757.585
       other specified anomalies - L 757.580
       short - x
       small - L 757.585
       unspecified anomalies - 757.920
Nail-patella syndrome - 756.830
Nares
       absent - 748.100
       atresia - L 748.000
       small - # 748.180
Narrow/narrowing
       aorta - 747.210
       biparietal - * 756.080
       bitemporal - * 756.080
       chest - 754.820
       hand - x
       nails - L 757.585
       palate - 750.250
       pulmonary artery - L 747.320

temporal - * 756.080
truncal valve - 746.900

Nasal bridge
     broad - # 748.180
     flat - # 748.180
     hypoplasia - # 748.180
     wide - # 748.180

Nasal septum
     absent - # 748.180
     deviation - # 754.020
     perforated - 748.140

Neck
     absent - #  744.900
     anomaly NOS - # 744.900
     benign neoplasm - # 216.400
     broad - # 744.500
     long - # 744.900
     other specified anomalies - L 744.880
     redundant skin folds - # 744.500
     short - # 744.900
     skin folds - # 744.500
     skin tag - L # 744.110
     teratoma - 238.020
     thick - # 744.500
     webbed - # 744.500
     wide - # 744.500

Nephrocalcinosis - x
Nephromegaly - L 753.340
Nephrotic syndrome, congenital - L 753.380

Nervous system
     other specified - 742.880
     unspecified - 742.990

Neu-Laxova syndrome - 759.890
Neurocutaneous melanosis syndrome - 757.300
Neurofibromatosis - 237.700
Neurofibromatosis-Noonan syndrome - 237.700

Nevus - see also skin-benign neoplasm
     blue - see skin-benign neoplasm
     flammeus - # 757.380
     hairy - *216.920
     not elsewhere classified - # 757.380

Nipple
     absent
         only - L 757.630
         with absent breast - L 757.600
     accessory
         only - L # 757.650
         with accessory breast - L 757.620
     asymmetric - # 757.680
     ectopic
         only - L # 757.650
         with ectopic breast - L 757.620
     hypoplastic
         only - L * 757.640
         with hypoplastic breast - L 757.610
     inverted - x
     small - L * 757.640

wide spaced - # 757.680
Noonan syndrome - 759.800
Norrie disease - 759.890
Nose
        absent - 748.100
        accessory - 748.110
        agenesis - 748.100
        asymmetry - # 748.180
        benign neoplasm (external) - # 216.300
        bent - # 754.020
        bifid - 748.120
        broad bridge - # 748.180
        cleft - 748.120
        fissured - 748.120
        flat bridge - # 748.180
        hypoplastic - 748.100
        notched - 748.120
        other specified anomalies - # 748.180
        skin tag - L # 744.110
        small - # 748.180
        tubular - 748.185
        underdevelopment - 748.100
        unspecified anomalies - 748.190
        wide bridge - # 748.180
Nostril
        single - 748.185
        small - # 748.180
Notched lip - 750.270
Nubbin
        finger - L 755.240
        toe - L 755.340
Nuchal folds - # 744.500
Nystagmus - # 379.500

**-O-**

OAV syndrome - 756.060
Obstruction
        alimentary tract, NOS - 751.900
        biliary - x
        bladder outlet - 753.690
        digestive system, NOS - 751.900
        intestinal - x
        lacrimal - L # 743.650
        meatus (urethral, urinary) - 753.630
        pyloric - 750.580
        ureteropelvic junction - L 753.210
        urethra (anterior) - 753.620
        urinary meatus - 753.630
        ventricular outflow tract (left or right) - 746.880
Obstructive uropathy
        at level of bladder or urethra - 753.690
        unilateral - L 753.290
Occipitocervical encephalocele - 742.000
Occiput
        flat - * 756.080
        prominent - * 756.080

short - * 756.080
Occult spina bifida - 756.100
Ochoa syndrome - 759.800
Oculoauriculovertebral dysplasia - 756.060
Oculomandibulofacial syndrome - 756.046
Oeis syndrome - 759.890
OFD syndrome, type I - 759.800
Olfactory nerve
        absent - 742.270
        hypoplastic - 742.270
Oligodactyly
        foot - L 755.340
        hand - L 755.240
        NOS - L 755.440
Ollier syndrome - 756.410
Omentum
        adhesion - 751.420
        band - 751.420
Omphalocele - 756.700
Omphalomesenteric duct - 751.000
Ondine's Curse syndrome - x
Onychauxis - 757.515
Opitz G/BBB syndrome - 759.800
Optic disc/nerve
        atrophy - L 743.520
        coloboma - L 743.520
        hypoplastic - L 743.520
        specified anomalies - L 743.520
Oral-facial-digital syndrome, type I - 759.800
Orbit (eye) anomalies - L 743.670
Orofaciodigital syndrome, type II - 759.800
Oro-mandibular-limb hypogenesis syndrome - 759.840
Organ of Corti anomalies - L 744.030
Ortolani positive hip - L 754.310
Ossicles (ear)
        fusion - L 744.020
Osteochondrodysplasia - 756.490
Osteodystrophy
        other specified - 756.580
        unspecified - 756.590
Osteogenesis imperfecta - 756.500
Osteopenia - x
Osteopetrosis - 756.540
Osteopoikilosis - 756.560
Osteoporosis - x
Osteopsathyrosis - 756.505
Ostium primum defect - * 745.600
Ostium secundum defect - 745.510
Oto-palato-digital syndrome - 759.800
Ovary
        absent - L 752.000
        accessory - L 752.020
        agenesis - L 752.000
        cyst
                multiple - L 752.085
                single - L 752.080
        hypoplastic - L 752.080

other specified anomalies - L 752.080
       streak - L 752.010
       torsion - L 752.080
       unspecified - L 752.090

Overlapping
       digit, NOS - L 755.880
       fingers - L # 755.500
       sutures - x
       toes - L # 755.600

Overriding
       aorta - 747.260
       pulmonary artery - L 747.380
       sutures - x

Ovotestis - 752.700
Oxycephaly - 754.080

**-P-**

Pachygyria - 742.280
Pachyonychia - 757.516
Palate
       absent
              hard - 749.030
              NOS - 749.090
              soft - 749.070
       anterior - see hard
       cleft
              with cleft lip - see cleft lip with any cleft palate
              without cleft lip
                     hard palate (alone) - L 749.000
                           central - 749.020
                           midline - 749.020
                    NOS (hard/soft not specified) - 749.090
                    soft and hard palate - 749.090
                    soft palate (alone) - L 749.040
                           central - 749.060
                           midline - 749.060
                    submucosal
                         hard - 749.020
                         NOS (hard/soft not specified) - 749.090
                         soft - 749.060
       high arched - # 750.240
       narrow - 750.250
       other anomalies - 750.250
       posterior - see soft
       small - 750.250
Palatoschisis - 749.090
Palmar creases
       abnormal - L # 757.200
       simian - L # 757.200
       transverse - L # 757.200
Palpebral fissures
       narrow - L 743.635
       slanting (up-, down-) - L # 743.800
       small - L 743.635
       thick - x
       thin - L 743.635

Palsy
    Bell's - L # 351.000
    brachial plexus - L # 767.600
    Erb's - L # 767.600
    facial - L # 351.000
Pancreas
    absent - 751.700
    accessory - 751.710
    agenesis - 751.700
    annular - 751.720
    cyst - 751.740
    divisum - 751.780
    ectopic - 751.730
    heterotopia - 751.780
    hypoplasia - 751.700
    multicystic - 751.780
    other specified anomalies - 751.780
    small - 751.700
    unspecified anomalies - 751.790
Papilloma - see skin-benign neoplasm
Parachute mitral valve - 746.505
Paraesophageal hernia - 750.600
Paralysis
    diaphragm - L 756.680
    vocal cord - x
Parathyroid gland anomalies - 759.230
Parovarian cyst - L 752.120
Partial anomalous pulmonary venous return - 747.430
Partial foramen ovale - * 745.590
Parvovirus infection, congenital - 771.280
Patau syndrome
    karyotype trisomy 13 - 758.100
    karyotype trisomy D, NOS - 758.110
    NOS - 758.190
    mosaic - 758.140
    translocation trisomy 13 - duplication or a 13 - 758.120
    translocation trisomy 13 - duplication or a D, NOS - 758.130
Patella
    absent - L 755.647
    rudimentary - L 755.647
Patent
    ductus arteriosus - * 747.000
    foramen ovale
        NOS - * 745.500
        vs ASD - * 745.590
        vs secundum ASD - * 745.590
    urachus - # 753.700
Pearson syndrome - 759.870
Pectoralis major muscle, absent - L 756.810
Pectus
    carinatum - 754.800
    excavatum - 754.810
    NOS - 754.820
Pelvicaliectasis - L 753.380
Pelvic kidney - L 753.330
Pelviectasis - L 753.380
Pelvis

anomalies - L 755.670
Pena-Shokier syndrome - L 755.800
Pena-Shokeir II syndrome - 759.840
Penis
     absent - 752.850
     adhesions - 752.860
     aplasia - 752.850
     buried - 752.860
     concealed - 752.860
     disappearing penis syndrome - 752.860
     hypoplastic - 752.865
     large - 752.880
     other anomalies - 752.860
     palmae - 752.860
     small - 752.865
     torsion - 752.860
     webbed - 752.621
Penoscrotal fusion - 752.880
Penoscrotal transposition - 752.880
Penoscrotal web - 752.860
Pentalogy of Cantrell - 759.890
Perforated nasal septum - 748.140
Pericardium anomalies - 746.850
Peripheral arteries, other anomalies - L 747.640
Peripheral pulmonary artery branch stenosis - L * 747.325
Peripheral pulmonary stenosis - L * 747.325
Peripheral vascular system
     other specified anomalies - L * 747.680
     unspecified anomalies - L 747.690
Peripheral veins, other anomalies - L 747.650
Peritoneum
     adhesion - 751.420
     band - 751.420
Peritonitis, meconium - # 777.600
Periventricular cyst - 742.420
Persistent hyperplastic primary vitreous - L 743.500
Persistent omphalomesenteric duct - 751.000
Persistent vitelline duct - 751.000
Pes
     cavus - L 754.700
     planus - L 754.610
     valgus - L 754.615
     varus - L 754.590
Petechiae - x
Peter's anomaly - L 743.440
Peutz-Jegher syndrome - 759.600
Pfeiffer syndrome - 756.057
PHACE syndrome - 759.890
Phalange
     absent (individual)
        finger - L 755.240
        toe - L 755.340
Pharynx/pharyngeal
     other anomalies - L 750.210
     other specified anomalies - 750.280
     pouch - L 750.200
     unspecified anomalies - 750.900

Phenylketonuria (PKU)
       classic - # 270.100
       hyperphenylalaninemia variant - # 270.110
       NOS - # 270.190
Philtrum
       long - 750.270
       prominent - 750.270
       smooth - 750.270
Phimosis - x
Phlebectasia - L 747.630
Phocomelia
       arm - L 755.210
       leg - L 755.310
       limb, NOS - L 755.410
Pierre-Robin sequence - * 524.080
Pigeon chest - 754.800
Pili torti - 757.420
Pilonidal sinus - # 685.100
Pinna
       absent - L 744.010
       accessory - L # 744.100
       benign neoplasm - L # 216.200
       enlarged - L 744.200
       hypoplastic - L 744.210
       large - L 744.200
Piriform aperature
       atresia - L 748.000
       stenosis - L 748.000
Pit
       auricular - L 744.280
       branchial cleft - L 744.400
       ear - L 744.280
       lip - 750.260
       preauricular - L # 744.410
Pituitary gland anomalies - 759.200
Pixie-like - L * 744.230
Plagiocephaly - L * 754.050
Plantar crease, deep - L 755.610
Plantar furrow - L 755.610
Platycephaly - 754.080
Platyspondyly - * 756.180
Pleura anomaly - L 748.800
Pneumothorax - x
Pointed chin - * 756.080
Pointed ear - L * 744.230
Poland syndrome or anomaly - L 756.800
Polycoria - L 743.440
Polycystic
       kidney
              adult type - 753.120
              autosomal dominant - 753.120
              autosomal recessive - 753.110
              infantile type - 753.110
              NOS - 753.130
       lung - L 748.410
Polydactyly
       digit, NOS - L 755.090

finger
>>> Type A - L 755.005
>>> Type B - L * 755.006
>>> NOS - L 755.095
>>> preaxial
>>>> index finger - L 755.010
>>>> thumb - L 755.010
>>> postaxial
>>>> finger - L 755.005
>>>> finger vs skin tag - L 755.007
>>>> NOS - L 755.007
>>>> skin tag - L * 755.006
>> thumb - L 755.010
>> toe
>>> big toe - L 755.030
>>> NOS - L 755.096
>>> preaxial - L 755.030
>>> postaxial - L 755.020
>>> second toe - L 755.030

Polymicrogyria - 742.250
Polyorchidism - 752.820
Polyotia - L # 744.100
Polyploidy - 758.585
Polysplenia - 759.040
Polystotic fibrous dysplasia - 756.510
Polythelia - L # 757.650
Pontine hypoplasia - 742.280
Porencephalic cyst - * 742.410
Porencephaly - * 742.410
Portal vein
> anomalous termination - 747.440
> hepatic artery fistula - 747.450
Port wine stain - # 757.380
Positional deformity
> arm - L 755.580
> foot - L 754.780
> leg - L 755.680
Posterior encephalocele - 742.000
Posterior fossa cyst - 742.230
Posteriorly rotated ears - L # 744.246
Posterior segment of eye
> specified anomalies - L 743.580
> unspecified anomalies - L 743.590
Posterior urethral obstruction - 753.600
Posterior urethral valves - 753.600
Posterolateral diaphragmatic hernia - L 756.615
Potter's facies - 754.010
Potter's syndrome - 753.000
Potter's sequence - 753.000
Pouch
> esophageal - 750.420
> phayngeal - L 750.200
Prader-Willi syndrome - 759.870
Preauricular
> appendage - L # 744.110
> cyst - L # 744.410
> lobule - L # 744.110

pit - L # 744.410
sinus - L # 744.410
tag - L # 744.110
Predislocation of hip - L 754.310
Preluxation of hip - L 754.310
Premature atrial contractions (PACs) - x
Primary pulmonary artery hypertension - L * 747.680
Primary vitreous, persistent hyperplastic - L 743.500
Proboscis - 748.185
Progressive diaphyseal dysplasia - 756.550
Prolapse
       bladder (mucosa) - 753.830
       mitral valve - 746.505
       tricuspid valve - 746.100
Prominent
       clitoris - * 752.450
       eye - L # 743.800
       glabella - # 748.180
       gum - 750.280
       heel - L 755.610
       labia (minora or majora) - * 752.440
       occiput - * 756.080
       philtrum - 750.270
       prepuce of clitoris - x
       renal pelvis - L 753.380
       tongue - x
       xyphoid process - x
Proptosis - L # 743.800
Prostate
       other anomalies - 752.840
Proteus syndrome - 759.890
Protruding/protuberant
       eye - L # 743.800
       tongue - x
Proximal femoral focal deficiency - L 755.380
Prune belly syndrome - 756.720
Pseudocircumcision - x
Pseudocoarctation of aorta - 747.280
Pseudohermaphroditism
       female - 752.720
       male - 752.710
       NOS - 752.730
Pseudotruncus arteriosus - 747.200
Ptosis - L 743.600
Pterygium colli - # 744.500
Pubis anomalies - L 755.670
Pulmonary/pulmonic
       arteriovenous malformation or aneurysm - L 747.340
       artery
              absent - L 747.300
              absent septum between aorta and - 745.000
              agenesis - L 747.300
              aneurysm - 747.330
              atresia
                     without septal defect - L 747.300
                     with septal defect - L 747.310
              collateral vessel involving (but not involving aorta) - L 747.380

dilatation - 747.330
enlarged - 747.330
hypertension, primary - L * 747.680
hypoplasia - L 747.380
narrow - L 747.320
other specified anomalies - L 747.380
overriding - L 747.380
small - L 747.380
stenosis - L 747.320
stenosis, branch - L * 747.325
stenosis, peripheral - L * 747.325
unspecified anomaly - L 747.390
hyperplasia - x
hypoplasia (lung) - L * 748.510
infundibular stenosis - 746.830
insufficiency or regurgitation - * 746.020
NOS (heart)
atresia - 746.995
hypoplasia - 746.995
stenosis - 746.995
subvalvular stenosis - 746.830
supravalvular stenosis - L 747.320
valve
absent - 746.000
atresia - 746.000
bicuspid - 746.080
dilated - 746.080
dysmorphic - 746.080
dysplasia - 746.080
enlarged - 746.080
hypoplasia - 746.000
insufficiency - * 746.020
other specified anomalies - 746.080
redundant - 746.080
regurgitation - * 746.020
small - 746.000
stenosis - 746.010
thickened - 746.080
unspecified - 746.090
vein
atresia - 747.480
stenosis - 747.480
Punctum lacrimale, absent - L 743.640
Pupil - see also iris
ectopic - L 743.440
Pyelectasis - L 753.380
Pyelocaliectasis - L 753.380
Pyelon duplex or triplex - L 753.310
Pyloric
atresia - 751.100
duplication - 751.500
obstruction - 750.580
spasm - # 750.500
stenosis - 750.510
Pylorospasm - # 750.500


**-Q-**

quadricuspid aortic valve - 746.480

**-R-**

Rachischisis - see spina bifida
Radial ray defect, NOS - L 755.260
Radioulnar
 dysostosis - L 755.535
 synostosis - L 755.536
Radius/radial
 absent
  only (total or partial) - L 755.260
  with absent humerus (total or partial) and ulna - L 755.210
  with absent humerus (total or partial), ulna, and hand - L 755.200
  with absent thumb - L 755.260
  with absent ulna - L 755.220
  with absent ulna (total or partial) and hand - L 755.230
 deviation of hand/wrist with no mention of radial defect - L 755.520
 deviation of hand/wrist with mention of radial defect - L 754.840
 fused with ulna - L 755.536
 hypoplastic - L 755.530
 other specified anomalies - L 755.530
 short - L 755.530
Ranula - x
Receding chin - 524.000
Rectourethral fistula - 753.860
Rectovaginal fistula - 752.420
Rectovesical - 753.860
Rectum/rectal
 absent
  with fistula - 751.210
  without fistula - 751.220
 atresia
  with fistula - 751.210
  without fistula - 751.220
 fissure - * 751.580
 fistula - 751.540
 short - 751.220
 small - 751.220
 stenosis
  with fistula - 751.210
  without fistula - 751.220
Red cell aplasia - # 284.000
Reduction defect of the brain
 brainstem - 742.280
 other - 742.280
 unspecified - 742.290
Redundant foreskin - x
Redundant
 mitral valve - x
 pulmonary valve - 746.080
 tricuspid valve - 746.100
Reflux
 gastroesophageal (GER) - x
 vesicoureteral - L 753.485
Regurgitation

aortic valve - * 746.400
mitral valve - * 746.600
pulmonary valve - * 746.020
single atrioventricular valve - 746.900
tricuspid valve - * 746.105
truncal valve - 746.900
Rieger's anomaly - L 743.480
Renal - see also kidney
agenesis, NOS - 753.009
artery
absent - L 747.610
other anomalies - L 747.610
stenosis - L 747.600
calculi - L 753.350
collecting system
dilation (central) - L 753.380
dilation (lower) - L 753.480
dilation (upper) - L 753.480
duplex - L 753.410
cyst (single) - L 753.100
dysplasia, NOS - 753.009
multicystic (dysplasia) - L 753.160
pelvis
dilated - L 753.380
enlarged - L 753.380
extra - L 753.380
other and unspecified obstructive defects - L 753.290
prominent - L 753.380
Renomegaly - L 753.340
Respiratory system
anomaly NOS - 748.900
other specified anomalies - L 748.880
unspecified anomalies - 748.900
Retina
aneurysm - L 743.510
coloboma - L 743.535
degeneration, peripheral - 362.600
hemangioma - L * 228.030
specified anomalies - L 743.510
unspecified anomalies - L 743.590
Retinitis pigmentosa -362.700
Retractile testicle - x
Retrognathia - 524.000
Reversal
complete mirror reversal of abdominal organs with normal thoracic organs - 759.330
complete mirror reversal of all organs - 759.300
complete mirror reversal of thoracic organs with normal abdominal organs - 759.320
Rhabdomyoma
heart - * 746.860
organs other than the heart - 759.680
Rhizomelia
arm - L 755.540
extremity NOS - L 755.880
leg - L 755.650
Rib
absent - L 756.300
bifid - L 756.310

cervical - L # 756.200  
dysplasia - L 756.340  
extra  
    in cervical region - L # 756.200  
    other - L 756.330  
fused - L 756.320  
gracile - L 756.340  
hypoplastic - L 756.340  
less than 12 - L 756.300  
misshapen - L 756.310  
more than 12 - L 756.330  
other anomalies - L 756.340  
rudimentary - L 756.340  
short - L 756.340  
small - L 756.340  
thin - L 756.340  
Rickets, hypophosphatemic - # 275.330  
Rieger's anomaly - L 743.480  
Rieger syndrome - 759.800  
Right  
    aortic arch - 747.230  
    atrioventricular valve - see tricuspid valve  
    lung with left lung bronchial pattern - 748.625  
    semilunar valve - see pulmonary valve  
    superior vena cava, absent -x  
Right sided stomach - 750.730  
Riley-Day syndrome - 742.810  
Ring  
    chromosome  
        X - 758.610  
    vascular - 747.250  
Roberts phocomelia syndrome - 759.840  
Robinow syndrome - 759.800  
Robin sequence - * 524.080  
Rocker-bottom foot - L # 755.616  
Roger's disease - 745.400  
Rokitansky sequence - 759.890  
Rotated ear - L # 744.246  
Rubella  
    congenital (in utero infection) - 771.000  
    syndrome, congenital - 771.00  
Rubenstein-Taybi syndrome - 759.840  
Rudimentary  
    eye - L 743.100  
    finger - L 755.240  
    patella - L 755.647  
    rib - L 756.340  
    toe - L 755.340  
Russell-Silver syndrome - 759.820  

**-S-**

Sacral/sacrococcygeal/sacrum  
    agenesis - 756.170  
    anomalies - 756.170  
    dimple - # 685.100  
    hair tuft - x

hemivertebrae - * 756.170
mass, NOS - 756.179
sinus - # 685.100
teratoma - 238.040
Sacroiliac joint fusion - L 755.670
Saethre-Chotzen syndrome - 756.056
Sagittal suture
closed - 756.005
craniosynostosis - 756.005
fused - 756.005
Salivary glands or ducts, other anomalies - L 750.230
Salmon patches - # 757.380
Scalp defects
aplasia cutis - 757.800
benign neoplasm - # 216.400
NOS - 757.800
Scaphocephaly (no mention of craniosynostosis) - * 754.060
Scaphoid abdomen - x
Scapula anomalies - L 755.550
Schinzel-Giedion syndrome - 759.860
Schizencephaly - 742.280
Schwachman Diamond syndrome - 759.870
Scimitar syndrome - L 748.690
Sclera, blue - L * 743.450
Sclerocornea - L 743.410
Scoliosis
cervical - 754.200
lumbar - 754.200
NOS - 754.200
postural - 754.200
sacral - 754.200
thoracic - 754.200
Scrotum
aplasia - L * 752.810
bifid - 752.820
fused - x
hypoplasia - L * 752.810
other anomalies - 752.820
shawl - 752.820
small - L * 752.810
underdeveloped/undeveloped - L * 752.810
Seckel syndrome - 759.820
Semilunar valve
left - see aortic valve
right - see pulmonary valve
Septal closure - see septal defect
Septal defect (heart)
aortic - 745.010
atrial
NOS - * 745.590
fenestrated - 745.510
fossa ovalis - 745.510
ostium primum - * 745.600
ostium secundum - 745.510
other specified - 745.580
primum - * 745.600
secundum - 745.510

vs PFO - * 745.590
            atrioventricular - see atroventricular canal
            auricular - * 745.590
            other - 745.800
            unspecified - 745.900
            ventricular (heart)
                    apical - 745.480
                    cystalline - 745.480
                    malalignment - 745.480
                    membranous - 745.480
                    mid-muscular - 745.480
                    muscular - 745.480
                    NOS - 745.490
                    other specified - 745.480
                    perimembranous - 745.480
                    septal - 745.480
                    sub-cystalline - 745.480
                    type I - 745.480
                    type II - 745.480
Septo-optic dysplasia - 742.880
Septum pellucidum
        absent - 742.210
        cavum - x
        enlarged - x
        hypoplasia - 742.210
Sequence - see syndrome
Sequestration lung - L 748.520
Sex chromosome
        additional, NOS - 758.860
                see also trisomy
        mosaic - see mosaic
        other specified anomalies - 758.880
        translocation - see translocation
        trisomy - see trisomy
        unspecified anomalies - 758.890
Shawl scrotum - 752.820
Shield chest - 754.825
Shone's complex - 746.880
Short
        achilles tendon - L 754.720
        arm - L 755.580
        colon - 751.520
        esophagus - x
        extremity NOS - L 755.880
        femur - L 755.650
        fibula - L * 755.630
        finger - L # 755.500
        foot - L 755.610
        forearm - L 755.530
        frenulum (tongue, lingual) - # 750.000
        hand - L 755.510
        humerus - L 755.540
        leg - L 755.680
        lower leg - L * 755.630
        nail - x
        neck - # 744.900
        occiput - * 756.080

radius - L 755.530
          rectum - 751.220
          rib - L 756.340
          small intestine - 751.190
          sterum - 756.380
          thigh - L 755.650
          tibia - L * 755.630
          toe - L # 755.600
          ulna - L 755.530
          umbilical cord - # 759.900
          ureter - L 753.480
          vagina - 752.410
Shoulder
          anomalies - L 755.550
          benign neoplasm - L # 216.600
          dislocation - x
Sickle cell disease
          SS - # 282.600
          SC - # 282.630
          other - # 282.690
Simian crease - L # 757.200
Single
          atrioventricular valve - 746.900
          atrium - 745.610
          leg (fused, not one absent) - 759.840
          lung cyst
                  multiple - L 748.410
                  single - L 748.400
          nostril - 748.185
          umbilical artery - # 747.500
          ventricle (heart) - 745.300
Sinus
          branchial cleft - L 744.400
          dermal, of head - L 744.480
          pilonidal - # 685.100
          preauricular - L # 744.410
          sacral - # 685.100
          urachal - # 753.700
Sinus of Valsalva aneurysm - 747.240
Sinus wall (nose) anomalies - 748.130
Sirenomelia - 759.840
Situs ambiguous - * 759.390
Situs inversus
          abdominis - 759.330
          complete, with dextrocardia - 759.300
          thoacis - 759.320
          unspecified - * 759.390
          with levocardia - 759.310
          with sinusitis and bronchitis - 759.340
Sjogren-Larsson syndrome - 757.120
Skeletal dysplasia - 756.490
Skin
          absent - 757.395
          benign neoplasm
                  abdominal wall - #216.500
                  anus - #216.500
                  arm - L # 216.600

auditory canal, external - L # 216.200
auricle - L # 216.200
axillary fold - #216.500
back - #216.500
breast - #216.500
buttock - #216.500
cheek, external - #216.300
chest wall - #216.500
ear - L # 216.200
eyebrow - #216.300
eyelid, including canthus - L # 216.100
face, other and unspecified parts - #216.300
genital organs
    female - #221.000
    male - # 222.000
groin - #216.500
hip - L # 216.700
leg - L # 216.700
lip - # 216.000
neck - # 216.400
nose, external - #216.300
other specified sites - L # 216.800
perianal - # 216.500
perineum - # 216.500
pinna - L # 216.200
scalp - #216.400
shoulder - L # 216.600
temple - # 216.300
trunk - # 216.500
umbilicus - # 216.500
unspecified site - # 216.900
cyst - # 757.390
hemangioma - * 228.010
hyperpigmentation - # 757.390
hypopigmentation - # 757.390
lipoma
    face - # 214.000
    other - # 214.100
meconium stained - x
other specified anomalies - # 757.390
specified syndromes, not elsewhere classified, involving skin anomalies - 757.300
tag
    cheek - L # 744.110
    ear - L # 744.120
    face - L # 744.110
    finger (postaxial) - L * 755.006
    hymen - * 752.480
    neck - L # 744.110
    nose - L # 744.110
    other - # 757.310
    preauricular - L # 744.110
    unspecified - # 757.310
    vagina - * 752.480
unspecified anomalies - 757.900

Skull

asymmetry - 754.055
deformity, NOS - 754.090

depressions - # 754.040
elongated - * 754.030
localized defects - * 756.080
misshapen - 754.090
other specified bone anomalies - * 756.080
other specified deformity (no mention of craniosynostosis) - 754.080
tower - 754.080
unspecified bone anomalies - 756.090
Slanting (up-, down-) palpebral fissures - L # 743.800
Small - see also hypoplastic, stenosis
aorta - 747.210
aortic valve - 746.300
auditory canal - L 744.000
bladder - x
brainstem - 742.280
breast - x
chest - 754.820
chin - 524.000
colon - 751.520
ear (not microtia) - L * 744.230
face - 744.910
finger, all other - L # 755.500
finger, thumb - L # 755.500
fontanelle - # 754.040
foot - L 755.610
gallbladder - x
hand - L 755.510
head - 742.100
kidney
       bilateral - 753.000
       NOS - 753.009
       unilateral - L 753.010
lips - # 744.830
lung - L * 748.510
mitral valve - 746.505
mouth - 744.810
nail - L 757.585
nares - # 748.180
nipple - L * 757.640
nose - # 748.180
nostril - # 748.180
oral cavity - 744.810
palate - 750.250
pancreas - 751.700
penis - 752.865
pulmonary artery - x
pulmonary valve - 746.000
rectum - 751.220
rib - L 756.340
scrotum - L * 752.810
spleen - 759.010
stomach - 750.700
testicle - L * 752.810
thymus - * 759.240
trachea - 748.330
tricuspid valve - 746.100
umbilical cord - # 759.900

uterus - L 752.380
　　　　uvula - x
　　　　vagina - 752.410
　　　　vena cava (inferior or superior) - 747.400
Smith-Lemli-Opitz syndrome - 759.820
Smith-Magenis syndrome - 759.800
Smooth
　　　　lip - 750.270
　　　　philtrum - 750.270
Sotos syndrome - 759.890
Spade-like hand - L 754.850
Spasms, infantile, congenital - # 345.600
Spermatic cord, torsion - L # 608.200
Sphenoid encephalocele - 752.080
Spherophakia - L 743.310
Sphrintzen syndrome - 759.890
Spina bifida
　　　　aperta - see open lesions
　　　　cystica - see closed lesions
　　　　closed lesions (open vs closed not stated)
　　　　　　　　with hydrocephalus
　　　　　　　　　　　　other - 741.080
　　　　　　　　　　　　with aqueductal stenosis, any site - 741.020
　　　　　　　　　　　　with Arnold-Chiari malformation, any site - 741.010
　　　　　　　　　　　　with hydrocephalus of late onset, any site - 741.070
　　　　　　　　　　　　without Arnold-Chiari malformation or aqueductal stenosis
　　　　　　　　　　　　　　　　cervical - 741.030
　　　　　　　　　　　　　　　　cervicothoracic - 741.085
　　　　　　　　　　　　　　　　lumbar - 741.050
　　　　　　　　　　　　　　　　lumbosacral - 741.087
　　　　　　　　　　　　　　　　sacral - 741.060
　　　　　　　　　　　　　　　　site unknown - 741.090
　　　　　　　　　　　　　　　　thoracic - 741.040
　　　　　　　　　　　　　　　　thoracolumbar - 741.086
　　　　　　　　without hydrocephalus
　　　　　　　　　　　　cervical - 741.910
　　　　　　　　　　　　cervicothoracic - 741.980
　　　　　　　　　　　　lipomeningocele - 741.985
　　　　　　　　　　　　lipomyelomeningocele, any site - 741.985
　　　　　　　　　　　　lumbar - 741.930
　　　　　　　　　　　　lumbosacral - 741.980
　　　　　　　　　　　　sacral - 741.940
　　　　　　　　　　　　site unknown - 741.990
　　　　　　　　　　　　thoracic - 741.920
　　　　　　　　　　　　thoracolumbar - 741.980
　　　　occipital - 742.000
　　　　occulta - 756.100
　　　　open lesions
　　　　　　　　with hydrocephalus, any site - 741.000
　　　　　　　　without hydrocephalus, any site - 741.900
Spinal cord
　　　　dysplasia - 742.510
　　　　hypoplasia - 742.510
　　　　other specified - 742.580
　　　　tethered - 742.580
　　　　unspecified - 742.910
Spinal dysraphism

cervical - 756.140
lumbar - 756.160
NOS - 756.180
sacral - 756.170
thoracic - 756.150
Spinal muscular atrophy, infantile - 335.000
Spine
unspecified anomalies - 756.190
Spleen
absent - 759.000
accessory - 759.040
cyst - 759.080
ectopic - 759.050
enlarged - # 759.020
hyperplasia - # 759.020
hypoplasia - 759.010
lobulated - 759.030
misshapen - 759.030
on right in heterotaxy syndrome - 759.050
other specified anomalies - 759.080
small - 759.010
unspecified anomalies - 759.090
Splenomegaly - # 759.020
Split - see also cleft
hand - L 755.250
foot - L 755.350
Spondylocostal dysostoses - 756.480
Spondyloepiphyseal dysplasia - 756.460
Spondylolisthesis - 756.130
Spondylometaphyseal dysplasia - 756.480
Spondylothoracic dysplasia - 756.480
Sprengel's deformity - L 755.556
Squamosal craniosynostosis - 756.000
Square cranium - 754.080
Square head - 754.080
Stenosis
anus
with fistula - 751.230
without fistula - 751.240
aortic - 746.300
aortic valve - 746.300
appendix - 751.200
aqueductal (without spina bifida) - 742.300
bladder neck - 753.610
other and unspecified - 753.690
bronchus - L 748.340
choanal - L 748.000
colon - 751.200
duodenum - 751.100
esophageal - 750.340
hepatic vein - L 747.650
ileum - 751.120
intestine
large - 751.200
small - 751.190
small, with fistula - 751.195
jejunum - 751.110

lacrimal duct - L # 743.650
larynx (not subglottic) - 748.300
meatus (urethral, urinary) - 753.630
meatus (external auditory, ear) - L 744.000
mitral valve - 746.500
piriform aperature - L 748.000
pulmonary
       artery - L 747.320
       artery, branch - L * 747.325
       artery, peripheral - L * 747.325
       infundibular - 746.830
       NOS (heart) - 746.995
       subvalvular - 746.830
       valve - 746.010
       vein - 747.480
pyloric - 750.510
rectum
       with fistula - 751.210
       without fistula - 751.220
renal artery - L 747.600
subglottic - * 748.310
subvalvular aortic - 746.300
subvalvular pulmonary - 746.830
supra-aortic - 747.220
supravalvular aortic - 747.220
supravalvular pulmonary - L 747.320
trachea - 748.330
tricuspid valve - 746.100
truncal valve - 746.900
ureter - L 753.210
ureteropelvic junction - L 753.210
urethral
       anterior - 753.620
       other and unspecified - 753.690
urinary meatus - 753.630
vena cava (inferior or superior) - 747.400

Sternocleidomastoid muscule
    absent - L 754.100
    anomalies - L 754.100
    contracture - L 754.100
    hypoplastic - L 754.100
    tumor - L 754.100

Sternum
    absent - 756.350
    bifid - 756.380
    curved - 754.820
    long - 756.380
    misshapen - 756.360
    other anomalies - 756.380
    short - 756.380
    wide - 756.380

Stickler syndrome - 759.860

Stomach
    absent
       with absent GI tract - 750.780
       with rest of GI tract intact - 750.700
    displacement - 750.730

diverticulum - 750.740
duplication - 750.750
other specified anomalies - 750.780
partial thoracic - 750.600
right sided - 750.730
small - 750.700
transposition - 750.730
unspecified - 750.920
Stork bite - # 757.380
Strabismus, NOS - # 378.900
Streak ovary - L 752.010
Streeter syndrome/dysplasia - # 658.800
Stricture - see also stenosis
       meatus (external auditory, ear) - L 744.000
       ureter - L 753.210
       urethral - 753.690
Stridor, laryngeal - * 748.360
Sturge-Weber syndrome - 759.610
Subclavian artery, aberrant - L 747.640
Subcoronal hypospadias with chordee - 752.625
Subcoronal hypospadias without chordee - 752.605
Subependymal cyst - 742.420
Subglottic
       stenosis - * 748.310
       web - 748.206
Subluxable hip - L 754.310
Subluxation knee - L 754.440
Subluxation of hip - L 754.310
Sunken eye - L # 743.800
Sun-setting eyes - x
Superior vena cava, right, absent - x
Supernumerary - see accessory, extra
Supraorbital ridges, hypoplastic - * 756.080
Suture
       closed
              basilar - 756.030
              coronal - L 756.010
              lambdoidal - L 756.020
              metopic - 756.006
              NOS - 756.000
              other - 756.030
              sagittal - 756.005
       fused
              basilar - 756.030
              coronal - L 756.010
              lambdoidal - L 756.020
              metopic - 756.006
              NOS - 756.000
              other - 756.030
              sagittal - 756.005
       overlapping - x
       overriding - x
Symblepharon - L * 743.630
Symbrachydactyly fingers - L #  755.500 and L 755.190-755.199 (depending on the laterality)
Symbrachydactyly toes - L #  755.600 and L 755.190-755.199 (depending on the laterality)
Symphalangism finger - L # 755.500
Symphalangism toe - L # 755.600

Syndactyly (fused vs webbed unspecified)
     fingers
          bilateral - 755.192
          NOS - 755.193
          unilateral - L 755.191
     NOS - L 755.190
     NOS - 755.199
     toes
          bilateral - 755.195
          NOS - 755.196
          unilateral - L 755.194
Syndrome (also anomaly, association, disease, sequence)
     Aarskog syndrome - 759.800
     Acrocallosal syndrome - 759.890
     Adams-Oliver syndrome - 759.840
     Adrenogenital syndrome - # 255.290
     Agnathia formation syndrome - 759.800
     Aicardi syndrome - 759.890
     Alagille syndrome - 759.870
     Albers-Schonberg syndrome - 756.540
     Albright-McCune-Sternberg syndrome - 756.510
     Alport syndrome - 759.870
     Amniotic band syndrome - # 658.800
     Androgen insensitivity syndrome - 257.800
     Angelman syndrome - 759.890
     Antimongolian syndrome - 758.300
     Apert syndrome - 756.055
     Baller-Gerold syndrome - 759.840
     Bart syndrome - 757.330
     Beals syndrome - 759.860
     Beckwith syndrome - 759.870
     Beckwith-Wiedemann syndrome - 759.870
     Beemer Langer syndrome - 759.860
     Blepharophimosis syndrome - 759.800
     Bloom syndrome - 759.890
     BOR syndrome - 759.800
     Bonneville-Ullrich syndrome, NOS - 758.690
     Bourneville's disease - 759.500
     Branchial arch syndrome - 759.800
     Brown syndrome - # 378.000
     Caffey syndrome - 756.530
     Camurati-Engelmann syndrome - 756.550
     Cardio-splenic syndrome - 759.890
     Carpenter syndrome - 759.840
     Cat eye syndrome - 758.580
     Caudal regression syndrome - 759.840
     Cerebro-oculo-facial-skeletal syndrome - 759.890
     CHARGE association - 759.890
     Chediak-Higashi syndrome - 757.300
     Clifford's syndrome - x
     Cockayne syndrome - 759.820
     Coffin-Siris syndrome - 759.800
     COFS syndrome - 759.890
     Congenital contractural arachnodactyly syndrome - 759.860
     Congenital rubella syndrome - 771.000
     Conradi syndrome - 756.575
     Constriction band syndrome - # 658.800

Cornelia de Lange syndrome - 759.820
Costello syndrome - 759.800
Cri du chat syndrome - 758.310
Crouzon's disease - 756.040
Diamond-Blackfan syndrome (anemia) - # 284.000
Diencephalic syndrome - 253.820
DiGeorge syndrome - 279.110
disappearing penis syndrome - 752.860
distal arthrogryposis syndrome - L 755.800
Down syndrome
       karyotype trisomy 21 - 758.000
       karyotype trisomy G, NOS - 758.010
       mosaic - 758.040
       NOS - 758.090
       translocation trisomy (duplication of a 21) - 758.020
       translocation trisomy (duplication of a G, NOS) - 758.030
Duane syndrome - # 378.000
Du Pan syndrome - 759.840
Eagle-Barrett's syndrome - 756.720
Ebstein's anomaly - 746.200
Ectrodactyly-Ectodermal dysplasia-Clefting syndrome - 759.840
Edwards syndrome
       karyotype normal (Edwards phenotype) - 758.295
       karyotype trisomy 18 - 758.200
       karyotype trisomy E, NOS - 758.210
       mosaic - 758.240
       NOS - 758.290
       translocation trisomy 18 (duplication or an 18) - 758.220
       translocation trisomy 18 (duplication or an E, NOS - 758.230
EEC syndrome - 759.840
Ehlers-Danlos syndrome - 756.850
Eisenmenger's  syndrome - 745.410
Ellis-van Creveld syndrome - 756.525
Engelmann syndrome - 756.550
Escobar syndrome - 759.840
epidermal nevus syndrome - 757.300
Facio-auricular-digital syndrome - 759.800
Facio-auriculo-vertebral syndrome - 756.060
Femoral fibular hypoplasia – unusual facies syndrome - 759.840
Femoral hypoplasia – unusual facies syndrome - 759.840
Femur-fibula-ulna syndrome - 759.840
Fetal Accutane (Isoretinoin) syndrome - 760.760
Fetal alcohol syndrome - 760.710
Fetal Dilantin syndrome - 760.750
Fetal hydantoin syndrome - 760.750
FG syndrome - 759.800
fragile X syndrome - 758.880
Franceschetti syndrome - 756.045
Frasier syndrome - 759.800
Freeman Sheldon syndrome - 759.800
Fryn syndrome - 759.840
Gardner syndrome - 759.630
Gaucher disease Type II - 759.870
Gerbode syndrome - 745.420
Goldenhar syndrome - 756.060
Goltz syndrome - 757.300
Hallermann-Streiff syndrome - 756.046

Heterotaxy syndrome - * 759.390
Holt-Oram syndrome - 759.840
Horner syndrome - L 744.880
Hurler syndrome - 277.510
Hypertelorism-hypospadias syndrome - 759.800
Hypoglossia-hypodactylia syndrome - 759.840
hypoplastic left heart syndrome - 746.700
immotile cilia syndrome - 759.340
Ivemark syndrome - 759.005
Jackson-Weiss syndrome - 756.046
Jacobsen syndrome - 757.300
Jadassohn-Lewandasky syndrome - 759.890
Jarcho Levin syndrome - 756.480
Jaw-winking syndrome - 742.800
Jeune syndrome - 756.400
Johansen-Blizzard syndrome - 759.870
Kabuki syndrome - 759.800
Kalischer's disease - 759.610
Kartagener (triad) syndrome - 759.340
Kast syndrome - 756.420
Kawasaki disease - x
Keratitis-ichthyosis-deafness syndrome - 757.190
KID syndrome - 757.190
kinky hair syndrome - 759.870
Klinefelter syndrome
       karyotype 47,XXY - 758.700
       karyotype 48,XXXY - 758.710
       karyotype 48,XXYY - 758.710
       karyotype 49,XXXXY - 758.710
       NOS - 758.790
       other karyotype with additional X chromosomes - 758.710
Klippel-Feil syndrome - 756.110
Klippel-Trenaunay-Weber syndrome - 759.840
Larsen's syndrome - 755.810
Laurence-Moon-Biedl syndrome - 759.820
Lethal multiple pterygium syndrome - 759.840
Limb-body wall complex - 759.840
Long QT syndrome - 746.880
Lowe syndrome - 759.870
Lutemabcher's syndrome - 745.520
Maffucci syndrome - 756.420
Marcus Gunn syndrome - 742.800
Marfan syndrome - 759.860
Meckel-Gruber syndrome - 759.890
Meconium plug syndrome - # 777.100
Melnick-Fraser syndrome - 759.800
Menkes syndrome - 759.870
Mermaid syndrome - 759.840
Miller-Dieker syndrome - 759.800
Milroy's disease - 757.000
Moebius syndrome - 352.600
Mohr syndrome - 759.800
Multiple pterygium syndrome - 759.840
Muscle-eye-brain disease - 759.890
Nager syndrome - 756.046
Nail-patella syndrome - 756.830
Nephrotic syndrome, congenital - L 753.380

Neu-Laxova syndrome - 759.890
Neurocutaneous melanosis syndrome - 757.300
Neurofibromatosis-Noonan syndrome - 237.700
Noonan syndrome - 759.800
Norrie disease - 759.890
OAV syndrome - 756.060
Ochoa syndrome - 759.800
oculoauriculovertebral syndrome - 756.060
oculomandibulofacial syndrome - 756.046
Oeis syndrome - 759.890
OFD syndrome, type I - 759.800
Ollier syndrome - 756.410
Opitz G/BBB syndrome - 759.800
oral-facial-digital syndrome, type I - 759.800
orofaciodigital syndrome, type II - 759.800
Oro-mandibular-limb hypogenesis syndrome - 759.840
other specified syndromes
      affecting facial appearance - 759.800
      associated with short stature - 759.820
      involving limbs - 759.840
      not elsewhere clasified - 759.890
      with metabolic disturbances - 759.870
      with other skeletal changes - 759.860
Oto-palato-digital syndrome - 759.800
Patau syndrome
      karyotype trisomy 13 - 758.100
      karyotype trisomy D, NOS - 758.110
      mosaic - 758.140
      NOS - 758.190
      translocation trisomy 13 (duplication or a 13) - 758.120
      translocation trisomy 13 (duplication or a D, NOS) - 758.130
Pearson syndrome - 759.870
Pena-Shokier syndrome - L 755.800
Pena-Shokeir II syndrome - 759.840
Peter's anomaly - L 743.440
Peutz-Jegher syndrome - 759.600
Pfeiffer syndrome - 756.057
PHACE syndrome - 759.890
Pierre-Robin sequence - * 524.080
Poland syndrome (anomaly) - L 756.800
Potter's sequence (syndrome) - 753.000
Prader-Willi syndrome - 759.870
Proteus syndrome - 759.890
Prune belly syndrome - 756.720
Rieger's anomaly - L 743.480
Rieger syndrome - 759.800
Riley-Day syndrome - 742.810
Roberts phocomelia syndrome - 759.840
Robinow syndrome - 759.800
Robin sequence - * 524.080
Roger's disease - 745.400
Rokitansky sequence - 759.890
Rubella, congenital syndrome - 771.000
Rubenstein-Taybi syndrome - 759.840
Russell-Silver syndrome - 759.820
Saethre-Chotzen syndrome - 756.056
Schinzel-Giedion syndrome - 759.860

Schwachman Diamond syndrome - 759.870
Scimitar syndrome - L 748.690
Seckel syndrome - 759.820
Shone's complex - 746.880
Short rib-polydactyly syndrome - 756.480
Sjogren-Larsson syndrome - 757.120
Smith-Lemli-Opitz syndrome - 759.820
Smith-Magenis syndrome - 759.800
Sotos syndrome - 759.890
Sphrintzen syndrome - 759.890
Stickler syndrome - 759.860
Streeter syndrome/dysplasia - # 658.800
Sturge-Weber syndrome - 759.610
TAR syndrome - 759.840
Taussig-Bing - 745.100
Tay-Sachs disease - # 330.100
Testicular feminization syndrome - 257.800
Thrombocytopenia-absent radius syndrome - 759.840
Townes-Brock syndrome - 759.890
Treacher-Collins syndrome - 756.045
Turner syndrome
      isochromosome - 758.610
      karyotype 45,X [XO] - 758.600
      mosaic (including XO) - 758.610
      NOS - 758.610
      partial X deletion - 758.610
      ring - 758.610
      variant karyotypes - 758.610
Uhl's syndrome - 746.882
VACTERL association - 759.890
VATER association - 759.890
Velocardiofacial syndrome (VCFS) - 279.110
Von Hipple-Lindau syndrome - 759.620
Von Willebrand syndrome - # 286.400
Waardenburg syndrome - 759.800
Walker-Warburg syndrome - 742.880
Weaver syndrome - 759.890
Werdnig-Hoffman syndrome - 335.00
whistling face syndrome - 759.800
Wiedemann-Beckwith syndrome - 759.870
Wildervanck syndrome - 756.110
Williams syndrome - 759.800
Wilson-Mikity syndrome - x
Wolff-Hirschorn syndrome - 758.320
Wolff-Parkinson-White syndrome - 426.705
Zellweger syndrome - 759.870
Synophrys - L 744.880
Synostosis
    astragaloscaphoid - L 755.620
    cranial - see craniosynostosis
    radioulnar - L 755.536
Synotia - L 744.240
Syphilis, congenital (in utero infection) - # 090.000
Syringoadenoma - see skin-benign neoplasm
Syringohydromyelia - 742.540
Syringomyelia - 742.540

**-T-**

Tag - see skin tag
Talipes
        calcaneovalgus - L 754.600
        calcaneovarus - L 754.510
        equinovalgus - L 754.680
        equinovarus - L 754.500
        NOS - L 754.730
Talipomanus - L 754.840
Taenzer's hair - 757.430
Tarsal bones, absent - L 755.340
TAR syndrome - 759.840
Taussig-Bing syndrome - 745.110
Tay-Sachs disease - # 330.100
Teeth, natal - # 520.600
Telecanthus - 756.085
Temporal narrowing - * 756.080
Tendon
        absent - L 756.820
        other specified anomalies - L 756.880
        unspecified anomalies - 756.910
Teratoma
        abdomen - 238.030
        coccygeal - 238.040
        face - 238.010
        head - 238.010
        neck - 238.020
        NOS - 238.000
        other specified - 238.080
        sacral/sacrococcygeal - 238.040
Testicle/testis
        absent - L 752.800
        aplasia - L * 752.810
        appendix - L 752.870
        atrophy - L * 752.810
        ectopic - L 752.530
        hypoplasia - L * 752.810
        in inguinal canal - see undescended

        large - 752.820
        non-palpable - see undescended
        other anomalies - 752.820
        regression - L 752.800
        retractile - x
        small - L * 752.810
        torsion - L # 608.200
        undescended
            bilateral - * 752.514
            NOS - * 752.520
            unilateral - L * 752.500
        vanishing - L 752.800
Testicular feminization syndrome - 257.800
Tethered spinal cord - 742.580
Tetralogy of Fallot
        with ASD - 745.210
        without ASD - 745.200

Thalami, fused - 742.260
Thalamus, hypoplastic - 742.280
Thanatophoric dwarfism - 756.447
Thick/thickened
        aortic valve - 746.480
        bladder - x
        frenulum - x
        mitral valve - 746.500
        neck - # 744.500
        palpebral fissure - x
        pulmonary valve - 746.080
        tongue - 750.120
        tricuspid valve - 746.100
        urethra - x
        ventricular septum - * 746.860
Thigh
        absent
                with absent lower leg - L 755.310
        hyperextended - x
        short - L 755.650
Thin
        lips - # 744.830
        palpebral fissure - L 743.635
        rib - L 756.340
Thoracic cage
        unspecified anomalies - 756.390
Thoracic-pelvic-phalangeal dysplasia - 756.400
Thorax - see chest
Thrombocytopenia-absent radius syndrome - 759.840
Thumb - see finger
Thymus
        absent - * 759.240
        anomalies - * 759.240
        enlarged - * 759.240
        hypoplastic - * 759.240
        hypertrophy - * 759.240
        small - * 759.240
Thyroglossal cyst - 759.220
Thyroglossal duct anomalies - 759.220
Thyroid gland anomalies - 759.210
Tibia
        absent
                only (total or partial) - L 755.365
                with absent femur (total or partial) and fibula (total or partial)- L 755.310
                with absent femur (total or partial), fibula, and foot - L 755.300
                with absent fibula - L 755.320
                with absent fibula (total or partial) and foot - L 755.330
                with absent first toe (with or without second toe) - L 755.365
        angulation - L * 755.630
        bowed - L 754.410
        hemimelia - L 755.365
        hypoplastic - L * 755.630
        other specified anomalies - L * 755.630
        short - L * 755.630
        torsion - L * 755.630
Tibial ray defect, NOS - L 755.365
Toe

absent
fifth (with or without fourth) - L 755.366
first toe (with or without second toe) - L 755.365
first toe (with or without second toe) and tibia (total or partial) - L 755.365
NOS - L 755.340
third (with or without second, fourth)- L 755.350
with absent long bone leg - L 755.360
acrodactylia - L # 755.600
anomalies - L # 755.600
arachnodactyly - L # 755.600
brachydactyly - L # 755.600
camptodactyly - L # 755.600
clinodactyly - L # 755.600
digitalized (great toe) - L # 755.600
flexion deformity - L # 755.600
fused - L 755.120
hammer - L # 755.600
hyperextension - L # 755.600
hypoplastic
all other - L 755.685
first - L 755.365
incurving - L # 755.600
long - L # 755.600
nubbin - L 755.340
other specified deformities - L 754.780
overlapping - L # 755.600
rudimentary - L 755.340
short - L # 755.600
symbrachydactyly - L # 755.600 and L 755.190-755.199 (depending on the laterality)
symphalangism - L # 755.600
syndactyly, unspecified
bilateral - 755.195
NOS - 755.196
unilateral - L 755.194
triphalangeal (geat toe) - L # 755.600
webbed - L * 755.130
widely spaced first and second - L # 755.600
Tongue
absent - 750.100
cleft - 750.140
cyst - x
dislocation - 750.130
displacement - 750.130
large - 750.120
other specified - 750.180
prominent - x
protruding - x
small - 750.110
thick - 750.120
tie - # 750.000
unspecified - 750.190
Tooth, natal - # 520.600
TORCH infection, unspecified - # 771.090
Torsion
femur - L 755.650
ovary - L 752.080
penile - 752.860

spermatic cord - L # 608.200

testicle - L # 608.200

tibia - L * 755.630

Torticollis - L 756.860

Total anomalous pulmonary venous return - 747.420

Tower head - 754.080

Tower skull - 754.080

Townes-Brock syndrome - 759.890

Toxoplasmosis, congenital (in utero infection) - # 771.210

Trabeculated bladder - x

Trachea

atresia - 748.330

other anomalies - 748.330

small - 748.330

stenosis - 748.330

unspecified anomalies - 748.390

Tracheomalacia - x

Tracheoesophageal

fistula

H type - 750.325

with esophageal atresia - 750.310

without esophageal atresia - 750.320

other anomalies - 750.380

Translocation

balanced autosomal (in normal individual) - 758.400

other (autosome) - 758.540

trisomy 13 - 758.120

trisomy 18 - 758.220

trisomy 21 - 758.020

trisomy D, NOS - 758.130

trisomy E, NOS - 758.230

trisomy G, NOS - 758.030

Transposition of

great arteries

complete - 745.100

corrected - 745.120

incomplete - 745.110

L- - 745.120

other - 745.180

unspecified - 745.190

with inlet VSD - 745.110

with muscular VSD - 745.100 and 745.480

without VSD - 745.100

with perimembraneous VSD - 745.110

with VSD - 745.110

great vessels - see great arteries

penoscrotal - 752.880

stomach - 750.730

Transverse liver - # 751.620

Transverse reduction defect, NOS

arm - L 755.285

leg - L 755.385

limb, NOS - L 755.420

Treacher-Collins syndrome - 756.045

Triangular

face - 744.910

head shape - 754.070

Tricuspid valve
 abnormal - 746.100
 aneurysm - 746.100
 atresia - 746.100
 bicuspid - 746.100
 cleft - 746.100
 dilated - 746.100
 dysplastic - 746.100
 enlarged - 746.100
 hypoplasia - 746.100
 incompetence - * 746.105
 insufficiency - * 746.105
 other specified anomalies - 746.100
 prolapse - 746.100
 redundant - 746.100
 regurgitation - * 746.105
 small - 746.100
 stenosis - 746.100
 thickened - 746.100
Trigonocephaly (no mention of craniosynostosis) - 754.070
Trilogy of Fallot - 746.840
Triphalangeal (thumb) - L # 755.500
Triphalangeal (great toe) - L # 755.600
Triploidy - 758.586
Trisomy
 1 - 758.520
 2 - 758.520
 3 - 758.520
 4 - 758.520
 5 - 758.520
 6 - 758.510
 7 - 758.510
 8 - 758.500
 9 - 758.510
 10 - 758.510
 11 - 758.510
 12 - 758.510
 13 - 758.100
 14 - 758.520
 15 - 758.520
 16 - 758.520
 17 - 758.520
 18 - 758.200
 19 - 758.520
 20 - 758.520
 21 - 758.000
 22 - 758.520
 C, NOS - 758.510
 D, NOS - 758.110
 E, NOS - 758.210
 G, NOS - 758.010
 NOS (autosome) - 758.520
 NOS - 758.910
 other total (autosome) - 758.520
 partial (autosome) - 758.530
 XXX female - 758.850
 XYY male - 758.840

Trophedema, hereditary - 757.000
Truncal valve - 746.900
  insufficiency - 746.900
  narrow - 746.900
  regurgitation - 746.900
  stenosis - 746.900
Truncus arteriosus - 745.000
Tuberous sclerosis - 759.500
Tubular hypoplasia of aorta - 747.210
Tumor
  heart - 746.880
  sternocleidomastoid muscule - L 754.100
Turner syndrome
  isochromosome - 758.610
  karyotype 45,X [XO] - 758.600
  mosaic (including XO) - 758.610
  NOS - 758.610
  partial X deletion - 758.610
  ring - 758.610
  variant karyotypes - 758.610
Turricephaly - 754.080
Twin reversed arterial perfusion (TRAP) sequence - 759.890
Twins
  acardiac - 759.480
  conjoined
    craniopagus (head-joined twins) - 759.410
    dicephalus (two heads) - 759.400
    ischiopagus - 759.480
    other specified - 759.480
    pelvis-joined twins - 759.480
    pygophagus (buttock-joined twins) - 759.440
    thoracopagus (thorax-joined twins) - 759.420
    unspecified - 759.490
    xiphopagus (xiphoid-joined twins) - 759.430
Twisted hair - 757.420
Two vessel umbilical cord - # 747.500
Tympanic membrane anomalies - L 744.020

**-U-**

Uhl's disease - 746.882
Ulna/ulnar
  absent
    only (total or partial) - L 755.270
    with absent humerus (total or partial) and radius - L 755.210
    with absent humerus (total or partial), radius, and hand - L 755.200
    with absent radius - L 755.220
    with absent radius (total or partial) and hand - L 755.230
  bowed without Madelung deformity - L 755.530
  deviation of hand/wrist with no mention of ulnar defect - L 755.520
  deviation of hand/wrist with mention of ulnar defect - L 754.840
  fused with radius - L 755.536
  hypoplastic - L 755.530
  other specified anomalies - L 755.530
  short - L 755.530
Ulnar ray defect, NOS - L 755.270
Umbilical artery hypoplasia - # 747.500

Umbilical cord/umbilicus
    anomalies - # 759.900
    atrophy - # 759.900
    benign neoplasm - # 216.500
    Four vessel - L * 747.680
    hernia - # 553.100
    low-lying - # 759.900
    short - # 759.900
    single artery - # 747.500
    small - # 759.900
    two vessels - # 747.500
Underdevelopment
    nose - 748.100
Undescended testicle
    bilateral - * 752.514
    NOS - * 752.520
    unilateral - L * 752.500
Unicornate uterus - L 752.380
Unstable of hip - L 754.310
Upper
    alimentary tract
        other specified anomalies - 750.800
        unspecified anomalies - 750.990
    arm
        absent
            only - L 755.220
            with absent forearm - L 755.210
        anomalies - L 755.540
    leg - see also thigh
        anomalies - L 755.650
    limb - see arm
Urachus/urachal
    cyst - 753.710
    other and unspecified anomaly - 753.790
    patent - # 753.700
    remnant - 753.790
    sinus - # 753.700
Ureter
    absent - L 753.400
    accessory - L 753.410
    atresia - L 753.210
    dilated - L 753.220
    double - L 753.410
    ectopic - L 753.420
    hypoplastic - L 753.210
    other and unspecified obstructive defects - L 753.290
    other specified anomalies - L 753.480
    short - L 753.480
    stenosis - L 753.210
    stricture - L 753.210
    unspecified anomalies - L 753.910
Ureterectasis - L 753.220
Ureterocele - L 753.480
Ureteropelvic junction
    obstruction - L 753.210
    stenosis - L 753.210
Ureterovesical junction - see ureteropelvic junction

Urethra/urethral
     absent - 753.800
     anterior
          atresia - 753.620
          obstruction - 753.620
          stenosis - 753.620
          valve - 753.620
     diverticulum - 753.880
     double - 753.840
     ectopic - 753.850
     enlarged - x
     fistula, NOS - 753.870
     hypertrophy - x
     obstruction (posterior) - 753.600
     orifice
          ectopic - 753.850
     other and unspecified atresia and stenosis - 753.690
     other specified anomalies - 753.880
     stricture - 753.690
     thickened - x
     unspecified anomalies - 753.930
     valves (posterior) - 753.600
Urethrorectal fistula - 753.860
Urinary meatus
     atresia - 753.630
     double - 753.840
     obstruction - 753.630
     stenosis - 753.630
Urinary system/tract
     fistula with digestive system - 753.860
     unspecified anomalies - 753.990
Urogenital sinus malformation - 753.880
Uropathy, obstructive
     at level of bladder or urethra - 753.690
     unilateral - L 753.290
Urticaria pigmentosa - 757.320
Uterointestinal fistula - 752.320
Uterovesical fistula - 752.320
Uterus
     absent - 752.300
     agenesis - 752.300
     bicornate - L 752.380
     didelphys - 752.200
     displaced - 752.310
     doubling - 752.200
     fistula (with digestive or urinary tract) - 752.320
     other anomalies - L 752.380
     septate - L 752.380
     small - L 752.380
     unicornate - L 752.380
     unspecified anomalies - 752.390
Uvula
     absent - 749.080
     bifid - 749.080
     cleft - 749.080
     enlarged - x
     small - x

**-V-**

VACTERL association - 759.890
Vagina
        absent (complete or partial) - 752.410
        agenesis (complete or partial) - 752.410
        atresia (complete or partial) - 752.410
        cyst
                embryonal - # 752.460
                other - 752.470
        doubling - * 752.480
        other specified anomalies - * 752.480
        short - 752.410
        small - 752.410
        tag - * 752.480
        unspecified anomalies - 752.490
Vaginocele - * 752.480
Valga/valgum/valgus
        coxa - L 755.660
        cubitus - L 755.540
        genu - L 755.645
        hallux - L 755.605
        knee - L 755.645
        other specified deformities of foot - L 754.680
        pes - L 754.615
        unspecified deformities of foot - L 754.690
Valve
        aortic
                absent - 746.480
                atresia - 746.480
                bicuspid - * 746.400
                dysmorphic - 746.480
                dysplastic - 746.480
                hypoplastic - 746.480
                incompetence - * 746.400
                insufficiency - * 746.400
                other specified - 746.480
                quadricuspid - 746.480
                regurgitation - * 746.400
                small - 746.300
                stenosis - 746.300
                thickened - 746.480
                unspecified - 746.490
        mitral
                absent - 746.505
                anomaly - 746.505
                atresia - 746.505
                cleft - 746.505
                dysmorphic - 746.505
                dysplastic - 746.505
                hypoplasia - 746.505
                insufficiency - * 746.600
                parachute - 746.505
                prolapse - 746.505
                redundant - x
                regurgitation - * 746.600

              small - 746.505
              stenosis - 746.500
              thickened - 746.500
        pulmonary
              absent - 746.000
              atresia - 746.000
              bicuspid - 746.080
              dilated - 746.080
              dysmorphic - 746.080
              dysplasia - 746.080
              enlarged - 746.080
              hypoplasia - 746.000
              insufficiency - * 746.020
              other specified anomalies - 746.080
              redundant - 746.080
              regurgitation - * 746.020
              small - 746.000
              stenosis - 746.010
              thickened - 746.080
              unspecified - 746.090
        tricuspid
              abnormal - 746.100
              aneurysm - 746.100
              atresia - 746.100
              bicuspid - 746.100
              cleft - 746.100
              dilated - 746.100
              dysplastic - 746.100
              enlarged - 746.100
              hypoplasia - 746.100
              incompetence - * 746.105
              insufficiency - * 746.105
              other specified anomalies - 746.100
              prolapse - 746.100
              redundant - 746.100
              regurgitation - * 746.105
              small - 746.100
              stenosis - 746.100
              thickened - 746.100
              unspecified anomalies - 746.900

Vanishing testicle - L 752.800
Varix- L 747.630
Vara/Varum/varus
      complex deformities - L 754.530
      coxa - L 755.660
      genu - L 755.646
      hallux - L 755.606
      metatarsus - L # 754.520
      unspecified (of feet) - L 754.590
Varicella, congenital (in utero infections) - # 052.000
Vascular ring - 747.250
Vas deferens
      atresia - L 752.830
      other anomalies - 752.840
VATER association - 759.890
Vein of Galen anomalies - L 747.810
Velocardiofacial syndrome (VCFS) - 279.110

Vena cava
        absent (except left superior) - 747.480
        bilateral inferior - 747.480
        bilateral superior - 747.410
        dilated - 747.480
        enlarged - 747.480
        interrupted inferior - 747.480
        left superior - 747.410
        small (inferior or superior) - 747.400
        stenosis (inferior or superior) - 747.400
Ventri in version - 745.120
Ventricle/ventricular (brain)
        cyst - * 742.485
        dilatation - 742.390
        enlarged - 742.390
Ventricle/ventricular (heart)
        common - 745.300
        dilatation - x
        double inlet left - 745.300
        double inlet right - 745.300
        double outlet left - 745.180
        double outlet right - 745.180
        enlarged - x
        hypertrophy - L * 746.886
        hypoplastic left - 746.881
        hypoplastic NOS - 746.883
        hypoplastic right - 746.882
        inversion - 745.120
outflow tract obstruction (left or right) - 746.880
        septal defect
                apical - 745.480
                cystalline - 745.480
                hypertrophy - * 746.860
                malalignment - 745.480
                membranous - 745.480
                mid-muscular - 845.480
                muscular - 745.480
                NOS - 745.490
                other specified - 745.480
                perimembranous - 745.480
                septal - 745.480
                sub-cystalline - 745.480
                thickened - * 746.860
                type I - 745.480
                type II - 745.480
        single - 745.300
Ventriculomegaly - 742.390
Vermian atrophy - 742.230
Vermis (inferior) anomalies - 742.230
Vertebra
        cervical
                agenesis - 756.146
                anomalies - 756.140
                bifid - 756.140
                butterfly - 756.140
                cleft - 756.140
                fused - 756.140

hypoplastic - 756.140
segmentation anomalies - 756.140
lumbar
agenesis - 756.166
anomalies - 756.160
bifid - 756.160
butterfly - 756.160
cleft - 756.160
fused - 756.160
hypoplastic - 756.160
segmentation anomalies - 756.160
NOS
bifid - 756.180
butterfly - 756.180
cleft - 756.180
fused - 756.180
hypoplastic - 756.180
other specified anomalies - 756.180
segmentation anomalies - 756.180
unspecified anomalies - 756.190
sacral/sacrum
agenesis - 756.170
anomalies - 756.170
bifid - 756.170
butterfly - 756.170
cleft - 756.170
fused - 756.170
hypoplastic - 756.170
segmentation anomalies - 756.170
thoracic
agenesis - 756.156
anomalies - 756.150
bifid - 756.150
butterfly - 756.150
cleft - 756.150
fused - 756.150
hypoplastic - 756.150
segmentation anomalies - 756.150
Vertical talus foot - L # 755.616
Vesicoureteral reflux - L 753.485
Vesicovaginal fistula - 752.420
Vesiculobullous dermatosis - x
Vitelline duct - 751.000
Vitreous humor anomalies - L 743.500
Vocal cord paralysis - x
Volvulus
gastric - x
intestinal - x
Von Hipple-Lindau syndrome - 759.620
Von Willebrand disease - # 286.400
Vulva
absent - * 752.440
cyst - 752.470
fused - * 752.440
other anomaly - * 752.440


**-W-**

Waardenburg syndrome - 759.800
Walker-Warburg syndrome - 742.880
Weaver syndrome - 759.890
Web/webbed
       duodenal - 751.560
       elbow - L 755.800
       esophageal - 750.350
       fingers - L 755.110
       hip - L 755.800
       jejunal - * 751.580
       knee - L 755.640
       larynx
              glottic - 748.205
              NOS - 748.209
              subglottic - 748.206
       neck - # 744.500
       penis - 752.621
       penoscrotal - 752.860
       toes - L * 755.130
Werdnig-Hoffman disease - 335.000
Werner mesomelic dysplasia - 756.480
Wharton duct cyst - x
Whistling face syndrome - 759.800
White forelock - # 757.390
Widely spaced first and second toes - L # 755.600
Widely spaced nipples - # 757.680
Wide neck - # 744.500
Wide set eyes - 756.085
Wide sternum - 756.380
Wiedemann-Beckwith syndrome - 759.870
Wildervanck syndrome - 756.110
Williams syndrome - 759.800
Wilson-Mikity syndrome - x
Wolff-Hirschorn syndrome - 758.320
Wolff-Parkinson-White syndrome - 426.705
Wolffian duct cyst - L 752.870
Wrist
       anomalies - L 755.520
       flexed - L 755.520
       ulnar deviation - L 755.520

**-X-**

Xeroderma pigmentosum - 757.360
XK aprosencephaly - 759.800
Xyphoid process
       bifid - 756.380
       prominent - x

**-Y-**

**-Z-**

Zellweger syndrome - 759.870

**-KARYOTYPES-**

45,X - 758.600
45,X,inv(9) - 758.600
45,X/46,X+mar - 758.610
45,X/46,X,9(X)(q10) - 758.610
45,X/46,X,r - 758.610
45,X/46,X,r(X)(p22.3;q24) - 758.610
45,X/46,XX (with Turner syndrome phenotype) - 758.610
45,X/46,XX (without Turner syndrome phenotype) - 758.810
45,X/46,XX/46,X,+15/47,XX+15 - 758.520
45,X/46,XX/46,X,+15/47,XX+15 - 758.880
45,X/46,XY - 758.800
45,X/46X,r(X) - 758.610
45,X/47,XXX - 758.610
45,XX,der(14;21)(q10;q10)/46,XX,der(14;21)(q10;q10),+21 - 758.040
45,XX,der(14;21)(q10;q10)/46,XX,der(14;21)(q10;q10),+21 - 758.020
45,XX,der(5)(5;15)(p15.3;q13),-15.15Hder(5)(PML+D15S10-,141-,D5S23+74+) - 758.540
45,XX,der(7)+(7;21)(q35;q10)-21 - 758.540
45,XX,der,(16;22)(p13.3;q11,2),-22 - 758.380
45,XY,?dic(7;20)(p22;?p13)/46,XY,?dic(7;20)(p22;?p13),+mar - 758.540
46 XY,+21,der(21;21)(q10;q10) - 758.020
46,del(13q) - 758.330
46,X,del(X)(p22.2) - 758.610
46,X,i(Xq) - 758.610
46,X,inv(Y)+mar - 758.580
46,XX,+21,der(21;21) - 758.020
46,XX,+21,der(21;21)(q10;q10) - 758.020
46,XX,-14,+t(13;14)/45XX,-14,+t(13;14) - 758.120
46,XX,-18,+der(18)+(18;?)(q12.3;?),var(14) - 758.530
46,XX,-18,+der(18)+(18;?)(q12.3;?),var(14) - 758.340
46,XX,-20,+mar - 758.580
46,XX,-21,+t(21q;21q) - 758.020
46,XX,?del(15)(q11.2)/47,XX+mar/46,XX - 758.380
46,XX,?del(15)(q11.2)/47,XX+mar/46,XX - 758.580
46,XX,9,qh+ - 758.580
46,XX,add(4)(p16) - 758.530
46,XX,add(4)(p16) - 758.530
46,XX,add(6)(p15.1) - 758.530
46,XX,add(8)(p23) - 758.530
46,XX,del(1)(p22) - 758.380
46,XX,del(1)(p36.3) - 758.380
46,XX,del(1)(p36.3),inv(9)(p11;q12)ish del(1)(P36.3)(P58-,D1Z2) - 758.380
46,XX,del(11)(q23) - 758.380
46,XX,del(14;21)(q10;q10)mat,+21 - 758.020
46,XX,del(15)(q11.2;q13) - 758.380
46,XX,del(17)(p11.2;p13) - 758.380
46,XX,del(18)(p11.2) - 758.350
46,XX,del(18)(q?21.1) - 758.340
46,XX,del(22)(q11.2;q11.2) - 758.380
46,XX,del(22)(q11.2;q11.2)(D22575-) - 758.380
46,XX,del(22)(q11.2;q11.2)(D22S75) - 758.380
46,XX,del(3)(q23;q25 or q25;q26.2)DISH del(3)(WCP3+) - 758.380
46,XX,del(4)(q32.1) - 758.380
46,XX,del(5)(p14) - 758.310
46,XX,del(9)(p22) - 758.380
46,XX,del(9)(p22-pter) - 758.380
46,XX,der(?18)t(13;18)(?q11;?p11.1).ISH 46,XX,der(18)t(13;18)(q12;p11.2)(D18Z1+) - 758.120

46,XX,der(13)+(2;13)(q37.1;q32.2) - 758.540
46,XX,der(13)t(13;?)(q;?) - 758.330
46,XX,der(13)t(13;?)(q;?) - 758.530
46,XX,der(14)+(14;17)(p12;p11.2)pat.ISH der(14)+(14;17)(p12;p11.2)(D17S29-)pat - 758.380
46,XX,der(14)+(14;17)(p12;p11.2)pat.ISH der(14)+(14;17)(p12;p11.2)(D17S29-)pat - 758.530
46,XX,der(14;21)(q10;q10),+21 - 758.020
46,XX,der(15)t(15.15)(p13;q26.1) - 758.580
46,XX,der(21)+(5:21) - 758.530
46,XX,der(21)+(5:21) - 758.540
46,XX,der(21:21)(q10;q10),+21 - 758.020
46,XX,der(5)+(5;10)(p15.1;p11.21).ISH der(5)+(5;10)(p15.1;p11.21)WCP 10+,D5S23-) - 758.540
46,XX,der(7) - 758.580
46,XX,der(8p) - 758.580
46,XX,dup(4)(q28;q33) - 758.530
46,XX,dup(5)(q11.2;q12) - 758.530
46,XX,inv(2) - 758.580
46,XX,inv(3)(?p13;?q21) - 758.580
46,XX,inv(6)(p21.3;q15)pat - 758.580
46,XX,inv(9)(p11;q12),r(13)(p11.2;q22)/45,XX,inv(9)(p11;q12),-13 - 758.380
46,XX,inv(9)(p12;q13) - 758.580
46,XX,inv(9),(p11;q13) - 758.580
46,XX,inv(9),(p12;q13) - 758.580
46,XX,ish del(15)(q11.2;q11.2)(SNRPN-) - 758.380
46,XX male - 758.880
46,XX,r(22)(p11.2;q13.3) - 758.580
46,XX,r(8) - 758.580
46,XX,rob(21q;21q) - 758.020
46,XX,t(11;12)(q22.1;q23) - 758.400
46,XX,t(14;21)(q10;q10),+21 - 758.020
46,XX,t(14q;21q) - 758.020
46,XX,t(21;21)(q10;q10),+21 - 758.020
46,XX,t(21q;21q) - 758.020
46,XX,t(6;7)(p22.2;15.3) - 758.540
46,XX,t(9:13)(q22;q14)pat - 758.400
46,XX.ISH del(22)(q11.2;q11.2)(D22S75-) - 758.380
46,XX.ISH del(22)(q11.2;q11.2)(F5-)/46,X,fra(X)(q27.3).ISH del(22)(q11.2;q11.2)(F5-_ - 758.880
46,XX.ISH del(22)(q11.2;q11.2)(F5-)/46,X,fra(X)(q27.3).ISH del(22)(q11.2;q11.2)(F5-_ - 758.380
46,XX.ISH del(22q11.2;q11.2)(D22575-) - 758.380
46,XX/45,X - 758.610
46,XX/46,XX,fra(X)(q28) - 758.880
46,XX/47,XX,+13 - 758.100
46,XX/47,XX,+21 - 758.040
46,XXI(18)(q10) - 758.220
46,XY,+13,der(13;13)(q10;q10) - 758.120
46,XY,+13,der(13;14)(q10;q10) - 758.120
46,XY,+21,der(21;21)(q10;q10) - 758.020
46,XY,+21,der(21;21)(q10;q10)de novo - 758.020
46,XY,-10,der(10)t(3;10)(p25;q26)mat - 758.400
46,XY,-14,+der(14)rob(13q;14q) - 758.120
46,XY,-14,+t(13:14)(p11;q11) - 758.120
46,XY,-14,+t(14q;21q) - 758.020
46,XY,-21,+der(21) - 758.000
46,XY,-21,+t(21q;21q) - 758.020
46,XY,?del(7)(q36) - 758.380
46,XY,1qh+ - 758.580
46,XY,add(20)(p16qh+) - 758.530
46,XY,add(8)(p23) - 758.530

46,XY,del(13p) - 758.380
46,XY,del(15)(q11.2;q13) - 758.380
46,XY,del(22)(q11.2).ISH del(22)(q11.2;q11.2)(D22S7S-) - 758.380
46,XY,del(3)(q21;q23) - 758.380
46,XY,del(4)(p15.2) - 758.320
46,XY,del(5)(p14.1) - 758.310
46,XY,del(6)(q25.1;q25.31) - 758.380
46,XY,del(p13) - 758.310
46,XY,der(13)+(13:?)(q32:?) - 758.530
46,XY,der(13)+(13:?)(q32:?) - 758.540
46,XY,der(13:13)(q10;q10),+13 - 758.120
46,XY,der(13;14)(q10;q10)+14/45,XY,der(13;14)(q10;q10) - 758.520
46,XY,der(14:21)(q10;q10),+21 - 758.020
46,XY,der(14;21)(q10,q10) - 758.020
46,XY,der(21;21)(q10;q10),+21 - 758.020
46,XY,der(4)(p15.3).ISH DER (4) (WCP4-, D4F26-, D4596-) - 758.320
46,XY female, with a diagnosis of androgen insensitivity - 257.800
46,XY female, without a diagnosis of androgen insensitivity - 758.880
46,XY,inv(1)(p32;q31),3+der(3)+(1;3)(q31;p24) - 758.400
46,XY,inv(12) - 758.580
46,XY,inv(9)(p11;q12) - 758.580
46,XY,inv(9)(p12;q13) - 758.580
46,XY,inv(9)(p12;q13)mat,17 CHEV,+pat - 758.580
46,XY,inv(9)(pg12a13) - 758.580
46,XY,ISH del(22)(q11.2;q11.2)(D22S75-) - 758.380
46,XY,rob(14q;21q) - 758.540
46,XY,rob(14q;21q) - 758.020
46,XY,t(14q;21q) - 758.020
46,XY,t(16;17)(q13;q23) - 758.540
46,XY,t(17;19)(q21.2;q13.2) - 758.400
46,XY,t(3;18)(p13;q23) - 758.400
46,XY,t(4;14) - 758.400
46,XY,var(15)(q11.2) - 758.580
46,XY,var(15q) - 758.580
46,XY,var(22) - 758.580
46,XY,var21(+p) - 758.580
46,XY.ISH del(22)(q11.2;q11.2)(D22S75-) - 758.380
46,XY.ISH del(22)(q11.2;q11.2)(D22S75-) - 758.380
46,XY.ISH del(22)(q11.2)(D22S75X2) - 758.380
46,XY/45,X - 758.800
46,XY/45,XY,-14-18,+der(14)+(14;18) (q11.1;p11.2) - 758.540
46,XY/45,XY,-14-18,+der(14)+(14;18)(q11.1;p11.2) - 758.380
46,XY/45,XY,-19 - 758.380
46,XY/46,XY,-20,+der(20) - 758.580
46,XY/47,XXY (without Klinefelter syndrome phenotype) - 758.820
46,XY/47,XY+mar - 758.580
46,XY/47,XY,+16 - 758.520
46,XY/47,XY,+18 - 758.200
46,XY/47,XY,+21 - 758.040
46,XY/49,XXXXY (without Klinefelter syndrome phenotype) - 758.830
46,Y,der(X) - 758.880
47,X,fra(X)(q27.3)/47,XX,+21 - 758.000
47,XX,+(15;17)(q11.2?;q25),+18 - 758.200
47,XX,+1 - 758.520
47,XX,+10 - 758.510
47,XX,+11 - 758.510
47,XX,+12 - 758.510

47,XX,+13 - 758.100
47,XX,+13,inv(9)(pg12;q13) - 758.100
47,XX,+13,inv(9)(pg12;q13) - 758.580
47,XX,+14 - 758.520
47,XX,+15 - 758.520
47,XX,+16 - 758.520
47,XX,+17 - 758.520
47,XX,+18 - 758.200
47,XX,+19 - 758.520
47,XX,+2 - 758.520
47,XX,+20 - 758.520
47,XX,+20(p10)/46,XX - 758.520
47,XX,+21 - 758.000
47,XX,+21,16(qh+) - 758.000
47,XX,+21,16qht - 758.000
47,XX,+21/46,XX - 758.040
47,XX,+21;inv(9)(p11;q12) - 758.000
47,XX,+22 - 758.520
47,XX,+3 - 758.520
47,XX,+4 - 758.520
47,XX,+5 - 758.520
47,XX,+6 - 758.510
47,XX,+7 - 758.510
47,XX,+8 - 758.500
47,XX,+8/46,XX - 758.500
47,XX,+9 - 758.510
47,XX,9qht,+21 - 758.000
47,XX,inv(9)(p11;q13),+21/46,XX,inv(9)(p11;q13) - 758.040
47,XX,inv(9)(p11;q13),+21/46,XX,inv(9)(p11;q13) - 758.580
47,XX,t(7;8)(q11.2;p21.1)+21 - 758.540
47,XX,t(7;8)(q11.2;p21.1),+21 - 758.000
47,XXX - 758.850
47,XXY - 758.700
47,XXY/46,XY - 758.820
47,XY,+1 - 758.520
47,XY,+10 - 758.510
47,XY,+11 - 758.510
47,XY,+12 - 758.510
47,XY,+13 - 758.100
47,XY,+14 - 758.520
47,XY,+15 - 758.520
47,XY,+16 - 758.520
47,XY,+17 - 758.520
47,XY,+18 - 758.200
47,XY,+18,inv(9)(p11;q12) - 758.200
47,XY,+19 - 758.520
47,XY,+2 - 758.520
47,XY,+20 - 758.520
47,XY,+21 - 758.000
47,XY,+21,+22 PSS - 758.000
47,XY,+21,1qht - 758.000
47,XY,+21/46,XY - 758.040
47,XY,+22 - 758.520
47,XY,+8/46,XY - 758.500
47,XY,+3 - 758.520
47,XY,+4 - 758.520
47,XY,+5 - 758.520

47,XY,+6 - 758.510
47,XY,+7 - 758.510
47,XY,+8 - 758.500
47,XY,+9 - 758.510
47,XY,+del(18)(q21.2) - 758.200
47,XY,+der(22) - 758.530
47,XY,+der(22)t(11,22)(q23;q11)mat - 758.530
47,XY,+mar - 758.580
47,XY,i(21)(q10)+mar - 758.580
47,XY,i(21)(q10)+mar - 758.020
47,XY,inv(2)(p11.2;q13),+21 - 758.000
47,XY,inv(9)(p11;q12),+21 - 758.000
47,XY,inv(9)(p11;q12),+21 - 758.000
47,XY,t(2;9)(p25.1;q34.11),+21 - 758.020
47,XYY - 758.840
47,XYY/46,XY - 758.840
48,XXXY - 758.710
48,XXYY - 758.710
48,XY,+21,+mar(pat) - 758.000
48,XY,+21,+mar(pat) - 758.580
49,XXXXY - 758.710
69,XXX - 758.586
69,XXY - 758.586

## Appendix 5.2
## 6-Digit CDC Codes

BIRTH DEFECTS AND GENETIC DISEASES BRANCH 6-DIGIT CODE

For Reportable Congenital Anomalies

Based on the 1979 British Pediatric Association (BPA) Classification of Diseases and the World Health Organization's 1979 International Classification of Diseases, 9th Revision, Clinical Modification (ICD-9-CM)

Code modifications developed by Division of Birth Defects and Developmental Disabilities, National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention, Public Health Service, U.S. Department of Health and Human Services, Atlanta, Georgia 30333

Doc. No. 6digit88
Version 06/07
Replaces Versions 06/04 and 05/07

```
                    Explanation of 6-Digit Code


   6th Digit Code - Master
      .000  Blank
      .001  Left Only
      .002  Right Only
      .003  Unilateral Unspecified
      .004  Bilateral
      .005
      .006
      .007
      .008  Possible, Probable, Borderline, or Rule Out;
            Defects only diagnosed prenatally should be coded with the last digit 8
            when the prenatal diagnosis is not definitive.
      .009  Not Otherwise Specified (NOS)



Notes:
      An asterisk (*) beside a disease code indicates that the code was created by
      CDC.

      A pound symbol (#) beside a disease code indicates that the condition or
      defect is listed on the MACDP Exclusion List.

      A check (T) beside a disease code indicates that an addition/revision was
      made since the last printing of the Procedure Manual. Use of the code should
      be according to the exclusion list criteria.

      The abbreviations NEC and NOS used in this code are defined as not elsewhere
      classified and as not otherwise specified, respectively.
```

R = Rev. 6/07
N = Rev. 5/07                          A - 33
T = Rev. 6/04
* = code created by CDC
# = on the MACDP Excl List

CONGENITAL ANOMALIES

**Anencephalus and Similar Anomalies**

**740.0  Anencephalus**

    740.000  Absence of brain
    740.010  Acrania
    740.020  Anencephaly
    740.030  Hemianencephaly, hemicephaly
    740.080  Other

**740.1  Craniorachischisis**

    740.100  Craniorachischisis

**740.2  Iniencephaly**

    740.200  Closed iniencephaly
    740.210  Open iniencephaly
    740.290  Unspecified iniencephaly


**741     Spina Bifida**
Includes:Spina bifida <u>aperta</u> (open lesions)
            myelocele
            rachischisis
Spina bifida <u>cystica</u> (closed lesions)
            meningocele
            meningomyelocele
            myelomeningocele
Excludes:Spina bifida occulta (see 756.100)
            craniorachischisis (see 740.100)

**741.0  Spina Bifida with Hydrocephalus**

    741.000  Spina bifida <u>aperta</u>, any site, with hydrocephalus

    741.010  Spina bifida <u>cystica</u>, any site, with hydrocephalus
             and Arnold-Chiari malformation
             Arnold-Chiari malformation, NOS
    741.020  Spina bifida <u>cystica</u>, any site, with stenosed
             aqueduct of Sylvius
    741.030  Spina bifida <u>cystica</u>, cervical, with unspecified
             hydrocephalus
             Spina bifida <u>cystica</u>, cervical, with
             hydrocephalus but without mention of
             Arnold-Chiari malformation or aqueduct stenosis
    741.040  Spina bifida <u>cystica</u>, thoracic, with unspecified
             hydrocephalus, no mention of Arnold-Chiari
    741.050  Spina bifida <u>cystica</u>, lumbar, with unspecified
             hydrocephalus, no mention of Arnold-Chiari
    741.060  Spina bifida <u>cystica</u>, sacral, with unspecified
             hydrocephalus, no mention of Arnold-Chiari
    741.070  Spina bifida of any site with hydrocephalus of
             late onset

R = Rev. 6/07
N = Rev. 5/07                          A - 34
T = Rev. 6/04
* = code created by CDC
# = on the MACDP Excl List

```
        741.080   Other spina bifida, meningocele of specified site
                  with hydrocephalus
        741.085   Spina bifida, meningocele, cervicothoracic, with
                  hydrocephalus
        741.086   Spina bifida, meningocele thoracolumbar, with
                  hydrocephalus
        741.087   Spina bifida, meningocele, lumbosacral with
                  hydrocephalus
        741.090   Spina bifida of any unspecified type
                  with hydrocephalus
```

## 741.9   Spina bifida without mention of hydrocephalus

```
        741.900   Spina bifida (aperta), without hydrocephalus
        741.910   Spina bifida (cystica), cervical, without hydrocephalus
        741.920   Spina bifida (cystica), thoracic, without hydrocephalus
        741.930   Spina bifida (cystica), lumbar, without hydrocephalus
        741.940   Spina bifida (cystica), sacral, without hydrocephalus
        741.980   Spina bifida, other specified site, without hydrocephalus
                  Includes: cervicothoracic, thoracolumbar, lumbosacral
        741.985   Lipomyelomeningocele
        741.990   Spina bifida, site unspecified, without hydrocephalus
                  (myelocoele, myelomeningocele, meningomyelocele)
```

## 742   Other Congenital Anomalies of Nervous System

## 742.0   Encephalocele

```
        742.000   Occipital encephalocele
        742.080   Other encephalocele of specified site
                  (includes midline defects)
        742.085   Frontal encephalocele
        742.086   Parietal encephalocele
        742.090   Unspecified encephalocele
```

## 742.1   Microcephalus
```
        742.100   Microcephalus
```

## 742.2   Reduction deformities of brain

```
        742.200   Anomalies of cerebrum
        742.210   Anomalies of corpus callosum
        742.220   Anomalies of hypothalamus
        742.230   Anomalies of cerebellum
        742.240   Agyria and lissencephaly
        742.250   Microgyria, polymicrogyria
        742.260   Holoprosencephaly
        742.270   Arrhinencephaly
        742.280   Other specified reduction defect of brain
        742.290   Unspecified reduction defect of brain
```

## 742.3   Congenital hydrocephalus
```
        Excludes:hydrocephalus with any condition in 741.9 (use 741.0)

        742.300   Anomalies of aqueduct of Sylvius
        742.310   Atresia of foramina of Magendie and Luschka
```

```
                    Dandy-Walker syndrome
        742.320   Hydranencephaly
        742.380   Other specified hydrocephaly
                    Includes:     communicating hydrocephaly
    #   742.385   Hydrocephalus secondary to intraventricular
                    hemorrhage (IVH) or CNS bleed
        742.390   Unspecified hydrocephaly, NOS
```

**742.4  Other specified anomalies of brain**

```
        742.400   Enlarged brain and/or head
                    megalencephaly
                    macrocephaly
        742.410   Porencephaly
                    Includes:     porencephalic cysts
        742.420   Cerebral cysts
        742.480   Other specified anomalies of brain
                    Includes:     cortical atrophy
                                  cranial nerve defects
        742.485   Ventricular cysts
                    Excludes:     arachnoid cysts
        742.486   Small brain
```

**742.5  Other specified anomalies of spinal cord**

```
        742.500   Amyelia
        742.510   Hypoplasia and dysplasia of spinal cord
                    atelomyelia
                    myelodysplasia
        742.520   Diastematomyelia
        742.530   Other cauda equina anomalies
        742.540   Hydromyelia
                    Hydrorachis
        742.580   Other specified anomalies of spinal cord and membranes
                    Includes:     congenital tethered cord
```

**742.8  Other specified anomalies of nervous system**
```
        Excludes:congenital oculofacial paralysis
                    Moebius syndrome (use 352.600)

        742.800   Jaw-winking syndrome
                    Marcus Gunn syndrome
        742.810   Familial dysautonomia
                    Riley-Day syndrome
        742.880   Other specified anomalies of nervous system
```

**742.9  Unspecified anomalies of brain, spinal cord and nervous systems**

```
        742.900   Brain, unspecified anomalies
        742.910   Spinal cord, unspecified anomalies
        742.990   Nervous system, unspecified anomalies
```

R = Rev. 6/07
N = Rev. 5/07                          A - 36
T = Rev. 6/04
* = code created by CDC
# = on the MACDP Excl List

**743 Congenital Anomalies of Eye**

    743.000  Anophthalmos
                agenesis of eye
                cryptophthalmos
    743.100  Microphthalmos, small eyes
                aplasia of eye
                hypoplasia of eye
                dysplasia of eye
                rudimentary eye

**743.2 Buphthalmos**

    743.200  Buphthalmos
                congenital glaucoma
                hydrophthalmos
    743.210  Enlarged eye, NOS
    743.220  Enlarged cornea
                keratoglobus
                congenital megalocornea

**743.3 Congenital cataract and lens anomalies**

    743.300  Absence of lens
                congenital aphakia
    743.310  Spherical lens
                Spherophakia
    743.320  Cataract, NOS
    743.325  Cataract, anterior polar
    743.326  Cataract, other specified
    743.330  Displaced lens
    743.340  Coloboma of lens
    743.380  Other specified lens anomalies
    743.390  Unspecified lens anomalies

**743.4 Coloboma and other anomalies of anterior segments**

    743.400  Corneal opacity
    743.410  Other corneal anomalies
                Excludes:    megalocornea (use 743.220)
    743.420  Absence of iris
                aniridia
    743.430  Coloboma of iris
    743.440  Other anomalies of iris
                polycoria
                ectopic pupil
                Peter's anomaly
      #         Excludes:    brushfield spots (use 743.800)
    743.450  Blue sclera
         #  If <36 weeks gestation, code only if another reportable
                defect is present.
                Always code if ≥36 weeks gestation.
    743.480  Other specified colobomas and anomalies of anterior segments
                Rieger's anomaly
    743.490  Unspecified colobomas and anomalies of anterior eye segments

**743.5  Congenital anomalies of posterior segment**

      743.500   Specified anomalies of vitreous humour
      743.510   Specified anomalies of retina
                congenital retinal aneurysm
                Excludes:    Stickler syndrome (use 759.860)
      743.520   Specified anomalies of optic disc
                hypoplastic optic nerve

                coloboma of the optic disc
      743.530   Specified anomalies of choroid
      743.535   Coloboma of choroid
      743.580   Other specified anomalies of posterior segment of eye
      743.590   Unspecified anomalies of posterior segment of eye

**743.6  Congenital anomalies of eyelids, lacrimal system, and orbit**

      743.600   Blepharoptosis
                congenital ptosis
      743.610   Ectropion
      743.620   Entropion
   #  743.630   Other anomalies of eyelids
                absence of eyelashes
                long eyelashes
                weakness of eyelids
T  #            fused eyelids (exclude if <25 weeks gestation unless another
                reportable defect is present)
      743.635   Blepharophimosis
                small or narrow palpebral fissures
      743.636   Coloboma of the eyelids
      743.640   Absence or agenesis of lacrimal apparatus
                absence of punctum lacrimale
   #  743.650   Stenosis or stricture of lacrimal duct
      743.660   Other anomalies of lacrimal apparatus (e.g., cyst)
      743.670   Anomalies of orbit

**743.8  Other specified anomalies of eye**

   #  743.800   Other specified anomalies of eye
                Includes:  exophthalmos
                           epicanthal folds
                           antimongoloid slant
                           upward eye slant
                           Brushfield spots
                Excludes:  congenital nystagmus (use 379.500)
                           retinitis pigmentosa (use 362.700)
                           ocular albinism (use 270.200)
                           wide spaced eyes, hypertelorism (use 756.085)

   *  743.810   Epibulbar dermoid cyst

**743.9  Unspecified anomalies of eye**

      743.900   Unspecified anomalies of eye
                congenital: of eye (any part)
                anomaly, NOS
                deformity, NOS

R = Rev. 6/07
N = Rev. 5/07                          A - 38
T = Rev. 6/04
* = code created by CDC
# = on the MACDP Excl List

**744  Congenital Anomalies of Ear, Face, and Neck**

**744.0  Anomalies of ear causing impairment of hearing**

    744.000  Absence or stricture of auditory canal
    744.010  Absence of auricle (pinna)
           absence of ear, NOS
    744.020  Anomaly of middle ear
           fusion of ossicles
    744.030  Anomaly of inner ear
           Includes:    congenital anomaly of membranous
                         labyrinth organ of Corti
    744.090  Unspecified anomalies of ear with hearing impairment
           Includes: congenital deafness, NOS

**744.1  Accessory auricle**

  # 744.100  Accessory auricle
          Polyotia
  # 744.110  Preauricular appendage, tag, or lobule
          (in front of ear canal)
  # 744.120  Other appendage, tag, or lobule include papillomas,
          ear tags

**744.2  Other specified anomalies of ear**

    744.200  Macrotia (enlarged pinna)
    744.210  Microtia (hypoplastic pinna and absence or
           stricture of external auditory meatus)
    744.220  Bat ear
T  # 744.230  Other misshapen ear
           pointed ear
           elfin
           pixie-like
           lop ear
           cauliflower ear
           cleft in ear
           malformed ear
           absent or decreased cartilage
    744.240  Misplaced ears
  # 744.245  Low set ears
  # 744.246  Posteriorly rotated ears
    744.250  Absence or anomaly of eustachian tube
    744.280  Other specified anomalies of ear (see also 744.230)
  #         Excludes:    Darwin's tubercle

**744.3  Unspecified anomalies of ear**

    744.300  Unspecified anomalies of ear
           Congenital:  ear (any part)
           anomaly, deformity, NOS

**744.4  Branchial cleft, cyst, or fistula; preauricular sinus**

    744.400  Branchial cleft, sinus, fistula cyst, or pit
  # 744.410  Preauricular sinus, cyst, or pit

```
        744.480   Other branchial cleft anomalies
                  Includes: dermal sinus of head
    #   744.500   Webbing of neck
                  Includes:    pterygium colli,
                               redundant neck skin folds
```

## 744.8 Other unspecified anomalies of face and neck

```
        744.800   Macrostomia (large mouth)
        744.810   Microstomia (small mouth)
    #   744.820   Macrocheilia (large lips)
    #   744.830   Microcheilia (small lips)
        744.880   Other specified anomalies of face/neck
```

## 744.9 Unspecified anomalies of face and neck

```
    #   744.900   Congenital anomaly of neck, NOS
                  Includes: short neck
        744.910   Congenital anomaly of face, NOS
                  Abnormal facies
```

**745      Bulbus Cordis Anomalies and Anomalies of Cardiac Septal Closure**

**745.0   Common truncus (see 747.200 for pseudotruncus)**

    745.000   Persistent truncus arteriosus
              absent septum between aorta and pulmonary
              artery
    745.010   Aortic septal defect
              Includes: aortopulmonary window
              Excludes: <u>atrial</u> septal defect (use 745.590)

**745.1   Transposition of great vessels**

    745.100   Transposition of great vessels, complete (no VSD)
    745.110   Transposition of great vessels, incomplete (w/ VSD)
              Taussig-Bing syndrome
    745.120   Corrected transposition of great vessels,
              L-transposition, ventri in version
              Excludes: dextrocardia (use 746.800)
N   745.130   Double outlet right ventricle (DORV) with normally
              related great vessels
N   745.140   Double outlet right ventricle (DORV) with transposed
              great vessels
N   745.150   Double outlet right ventricle (DORV), relationship of great
              vessels not specified
N   745.180   Other specified transposition of great vessels,
              no mention of double outlet right ventricle (DORV)
    745.190   Unspecified transposition of great vessels

**745.2   Tetralogy of Fallot**

    745.200   Fallot's tetralogy
    745.210   Fallot's pentalogy
              Fallot's tetralogy plus ASD

**745.3   Single ventricle**

    745.300   Single ventricle
              Common ventricle
              Cor triloculare biatriatum

**745.4   Ventricular septal defect**

N   745.400   Roger's disease
              Note:  This is an oudated term and the code is no longer
                     used. If this diagnostic term is encountered in
                     the medical record, code it as a ventricular septal
                     defect.
    745.410   Eisenmenger's syndrome
    745.420   Gerbode defect
T   745.480   Other specified ventricular septal defect
              Includes:  cystalline
                         sub-cystalline
                         subarterial
                         conoventricular
N   745.485   Perimembranous VSD
              Includes:  membranous VSD

```
N      745.486   Muscular VSD
                 Includes:  mid-muscular and apical VSDs
N      745.487   Inlet VSD
                 Includes:  common atrioventricular (AV) canal type VSD
                 Note:  Code common atrioventricular (AV) canal as
                        745.630
                        Code common atrioventricular (AV) canal with
                        muscular VSD as 745.620
       745.490   Ventricular septal defect, NOS
                 Excludes: common atrioventricular canal type (use
                 745.620)
       745.498   Probable VSD
```

## 745.5    Ostium secundum type atrial septal defect

```
N    #  745.500   Nonclosure of foramen ovale, NOS
                  Patent foramen ovale (PFO)
                  1)Always code if ≥36 weeks of gestation at birth and defect
                  last noted at ≥6 weeks of age.
                  2)If ≥36 weeks gestation at birth and defect last noted
                  <6 weeks of age, code only if another reportable heart
                  defect is present.
                  3)Never code if <36 weeks gestation at birth regardless
                  of presence of other defects.
        745.510   Ostium (septum) secundum defect
N       745.520   Lutembacher's syndrome
                  Note:  This is an outdated term and the code is no
                         longer used. If this diagnostic term is
                         encountered in the medical record, code the
                         individual components, not the syndrome.
        745.580   Other specified atrial septal defect
        745.590   ASD (atrial septal defect), NOS
                  Auricular septal defect, NOS
                  Partial foramen ovale
                  PFO vs. ASD
```

## 745.6  Endocardial cushion defects

```
        745.600   Ostium primum defects
        745.610   Single common atrium, cor triloculare biventriculare
N       745.620   Common atrioventricular canal with ventricular
                  septal defect (VSD)
                  Includes: Common AV canal with muscular VSD
                  Excludes: Inlet VSD or common AV canal type VSD (code as
                  745.487
        745.630   Common atrioventricular canal
        745.680   Other specified cushion defect
        745.690   Endocardial cushion defect, NOS
```

## 745.7  Cor biloculare

```
        745.700   Cor biloculare
```

## 745.8  Other specified defects of septal closure

```
        745.800   Other specified defects of septal closure
```

R = Rev. 6/07
N = Rev. 5/07
T = Rev. 6/04
* = code created by CDC
# = on the MACDP Excl List

**745.9  Unspecified defect of septal closure**

745.900   Unspecified defect of septal closure


**746     Other Congenital Anomalies of Heart**

**746.0  Anomalies of pulmonary valve**

N       746.000   Atresia, hypoplasia of pulmonary valve
                  Note:  Code pulmonary artery atresia as 747.300
                         Code pulmonary artery hypoplasia as 747.380
                         Code "pulmonic" or "pulmonary" atresia or
                         hypoplasia, NOS (no mention of valve or
                         artery) as 746.995
N       746.010   Stenosis of pulmonary valve
    #             Excludes:    pulmonary infundibular
                               stenosis (use 746.830)
                  Note:  Code pulmonary artery stenosis as 747.320
                         Code "pulmonic" or "pulmonary" stenosis, NOS (no
                         mention of valve or artery) as 746.995
N    #  746.020   Pulmonary valve insufficiency or regurgitation,
                  congenital
                  Never code cases designated as 'mild', minimal', 'trivial',
                  or 'physiologic'.
                  Code cases designated as 'moderate' or 'severe' and those
                  where the degree is not specified (NOS) only if another
                  reportable heart defect is present.
        746.080   Other specified anomalies of pulmonary valve
    #             Excludes:    pulmonary infundibular
                               stenosis (use 746.830)
        746.090   Unspecified anomaly of pulmonary valve


**746.1  Tricuspid atresia and stenosis**

N       746.100   Tricuspid atresia only
                  Excludes:  tricuspid stenosis and hypoplasia
N    #  746.105   Tricuspid valve insufficiency or regurgitation,
                  congenital
                   Never code cases designated as 'mild', minimal', 'trivial',
                  or 'physiologic'.
                  Code cases designated as 'moderate' or 'severe' and those
                  where the degree is not specified (NOS) only if another
                  reportable heart defect is present.
N       746.106   Tricuspid stenosis or hypoplasia

**746.2  Ebstein's anomaly**

746.200   Ebstein's anomaly

**746.3  Congenital stenosis of aortic valve**

746.300   Congenital stenosis of aortic valve
          Includes:    congenital aortic stenosis
                       subvalvular aortic stenosis
          Excludes:    supravalvular aortic stenosis (747.220)

R = Rev. 6/07
N = Rev. 5/07
T = Rev. 6/04
* = code created by CDC
# = on the MACDP Excl List

**746.4  Congenital insufficiency of aortic valve**

N  # 746.400   Aortic valve insufficiency or regurgitation, congenital
                Excludes:  bicuspid aortic valve.
                Never code cases designated as 'mild', minimal', 'trivial',
                or 'physiologic'.
                Code cases designated as 'moderate' or 'severe' and those
                where the degree is not specified (NOS) only if another
                reportable heart defect is present.
N    746.470   Bicuspid aortic valve
   * 746.480   Other specified anomalies of the aortic valves
                Includes:    aortic valve atresia
                Excludes:    supravalvular aortic stenosis (747.220)
   * 746.490   Unspecified anomalies of the aortic valves

**746.5  Congenital mitral stenosis**

     746.500   Congenital mitral stenosis
     746.505   Absence, atresia, or hypoplasia of mitral valve

**746.6  Mitral valve insufficiency or regurgitation, congenital**

N  # 746.600   Mitral valve insufficiency or regurgitation, congenital
                Never code cases designated as 'mild', minimal', 'trivial',
                or 'physiologic'.
                Code cases designated as 'moderate' or 'severe' and those
                where the degree is not specified (NOS) only if another
                reportable heart defect is present.

**746.7  Hypoplastic left heart syndrome**

     746.700   Hypoplastic left heart syndrome
                Atresia, or marked hypoplasia of the
                ascending aorta and defective development
                of left ventricle (with mitral valve atresia)

**746.8  Other specified anomalies of the heart**

     746.800   Dextrocardia without situs inversus (situs solitus)
                Dextrocardia with no mention of situs inversus
                Excludes:    dextrocardia with situs inversus use 759.300)
N    746.810   Levocardia
                Note:  This condition has been moved to the never code
                       list.
     746.820   Cor triatriatum
     746.830   Pulmonary infundibular (subvalvular) stenosis
     746.840   Trilogy of Fallot
     746.850   Anomalies of pericardium
N  # 746.860   Anomalies of myocardium
                cardiomegaly, congenital, NOS
                cardiomyopathy, congenital
                cardiomyopathy, hypertrophic
                Note: Do not code cardiomyopathy of any type in a
                newborn of a diabetic mother (either gestational or pre-
                existing diabetes).

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```
           746.870   Congenital heart block

           746.880   Other specified anomalies of heart
                        Includes:    ectopia (ectopic) cordis (mesocardia),
                                     conduction defects, NOS
           746.881   Hypoplastic left ventricle
                        Excludes:    hypoplastic left heart syndrome (746.700)
           746.882   Hypoplastic right heart (ventricle)
                        Uhl's disease
         * 746.883   Hypoplastic ventricle, NOS
           746.885   Anomalies of coronary artery or sinus
    N      746.886   Ventricular hypertrophy (right or left)
                        Note: Do not code ventricular hypertrophy of any type in
                        a newborn of a diabetic mother (either gestational or
                        pre-existing diabetes).
           746.887   Other defects of the atria
                        Excludes:    congenital Wolfe-Parkinson-White
                                     (use 426.705)
                                     rhythm anomalies (use 426.-, 427.-)


     746.9   Unspecified anomalies of heart

           746.900   Unspecified anomalies of heart valves
           746.910   Anomalous bands of heart
           746.920   Acyanotic congenital heart disease, NOS
           746.930   Cyanotic congenital heart disease, NOS
                        Blue baby
           746.990   Unspecified anomaly of heart:
                        Includes:  congenital heart disease (CHD)
    N      746.995   "Pulmonic" or "pulmonary" atresia, stenosis, or
                        hypoplasia, NOS (no mention of valve or artery)
                        Note:  Code pulmonary valve atresia or hypoplasia as
                               746.000
                               Code pulmonary valve stenosis as 746.010
                               Code pulmonary artery atresia as 747.300
                               Code pulmonary artery stenosis as 747.320
                               Code pulmonary artery hypoplasia as 747.380


     747 Other Congenital Anomalies of Circulatory System

    N   # 747.000   Patent ductus arteriosus (PDA)
                        Note: 1)Always code if ≥36 weeks of gestation at birth and
                        defect last noted at ≥6 weeks of age.
                        2)If ≥36 weeks gestation at birth and defect last noted
                        <6 weeks of age, code only if the PDA was treated (e.g.
                        by ligation or indomethicin) or if another reportable
                        heart defect is present.
                        3)Never code if <36 weeks gestation at birth or if
                        treated with prostaglandins regardless of gestational
                        age. (See PDA Tree Appendix)
           747.008   Probable PDA


     747.1   Coarctation of aorta

           747.100   Preductal (proximal) coarctation of aorta
           747.110   Postductal (distal) coarctation of aorta
           747.190   Unspecified coarctation of aorta
```

**747.2  Other anomalies of aorta**

|   |   |   |
|---|---|---|
|   | 747.200 | Atresia of aorta |
|   |   | absence of aorta |
|   |   | pseudotruncus arteriosus |
|   | 747.210 | Hypoplasia of aorta |
|   |   | tubular hypoplasia of aorta |
| N | 747.215 | Interrupted aortic arch, Type A |
| N | 747.216 | Interrupted aortic arch, Type B |
| N | 747.217 | Interrupted aortic arch, Type C |
|   | 747.220 | Supra-aortic stenosis (supravalvular) |
|   |   | Excludes: aortic stenosis, |
|   |   |          congenital (see 746.300) |
|   | 747.230 | Persistent right aortic arch |
|   | 747.240 | Aneurysm of sinus of Valsalva |
|   | 747.250 | Vascular ring (aorta) |
|   |   | double aortic arch |
|   |   | Includes:    vascular ring compression of trachea |
|   | 747.260 | Overriding aorta |
|   |   | dextroposition of aorta |
|   | 747.270 | Congenital aneurysm of aorta |
|   |   | congenital dilatation of aorta |
|   | 747.280 | Other specified anomalies of aorta |
| N | 747.285 | Interrupted aortic arch, NOS, type not specified |
|   | 747.290 | Unspecified anomalies of aorta |

**747.3  Anomalies of pulmonary artery**

| N | 747.300 | Pulmonary artery atresia, absence or agenesis |
|---|---|---|
|   |   | Note:  Code pulmonary valve atresia as 746.000 |
|   |   |        Code "pulmonic" or "pulmonary" atresia, NOS (no mention of valve or artery) as 746.995 |
|   | 747.310 | Pulmonary artery atresia with septal defect |
| N | 747.320 | Pulmonary artery stenosis |
|   |   | Includes: Stenosis of the main pulmonary artery or of the right or left main branches |
|   |   | Note:  Code pulmonary valve stenosis as 746.010 |
|   |   |        Code "pulmonic" or "pulmonary" stenosis, NOS (no mention of valve or artery) as 746.995 |
| N | 747.325 | Peripheral pulmonary artery stenosis |
|   |   | Includes: Stenosis of a pulmonary artery peripheral to the main right or left main branches |
|   |   |           Peripheral pulmonic stenosis (PPS), NOS, documented by echocardiogram |
| # |   | Excludes: Peripheral pulmonic stenosis (PPS) murmur only (not documented by echocardiogram) |
|   |   | Note: 1)Always code if ≥36 weeks of gestation at birth and defect last noted at ≥6 weeks of age. |
|   |   | 2)If ≥36 weeks gestation at birth and defect last noted <6 weeks of age, code only if another reportable heart defect is present. |
|   |   | 3)Never code if <36 weeks gestation at birth. |
|   |   | (See PPS Tree Appendix) |
|   | 747.330 | Aneurysm of pulmonary artery |
|   |   | dilatation of pulmonary artery |
|   | 747.340 | Pulmonary arteriovenous malformation or aneurysm |

```
747.380   Other specified anomaly of pulmonary artery
          Includes:    pulmonary artery hypoplasia
          Note:  Code pulmonary valve hypoplasia as 746.000
                 Code "pulmonic" or "pulmonary" hypoplasia, NOS
                 (no mention of valve or artery) as 746.995
747.390   Unspecified anomaly of pulmonary artery
```

**747.4   Anomalies of great veins**

```
747.400   Stenosis of vena cava (inferior or superior)
747.410   Persistent left superior vena cava
747.420   (TAPVR) Total anomalous pulmonary venous return
747.430   Partial anomalous pulmonary venous return
747.440   Anomalous portal vein termination
747.450   Portal vein - hepatic artery fistula
747.480   Other specified anomalies of great veins
747.490   Unspecified anomalies of great veins
```

**747.5   Absence or hypoplasia of umbilical artery**

```
 #  747.500   Single umbilical artery
```

**747.6   Other anomalies of peripheral vascular system**

```
747.600   Stenosis of renal artery
747.610   Other anomalies of renal artery
747.620   Arteriovenous malformation (peripheral)
          Excludes: pulmonary (747.340)
                    cerebral (747.800)
                    retinal (743.510)
747.630   Congenital phlebectasia
          congenital varix
747.640   Other anomalies of peripheral arteries
          Includes:    aberrant subclavian artery
747.650   Other anomalies of peripheral veins
          Excludes:    Budd-Chiari - occlusion of hepatic vein (use
                       453.000)
```
N    `747.680   Other anomalies of peripheral vascular system`
#    `          Includes: primary pulmonary artery hypertension ONLY if`
    `          it is present in an infant at >7 days of age`
```
747.690   Unspecified anomalies of peripheral vascular system
```

**747.8   Other specified anomalies of circulatory system**

```
747.800   Arteriovenous (malformation) aneurysm of brain
747.810   Other anomalies of cerebral vessels
          Includes:    vein of Galen
747.880   Other specified anomalies of circulatory system
          Excludes:    congenital aneurysm:
                  coronary (746.880)
                  peripheral (747.640)
                  pulmonary (747.330)
                  retinal (743.510)
                  ruptured cerebral arteriovenous
                  aneurysm (430.000)
```

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ruptured cerebral aneurysm (430.000)

**747.9  Unspecified anomalies of circulatory system**

747.900   Unspecified anomalies of circulatory system

**748     Congenital Anomalies of Respiratory System**

**748.0  Choanal atresia**

    748.000   Choanal atresia
          atresia of nares, anterior or posterior
          congenital stenosis

**748.1  Other anomalies of nose**

    748.100   Agenesis or underdevelopment of nose
    748.110   Accessory nose
    748.120   Fissured, notched, or cleft nose
    748.130   Sinus wall anomalies
    748.140   Perforated nasal septum
  # 748.180   Other specified anomalies of nose
          flat bridge of nose
          wide nasal bridge
          small nose and nostril
          absent nasal septum
    748.185   Tubular nose, single nostril, proboscis
    748.190   Unspecified anomalies of nose
          Excludes:   congenital deviation of the nasal
                       septum (use 754.020)

**748.2  Web of larynx**

    748.205   Web of larynx-glottic
    748.206   Web of larynx-subglottic
    748.209   Web of larynx-NOS

**748.3  Other anomalies of larynx, trachea, and bronchus**

    748.300   Anomalies of larynx and supporting cartilage
T   748.310   Congenital subglottic stenosis – Never code if chart states
          the condition was acquired or secondary to endotracheal (ET)
          intubation or ventilation
    748.330   Other anomalies of trachea
#          Excludes: vascular ring compression of the
                 trachea (use 747.250)
    748.340   Stenosis of bronchus
    748.350   Other anomalies of bronchus
    748.360   Congenital laryngeal stridor, NOS
    748.380   Other specified anomalies of larynx and bronchus
    748.385   Cleft larynx, laryngotracheoesophageal cleft
    748.390   Unspecified anomalies of larynx, trachea, and bronchus

**748.4  Congenital cystic lung**

    748.400   Single cyst, lung or lung cyst
    748.410   Multiple cysts, lung
          Polycystic lung
    748.420   Honeycomb lung
    748.480   Other specified congenital cystic lung

**748.5  Agenesis or aplasia of lung**

      748.500   Agenesis or aplasia of lung
T     748.510   Hypoplasia of lung; Pulmonary hypoplasia
   #            Exclude if isolated defect in infants <36 weeks gestation.
      748.520   Sequestration of lung
      748.580   Other specified dysplasia of lung
                Fusion of lobes of lung
   * 748.590   Unspecified dysplasia of lung

**748.6  Other anomalies of lung**

      748.600   Ectopic tissues in lung
      748.610   Bronchiectasis
      748.620   Accessory lobe of lung
      748.625   Bilobar right lung or right lung with left lung bronchial pattern
      748.690   Other and unspecified anomalies of lung

**748.8  Other specified anomalies of respiratory system**

      748.800   Anomaly of pleura
      748.810   Congenital cyst of mediastinum
      748.880   Other specified respiratory system anomalies
                Includes:    congenital lobar emphysema
                           lymphangiectasia of lungs

**748.9  Unspecified anomalies of respiratory system**

      748.900   Unspecified anomalies of respiratory system
                Absence of respiratory organ, NOS
                Anomaly of respiratory system, NOS

**749    Cleft Palate and Cleft Lip**

**749.0  Cleft palate alone**
       (If description of condition includes Pierre Robin sequence,
        use additional code, 524.080)

       749.000   Cleft hard palate, unilateral
       749.010   Cleft hard palate, bilateral
       749.020   Cleft hard palate, central
       749.030   Cleft hard palate, NOS
       749.040   Cleft soft palate, alone unilateral
       749.050   Cleft soft palate, alone bilateral
       749.060   Cleft soft palate, alone central
       749.070   Cleft soft palate, alone, NOS
       749.080   Cleft uvula
       749.090   Cleft palate, NOS
               palatoschisis

**749.1  Cleft lip alone**
       Includes:alveolar ridge cleft
               cleft gum
               harelip
       749.100   Cleft lip, unilateral
       749.110   Cleft lip, bilateral
       749.120   Cleft lip, central
       749.190   Cleft lip, NOS (fused lip)
               cleft gum

**749.2  Cleft lip with cleft palate**

       749.200   Cleft lip, unilateral, with any cleft palate
       749.210   Cleft lip, bilateral, with any cleft palate
       749.220   Cleft lip, central, with any cleft palate
       749.290   Cleft lip, NOS, with any cleft palate

**750    Other Congenital Anomalies of Upper Alimentary Tract**

  #  750.000   Tongue tie
                 Ankyloglossia

**750.1  Other anomalies of tongue**
       Excludes:protruding tongue (never a defect)

       750.100   Aglossia
                 Absence of tongue
       750.110   Hypoglossia (small tongue)
                 Microglossia
       750.120   Macroglossia (large tongue)
       750.130   Dislocation or displacement of tongue
                 Glossoptosis
       750.140   Cleft tongue or split tongue
       750.180   Other specified anomalies of tongue
       750.190   Unspecified anomalies of tongue

**750.2  Other specified anomalies of mouth and pharynx**

       750.200   Pharyngeal pouch
       750.210   Other pharyngeal anomalies
       750.230   Other anomalies of salivary glands or ducts
    #  750.240   High arched palate
       750.250   Other anomalies of palate
       750.260   Lip fistulae or pits
       750.270   Other lip anomalies
                 Includes:    notched lip, prominent philtrum,
                              long philtrum
                 Excludes:    cleft lip (see 749)
       750.280   Other specified anomalies of mouth and pharynx
                 Excludes:    receding jaw (see 524.0)
                              large and small mouth (see 744.8)

**750.3  Tracheoesophageal (T-E) fistula, esophageal atresia and stenosis**

       750.300   Esophageal atresia without mention of T-E fistula
       750.310   Esophageal atresia with mention of T-E fistula
       750.320   Tracheoesophageal fistula without mention of esophageal atresia
       750.325   Tracheoesophageal fistula – "H" type
       750.330   Bronchoesophageal fistula with or without mention of esophageal
                 atresia
       750.340   Stenosis or stricture of esophagus
       750.350   Esophageal web
       750.380   Other tracheoesophageal anomalies

**750.4  Other specified anomalies of esophagus**

       750.400   Congenital dilatation of esophagus
                 giant esophagus
       750.410   Displacement of esophagus
       750.420   Diverticulum of esophagus
                 esophageal pouch
       750.430   Duplication of esophagus
       750.480   Other specified anomalies of esophagus

**750.5 Congenital hypertrophic pyloric stenosis**

    # 750.500   Pylorospasm
      750.510   Congenital hypertrophic pyloric stenosis
      750.580   Other congenital pyloric obstruction

**750.6 Congenital hiatus hernia**

      750.600   Congenital hiatus hernia
                 Cardia displacement through esophageal hiatus
                 Partial thoracic stomach
                 Excludes:    congenital diaphragmatic hernia (756.610)

**750.7 Other specified anomalies of stomach**

      750.700   Microgastria
      750.710   Megalogastria
      750.720   Cardiospasm
                 achalasia of cardia, congenital
      750.730   Displacement or transposition of stomach
      750.740   Diverticulum of stomach
      750.750   Duplication of stomach
      750.780   Other specified anomalies of stomach

**750.8 Other specified anomalies of upper alimentary tract**

      750.800   Other specified anomalies of upper alimentary tract

**750.9 Unspecified anomalies of upper alimentary tract**

      750.900   Unspecified anomalies of mouth and pharynx
      750.910   Unspecified anomalies of esophagus
      750.920   Unspecified anomalies of stomach
      750.990   Unspecified anomalies of upper alimentary tract

**751    Other Congenital Anomalies of Digestive System**

**751.0  Meckel's diverticulum**

    751.000  Persistent omphalomesenteric duct
                persistent vitelline duct
  # 751.010  Meckel's diverticulum

**751.1  Atresia and stenosis of small intestine**

    751.100  Stenosis, atresia or absence of duodenum
    751.110  Stenosis, atresia or absence of jejunum
    751.120  Stenosis, atresia or absence of ileum
    751.190  Stenosis, atresia or absence of small intestine
    751.195  Stenosis, atresia or absence of small intestine with fistula

**751.2  Atresia and stenosis of large intestine, rectum and anal canal**

    751.200  Stenosis, atresia or absence of large intestine
                Stenosis, atresia or absence of appendix
    751.210  Stenosis, atresia or absence of rectum <u>with</u> fistula
    751.220  Stenosis, atresia or absence of rectum <u>without</u> mention of fistula
    751.230  Stenosis, atresia or absence of anus with fistula
                Includes:    imperforate anus with fistula
    751.240  Stenosis, atresia or absence of anus without mention of fistula
                Includes:    imperforate anus without fistula

**751.3  Hirschsprung's disease and other congenital functional disorders of the colon**

    751.300  Total intestinal aganglionosis
    751.310  Long-segment Hirschsprung's disease; aganglionosis beyond the rectum
    751.320  Short-segment Hirschsprung's disease; aganglionosis involving no more than the anal sphincter and the rectum
    751.330  Hirschsprung's disease, NOS
    751.340  Congenital megacolon
                congenital macrocolon, not aganglionic

**751.4  Anomalies of intestinal fixation**

    751.400  Malrotation of cecum and/or colon
    751.410  Anomalies of mesentery
    751.420  Congenital adhesions or bands of omentum and peritoneum; Ladd's bands
    751.490  Other specified and unspecified malrotation
    751.495  Malrotation of small intestine alone

**751.5  Other anomalies of intestine**

    751.500  Duplication of anus, appendix, cecum, or intestine
                enterogenous cyst
    751.510  Transposition of appendix, colon, or intestine
    751.520  Microcolon

```
              751.530  Ectopic (displaced) anus
              751.540  Congenital anal fistula
              751.550  Persistent cloaca
   R          751.555  Exstrophy of cloaca
                       Excludes exstrophy of urinary bladder not associated with
                       imperforate anus (use 753.500)
        *     751.560  Duodenal web
        #     751.580  Other specified anomalies of intestine
                       Includes: rectal fissures
              751.590  Unspecified anomalies of intestine
```

**751.6  Anomalies of gallbladder, bile ducts, and liver**

```
              751.600  Absence or agenesis of liver, total or partial
              751.610  Cystic or fibrocystic disease of liver
        #     751.620  Other anomalies of liver
                       hepatomegaly
                       hepatosplenomegaly (also use code 759.020)
                       Excludes:   Budd-Chiari (use 453.000)
              751.630  Agenesis or hypoplasia of gallbladder
              751.640  Other anomalies of gallbladder
                       duplication of gallbladder
              751.650  Agenesis or atresia of hepatic or bile ducts
                       Includes:    biliary atresia
                       Excludes:    congenital or neonatal hepatitis
                                      (use 774.480 or 774.490)
              751.660  Choledochal cysts
              751.670  Other anomalies of hepatic or bile ducts
              751.680  Anomalies of biliary tract, NEC
```

**751.7  Anomalies of pancreas**
```
        Excludes:fibrocystic disease of pancreas (277.000)
                       diabetes mellitus,
                       congenital
                       neonatal

              751.700  Absence, agenesis or hypoplasia of pancreas
              751.710  Accessory pancreas
              751.720  Annular pancreas
              751.730  Ectopic pancreas
              751.740  Pancreatic cyst
              751.780  Other specified anomalies of pancreas
              751.790  Unspecified anomalies of pancreas
```

**751.8  Other specified anomalies of digestive system**

```
              751.800  Absence of alimentary tract, NOS
                       (complete or partial)
              751.810  Duplication of alimentary tract
              751.820  Ectopic digestive organs, NOS
              751.880  Other specified anomalies of digestive system
```

**751.9  Unspecified anomalies of digestive system**

      751.900   Unspecified anomalies of digestive system
                congenital of digestive system, NOS
                anomaly, NOS
                deformity, NOS
                obstruction, NOS

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**752    Congenital Anomalies of Genital Organs**
        Excludes:congenital hydrocele (778.600)
                 testicular feminization syndrome (257.800)
                 syndromes associated with anomalies in
                 number and form of chromosomes (758)

**752.0  Anomalies of ovaries**

        752.000   Absence or agenesis of ovaries
        752.010   Streak ovary
        752.020   Accessory ovary
        752.080   Other specified anomalies of ovaries
        752.085   Multiple ovarian cysts
        752.090   Unspecified anomalies of ovaries

**752.1  Anomalies of fallopian tubes and broad ligaments**

        752.100   Absence of fallopian tube or broad ligament
        752.110   Cyst of mesenteric remnant
                  epoophoron cyst
                  cyst of Gartner's duct
        752.120   Fimbrial cyst
                  parovarian cyst
        752.190   Other and unspecified anomalies of fallopian tube
                  and broad ligaments

**752.2  Doubling of uterus**

        752.200   Doubling of uterus
                  doubling of uterus (any degree) or
                  associated with doubling of cervix and
                  vagina

**752.3  Other anomalies of uterus**

        752.300   Absence or agenesis of uterus
        752.310   Displaced uterus
        752.320   Fistulae involving uterus with digestive or
                  urinary tract
                  Includes:     uterointestinal fistula
                                uterovesical fistula
        752.380   Other anomalies of uterus
                  bicornuate uterus
                  unicornis uterus
        752.390   Unspecified anomalies of uterus

**752.4  Anomalies of cervix, vagina, and external female genitalia**

        752.400   Absence, atresia or agenesis of cervix
        752.410   Absence or atresia of vagina, complete or partial
        752.420   Congenital rectovaginal fistula
     #  752.430   Imperforate hymen
     #  752.440   Absence or other anomaly of vulva
                  fusion of vulva
                  hypoplastic labia majora – Always code if ≥36weeks gestation. If
                  <36 weeks gestation, code only if another reportable defect is
                  present.

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```
  # 752.450  Absence or other anomaly of clitoris
                 Includes: clitoromegaly
                            enlarged clitoris
                            clitoral hypertrophy
                            prominent clitoris
  # 752.460  Embryonal cyst of vagina
    752.470  Other cyst of vagina, vulva, or canal of Nuck
  # 752.480  Other specified anomalies of cervix, vagina, or external female
                 genitalia
                 Includes:    vaginal tags
                              hymenal tags
    752.490  Unspecified anomalies of cervix, vagina, or external female
                 genitalia
```

## 752.5  Undescended testicle

```
  #          1)If < 36 weeks gestation, code only if there is a
                 medical/surgical intervention for this problem;
                 2)If ≥36 weeks gestation and defect last noted at <1
                 year of age, code only if there was a medical/surgical
                 intervention for this problem or if another reportable
                 defect is present
                 3)Always code if ≥36 weeks gestation and defect first
                 noted at ≥1 of age.

  # 752.500  Undescended testicle, unilateral
                 undescended, unpalpable
  # 752.501  Left undescended testicle
  # 752.502  Right undescended testicle
  # 752.514  Undescended testicle, bilateral
  # 752.520  Undescended testicle, NOS (Cryptorchidism)
    752.530  Ectopic testis, unilateral and bilateral
```

## 752.6  Hypospadias and epispadias

```
    752.600  Hypospadias (alone), NOS
    752.605  1°, glandular,coronal
    752.606  2°, penile
    752.607  3°, perineal, scrotal
    752.610  Epispadias
    752.620  Congenital chordee (with hypospadias), NOS
    752.621  Congenital chordee alone (chordee w/o hypospadias)
    752.625  Cong. chordee with 1°, coronal hypospadias
    752.626  Cong. chordee with 2°, penile hypospadias
    752.627  Cong. chordee with 3°, perineal, scrotal hypospadias
```

## 752.7  Indeterminate sex and pseudohermaphroditism
```
    Excludes:pseudohermaphroditism:
             female, with adrenocortical disorder (see 255.200)
             male, with gonadal disorder with specified chromosomal anomaly
             (see 758)

    752.700  True hermaphroditism
                 ovotestis
```

```
752.710  Pseudohermaphroditism, male
752.720  Pseudohermaphroditism, female
         pure gonadal dysgenesis
         Excludes:    gonadal agenesis (758.690)
752.730  Pseudohermaphrodite, NOS
752.790  Indeterminate sex, NOS
         ambiguous genitalia
```

**752.8   Other specified anomalies of male genital organs**

```
  752.800  Absence of testis
           monorchidism, NOS
# 752.810  Aplasia or hypoplasia of testis and scrotum
  752.820  Other anomalies of testis and scrotum
           polyorchidism
           bifid scrotum
           Excludes:    torsion of the testes or spermatic
                        cord (use #608.200)
  752.830  Atresia of vas deferens
  752.840  Other anomalies of vas deferens and prostate
  752.850  Absence or aplasia of penis
# 752.860  Other anomalies of penis
           absent or hooded foreskin
#          redundant foreskin (never a defect)
  752.865  Small penis, hypoplastic penis, or micropenis
  752.870  Cysts of embryonic remnants
           cyst: hydatid of Morgagni
                 Wolffian duct
                 appendix testis
  752.880  Other specified anomalies of genital organs
           microgenitalia
           macrogenitalia
```

**752.9   Unspecified anomalies of genital organs**

```
  752.900  Unspecified anomalies of genital organs
           Congenital: of genital organ, NEC
           anomaly, NOS or deformity, NOS
```

**753    Congenital Anomalies of Urinary System**

**753.0  Renal agenesis and dysgenesis**

    753.000   Bilateral absence, agenesis, dysplasia, or
                 hypoplasia of kidneys
                 Potter's syndrome
    753.009   Renal agenesis, NOS
    753.010   Unilateral absence, agenesis, dysplasia or
                 hypoplasia of kidneys

**753.1  Cystic kidney disease**

    753.100   Renal cyst (single)
    753.110   Polycystic kidneys, infantile type
    753.120   Polycystic kidneys, adult type
    753.130   Polycystic kidneys, NOS
    753.140   Medullary cystic disease, juvenile type
    753.150   Medullary cystic disease, adult type
                 Medullary sponge kidney
    753.160   Multicystic renal dysplasia
                  Multicystic kidney
    753.180   Other specified cystic disease
                  Includes: cystic kidneys, NOS

**753.2  Obstructive defects of renal pelvis and ureter**

    753.200   Congenital hydronephrosis
    753.210   Atresia, stricture, or stenosis of ureter
                 Includes:    uretero<u>pelvic</u> junction obstruction/stenosis
                            uretero<u>vesical</u> junction obstruction/stenosis
                            hypoplastic ureter
    753.220   Megaloureter, NOS
                 Includes:    hydroureter
    753.290   Other and unspecified obstructive defects of renal
                 pelvis and ureter

**753.3  Other specified anomalies of kidney**

    753.300   Accessory kidney
    753.310   Double or triple kidney and pelvis
                 pyelon duplex or triplex
    753.320   Lobulated, fused, or horseshoe kidney
    753.330   Ectopic kidney
    753.340   Enlarged, hyperplastic or giant kidney
    753.350   Congenital renal calculi
    753.380   Other specified anomalies of kidney

**753.4  Other specified anomalies of ureter**

    753.400   Absence of ureter
    753.410   Accessory ureter
                 double ureter, duplex collecting system
    753.420   Ectopic ureter
    753.480   Other specified anomalies of ureter
                  Includes:    ureterocele

R = Rev. 6/07
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* = code created by CDC
# = on the MACDP Excl List

753.485   Variations of vesicoureteral reflux


**753.5   Exstrophy of urinary bladder**

        753.500   Exstrophy of urinary bladder
                  ectopia vesicae
                  extroversion of bladder

**753.6   Atresia and stenosis of urethra and bladder neck**

        753.600   Congenital posterior urethral valves or posterior urethral
                  obstruction
        753.610   Other atresia, or stenosis of bladder neck
        753.620   Obstruction, atresia or stenosis of anterior urethra
        753.630   Obstruction, atresia or stenosis of urinary meatus
                  Includes:    meatal stenosis
        753.690   Other and unspecified atresia and stenosis of urethra and
                  bladder neck

**753.7   Anomalies of urachus**

  T  #   753.700   Patent urachus
        753.710   Cyst of urachus
        753.790   Other and unspecified anomaly of urachus

**753.8   Other specified anomalies of bladder and urethra**

        753.800   Absence of bladder or urethra
        753.810   Ectopic bladder
        753.820   Congenital diverticulum or hernia of bladder
        753.830   Congenital prolapse of bladder (mucosa)
        753.840   Double urethra or urinary meatus
        753.850   Ectopic urethra or urethral orifice
        753.860   Congenital digestive-urinary tract fistulae
                  rectovesical fistula
        753.870   Urethral fistula, NOS
        753.880   Other specified anomalies of bladder and urethra

**753.9   Unspecified anomalies of urinary system**

        753.900   Unspecified anomaly of kidney
        753.910   Unspecified anomaly of ureter
        753.920   Unspecified anomaly of bladder
        753.930   Unspecified anomaly of urethra
        753.990   Unspecified anomaly of urinary system, NOS

R = Rev. 6/07
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* = code created by CDC
# = on the MACDP Excl List

```
754        Certain Congenital Musculoskeletal Anomalies

754.0  Of skull, face, and jaw
       Excludes:dentofacial anomalies (524.0)
                Pierre Robin sequence (524.080)
                syphilitic saddle nose (090.000)

       754.000  Asymmetry of face
       754.010  Compression (Potter's) facies
     # 754.020  Congenital deviation of nasal septum
                bent nose
 T       754.030  Dolichocephaly
                Always code if ≥36 weeks gestation
     #          If <36 weeks gestation, code only if another reportable
                defect is present
     # 754.040  Depressions in skull
                Includes:    large fontanelle
                             small fontanelle
       754.050  Plagiocephaly
       754.055  Asymmetric head
 T   #   * 754.060  Scaphocephaly, no mention of craniosynostosis
         * 754.070  Trigonocephaly, no mention of craniosynostosis
                Always code if ≥36 weeks gestation
     #          If <36 weeks gestation, code only if another reportable
                defect is present
     * 754.080  Other specified skull deformity, no mention of
                craniosynostosis
                Includes:brachycephaly
                         acrocephaly
                         turricephaly
                         oxycephaly
     * 754.090  Deformity of skull, NOS

754.1  Anomalies of sternocleidomastoid muscle

       754.100  Anomalies of sternocleidomastoid muscle
     *          Includes: absent or hypoplastic sternocleidomastoid
                          contracture of sternocleidomastoid muscle
                          sternomastoid tumor
                Excludes: congenital sternocleidomastoid torticollis
                         (use 756.860)

754.2  Certain congenital musculoskeletal deformities of spine

       754.200  Congenital postural scoliosis
       754.210  Congenital postural lordosis
       754.220  Congenital postural curvature of spine, NOS

754.3  Congenital dislocation of hip

       754.300  Congenital dislocation of hip
       754.310  Unstable hip
                preluxation of hip
                subluxation of hip
                predislocation status of hip at birth
```

R = Rev. 6/07
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**754.4  Congenital genu recurvatum and bowing of long bones of leg**

     754.400   Bowing, femur
     754.410   Bowing, tibia and/or fibula
     754.420   Bow legs, NOS
     754.430   Genu recurvatum
     754.440   Dislocation of knee, congenital
     754.490   Deformity of leg, NOS

**754.5  Varus (inward) deformities of feet**

     754.500   Talipes equinovarus
     754.510   Talipes calcaneovarus
  # 754.520   Metatarsus varus or metatarsus adductus
     754.530   Complex varus deformities
     754.590   Unspecified varus deformities of feet

**754.6  Valgus (outward) deformities of feet**

     754.600   Talipes calcaneovalgus
     754.610   Congenital pes planus
     754.615   Pes valgus
     754.680   Other specified valgus deformities of foot
     754.690   Unspecified valgus deformities of foot

**754.7  Other deformities of feet**

     754.700   Pes cavus
                Claw foot (use 755.350 for claw foot)
     754.720   Short Achilles tendon
     754.730   Clubfoot, NOS
                talipes, NOS
     754.735   Congenital deformities of foot, NOS
     754.780   Other specified deformities of ankle and/or toes
                Includes:   dorsiflexion of foot
                Excludes:   widely spaced 1st and 2nd toes (use 755.600)

**754.8  Other specified congenital musculoskeletal deformities**

     754.800   Pigeon chest (pectus carinatum)
     754.810   Funnel chest (pectus excavatum)
     754.820   Other anomalies of chest wall
                Includes: deformed chest, barrel chest
     754.825   Shield chest
     754.830   Dislocation of elbow
     754.840   Club hand or fingers
     754.850   Spade-like hand
     754.880   Other specified deformity of hands
                (see 755.500 for specified anomalies of fingers)

**755     Other Congenital Anomalies of Limbs**

**755.0  Polydactyly**

      755.005   Accessory fingers (postaxial polydactyly, Type A)
  # 755.006   Skin tag (postaxial polydactyly, Type B)
              Exclude: Do not code in black infants.
      755.007   Unspecified finger or skin tag (postaxial polydactyly, NOS)
      755.010   Accessory thumbs (preaxial polydactyly)
      755.020   Accessory toes (postaxial)
      755.030   Accessory big toe (preaxial)
      755.090   Accessory digits, NOS (hand/foot not specified)
      755.095   Accessory digits hand, NOS (preaxial, postaxial not
              specified)
      755.096   Accessory digits foot, NOS (preaxial, postaxial not
              specified)

**755.1  Syndactyly**

      755.100   Fused fingers
      755.110   Webbed fingers
      755.120   Fused toes
T  # 755.130   Webbed toes
              Code webbing of the second and third toes only if another
              reportable defect is present. Always code webbing of other toes
              regardless of whether another reportable defect is present
      755.190   Unspecified syndactyly (see below for specified site)
      755.191   Unspecified syndactyly thumb and/or fingers, unilateral
      755.192   Unspecified syndactyly thumb and/or fingers, bilateral
      755.193   Unspecified (webbed vs. fused) syndactyly thumb and/or fingers,
              NOS
      755.194   Unspecified syndactyly toes unilateral
      755.195   Unspecified syndactyly toes bilateral
      755.196   Unspecified syndactyly toes, NOS
      755.199   Unspecified syndactyly (i.e., webbed vs. fused) digits not
              known

**755.2  Reduction defects of upper limb**

T      If description of the condition includes amniotic or constricting
      bands use additional code, 658.800 (Only use 658.800 if another
      reportable defect is present)
      Excludes shortening of upper limb (use 755.580) or hypoplasia of
      upper limb (use 755.585)

      755.200   Absence of upper limb
              Absent:  humerus (total or partial), radius,ulna and hand
              Includes: amelia of upper limb, NOS
                       infants with rudimentary or nubbin fingers
                       attached to stump of humerus or shoulder girdle
      755.210   Absence of upper arm and forearm
              Absent:  humerus (total or partial), radius and ulna
                 (total or partial)
              Present:  hand (total or partial)
              Includes: phocomelia of upper limb, NOS;
                     intercalary reduction defect of upper limb, NOS

R = Rev. 6/07
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* = code created by CDC
# = on the MACDP Excl List

```
755.220  Absence of forearm only or upper arm only
         Absent:   radius and ulna
         Present:  humerus, hand (total or partial)
                   or
         Absent:   humerus
         Present:  radius, ulna, and hand
755.230  Absence of forearm and hand
         Absent:   radius and ulna (total or partial) and hand
         Includes: infants with rudimentary or nubbin fingers
                   attached to stump of forearm or elbow
755.240  Absence of hand or fingers
         Absent:   hand or fingers (total or partial) not in
                   conjunction with ray or long bone reduction
         Includes: rudimentary or nubbin fingers;
                   absent individual phalanges;
                   absent or missing fingers, NOS
         Excludes: isolated absent or hypoplastic thumb
                   (use 755.260)
755.250  Split-hand malformation
         Absent:   central fingers (third with or without second,
                   fourth) and metacarpals (total or partial)
         Includes: monodactyly;
                   lobster-claw hand
         Excludes: isolated absent central fingers without
                   metacarpal defects (use 755.240)
755.260  Preaxial longitudinal reduction defect of upper limb
         Absent:   radius (total or partial) and/or thumb with or
                   without second finger (total or partial)
         Includes: isolated absent or hypoplastic thumb;
                   radial ray defect, NOS
755.265  Longitudinal reduction defect of upper limb, NOS
         Includes: absent forearm long bone with absent fingers,
                   NOS
755.270  Postaxial longitudinal reduction defect of upper limb
         Includes: isolated absent ulna (total or partial);
                   absent fifth with or without fourth finger
                   (total or partial) only if ulna or fifth ±
                   fourth metacarpal also totally or partially
                   absent;
                   ulnar ray defect, NOS
755.280  Other specified reduction defect of upper limb
755.285  Transverse reduction defect of upper limb, NOS
         Includes: congenital amputation of upper limb, NOS
755.290  Unspecified reduction defect of upper limb
```

**755.3  Reduction defects of lower limb**

T     If description of condition includes amniotic or constricting bands
      use additional code, 658.800 (Only use this code if another
      reportable defect is present)

      Excludes shortening of lower limb (use 755.680) and hypoplasia of
      lower limb (use 755.685)

R = Rev. 6/07
N = Rev. 5/07                                    A - 65
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* = code created by CDC
# = on the MACDP Excl List

```
755.300  Absence of lower limb
         Absent:   femur (total or partial), tibia, fibula, and
                   foot
         Includes: amelia of lower limb, NOS
                   infants with rudimentary or nubbin toes
                   attached to stump of femur or pelvic girdle
755.310  Absence of thigh and lower leg
         Absent:   femur (total or partial), tibia and fibula
                   (total or partial)
         Present:  foot (total or partial)
         Includes: phocomelia of lower limb, NOS;
                   intercalary reduction defect of lower limb, NOS
755.320  Absence of lower leg only or femur only
         Absent:   tibia and fibula
         Present:  femur, foot (total or partial)
                   or
         Absent:   femur
         Present:  tibia, fibula, and foot
755.330  Absence of lower leg and foot
         Absent:   tibia and fibula (total or partial), foot
         Includes: infants with rudimentary or nubbin toes attached
                   to stump of leg or knee
755.340  Absence of foot or toes
         Absent:   foot or toes (total or partial) not in
                   conjunction with ray or long bone reduction
         Includes: rudimentary or nubbin toes;
                   absent individual phalanges;
                   absent or missing toes, NOS
         Excludes: isolated absent or hypoplastic great toe
                   (use 755.365)
755.350  Split-foot malformation
         Absent:   central toes (third with or without second,
                   fourth) and metatarsals (total or partial)
         Includes: monodactyly;
                   lobster claw foot
         Excludes: isolated absent central toes without metatarsal
                   defects (use 755.340)
         Note: preaxial lower limb reductions can occur with
               split-hand malformations of the upper limb and these
               lower limb defects should be coded 755.365
755.360  Longitudinal reduction defect of lower limb, NOS
         Includes: absent long bone of leg with absent toes, NOS
755.365  Preaxial longitudinal reduction defect of lower limb
         Absent:   tibia (total or partial) and/OR great toe with
                   or without second toe (total or partial)
         Includes: isolated absent or hypoplastic great toe;
                   tibial ray defect, NOS
755.366  Postaxial longitudinal reduction defect of lower limb
         Includes: isolated absent fibula (total or partial);
                   absent fifth with or without fourth toe (total
                   or partial) only if fibula or fifth ± fourth
                   metatarsal also totally or partially absent;
                   fibular ray defect, NOS
755.380  Other specified reduction defect of lower limb
755.385  Transverse reduction defect of lower limb, NOS
             Includes: congenital amputation of lower limb, NOS
755.390  Unspecified reduction defect of lower limb
```

**755.4  Reduction defects of unspecified limb**

T    If description of condition includes amniotic or constricting bands
     use additional code, 658.800 (note: 658.00 should only be used with
     another reportable defect)

    755.400  Absence of limb, NOS
             Includes: amelia, NOS
    755.410  Phocomelia, NOS
             Includes: intercalary reduction defect, NOS
    755.420  Transverse reduction defect, NOS
             Includes: congenital amputation of unspecified limb
    755.430  Longitudinal reduction defect, NOS
             Includes: preaxial or postaxial reduction defect, NOS
    755.440  Absent digits, not specified whether fingers or toes
    755.480  Other specified reduction defect of unspecified limb
    755.490  Unspecified reduction defect of unspecified limb

**755.5  Other anomalies of upper limb, including shoulder girdle**
     Includes:complex anomalies involving all or part of upper limb

  #  755.500  Anomalies of fingers
             Includes:     camptodactyly
                           clinodactyly
                           macrodactylia
                           brachydactyly
                           triphalangeal thumb
                           incurving fingers
             Excludes:     acrocephalosyndactyly (see 756.050)
                           Apert's syndrome (see 756.055)
    755.510  Anomalies of hand
             Excludes:   simian crease (use 757.200)
    755.520  Anomalies of wrist
    755.525  Accessory carpal bones
    755.526  Madelung's deformity
    755.530  Anomalies of forearm, NOS
    755.535  Radioulnar dysostosis
    755.536  Radioulnar synostosis
    755.540  Anomalies of elbow and upper arm
    755.550  Anomalies of shoulder
    755.555  Cleidocranial dysostosis
    755.556  Sprengel's deformity
    755.560  Other anomalies of whole arm
    755.580  Other specified anomalies of upper limb
             Includes:     hyperextensibility of upper limb
                           shortening of arm
    755.585  Hypoplasia of upper limb
             Includes:     hypoplasia of fingers, hands, or arms
             Excludes:     aplasia or absent upper limb (see 755.2)
    755.590  Unspecified anomalies of upper limb

R = Rev. 6/07                      A - 67
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* = code created by CDC
# = on the MACDP Excl List

**755.6  Other anomalies of lower limb, including pelvic girdle**
        Includes:complex anomalies involving all
                or part of lower limb

    #  755.600   Anomalies of toes
                  Includes:     overlapping toes
                                hammer toes
                                widely spaced first and second toes
       755.605   Hallux valgus
       755.606   Hallux varus
       755.610   Anomalies of foot
                  Includes:    plantar furrow
                  Excludes:    lobster claw foot (use 755.350)
    #  755.616   Rocker-bottom foot
       755.620   Anomalies of ankle
                  astragaloscaphoid synostosis
    #  755.630   Anomalies of lower leg
                  angulation of tibia, tibial torsion
                  (exclude if clubfoot present)
       755.640   Anomalies of knee
                  hyperextended knee
       755.645   Genu valgum
       755.646   Genu varum
       755.647   Absent patella or rudimentary patella
       755.650   Anomalies of upper leg
                  anteversion of femur
       755.660   Anomalies of hip
                  Includes:     coxa vara
                                coxa valga
                                other abnormalities of hips
       755.665   Hip dysplasia, NOS
       755.666   Unilateral hip dysplasia
       755.667   Bilateral hip dysplasia
       755.670   Anomalies of pelvis
                  fusion of sacroiliac joint
       755.680   Other specified anomalies of lower limb
                  hyperextended legs
                  shortening of legs
       755.685   Hypoplasia of lower limb
                  Includes:    hypoplasia of toes, feet, legs
                  Excludes:    aplasia or absent lower limb (see 755.3)
       755.690   Unspecified anomalies of legs


**755.8  Other specified anomalies of unspecified limb**

       755.800   Arthrogryposis multiplex congenita
                  Includes:    distal arthrogryposis syndrome
       755.810   Larsen's syndrome
       755.880   Other specified anomalies of unspecified limb
                  Includes:    overlapping digits, NOS
                               hyperextended joints, NOS
                  Excludes:    hyperextended knees (use 755.640)


**755.9  Unspecified anomalies of unspecified limb**

755.900  Unspecified anomalies of unspecified limb

**756     Other Congenital Musculoskeletal Anomalies**

**756.0  Anomalies of skull and face bones**
         Excludes:skull and face deformities in 754
                  Pierre Robin sequence (use 524.080)

         756.000  Craniosynostosis, NOS
                  craniostenosis, NOS
                  closed-skull sutures, NOS
         756.005  Sagittal craniosynostosis
         756.006  Metopic craniosynostosis
         756.010  Coronal craniosynostosis
         756.020  Lambdoidal craniosynostosis
         756.030  Other types of craniosynostosis
                  Includes:    basilar craniosynostosis
         756.040  Craniofacial dysostosis
                  Includes:    Crouzon's disease
         756.045  Mandibulofacial dysostosis
                  Includes:    Franceschetti syndrome
                               Treacher-Collins syndrome
         756.046  Other craniofacial syndromes
                  Includes:    oculomandibulofacial syndrome
                               Hallermann-Streiff syndrome
         756.050  Acrocephalosyndactyly, NOS
         756.055  Acrocephalosyndactyly types I or II
                  Apert syndrome
         756.056  Acrocephalosyndactyly type III
         756.057  Other specified acrocephalosyndactylies
         756.060  Goldenhar syndrome
                  oculoauriculovertebral dysplasia
         756.065  Hemifacial microsomia
         756.080  Other specified skull and face bone anomalies
                  Includes:    localized skull defects
      #                        flat occiput
                               mid-facial hypoplasia
      #                        prominent occiput
                               prominent maxilla
                               hypotelorism
                  Excludes:    macrocephaly (use 742.400)
                               small chin (see 524.0)
                               Pierre Robin sequence (use 524.080)
         756.085  Hypertelorism, telecanthus, wide set eyes
         756.090  Unspecified skull and face bone anomalies
                  Excludes:    dentofacial anomalies (524.0)
                               skull defects associated with brain anomalies
                               such as:
                               anencephalus  (740.0)
                               encephalocele (742.0)
                               hydrocephalus (742.3)
                               microcephalus (742.100)

**756.1  Anomalies of spine**

         756.100  Spina bifida occulta
         756.110  Klippel-Feil syndrome
                  Wildervanck syndrome

R = Rev. 6/07                      A - 70
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* = code created by CDC
# = on the MACDP Excl List

```
         756.120  Kyphosis
                  kyphoscoliosis
         756.130  Congenital spondylolisthesis
         756.140  Anomalies of cervical vertebrae
         756.145  Hemivertebrae (cervical)
         756.146  Agenesis (cervical)
         756.150  Anomalies of thoracic vertebrae
         756.155  Hemivertebrae of thoracic vertebrae
         756.156  Agenesis of thoracic vertebrae
         756.160  Anomalies of lumbar vertebrae
         756.165  Hemivertebrae of lumbar vertebrae
         756.166  Agenesis of lumbar vertebrae
         756.170  Sacrococcygeal anomalies
                  Includes: agenesis of sacrum
                  Excludes: pilonidal sinus (see 685.100)
         756.179  Sacral mass, NOS
         756.180  Other specified vertebral anomalies
         756.185  Hemivertebrae, NOS
         756.190  Unspecified anomalies of spine
```

**756.2  Cervical rib**

```
      #  756.200  Cervical rib
                  supernumerary rib in cervical region
```

**756.3  Other anomalies of ribs and sternum**

```
         756.300  Absence of ribs
         756.310  Misshapen ribs
         756.320  Fused ribs
         756.330  Extra ribs
         756.340  Other anomalies of ribs
         756.350  Absence of sternum
         756.360  Misshapen sternum
         756.380  Other anomalies of sternum
                  Includes:    double ossification center in the manubrium,
                               bifid sternum, short sternum
         756.390  Anomalies of thoracic cage, unspecified
                  Excludes:    deformed chest (use 754.820)
```

**756.4  Chondrodystrophy**

```
         756.400  Asphyxiating thoracic dystrophy
                  Jeune syndrome
                  thoracic-pelvic-phalangeal dysplasia
                  Excludes: homozygous achondroplasia
         756.410  Chondrodysplasia
                  Ollier syndrome, enchondromatosis
         756.420  Chondrodysplasia with hemangioma
                  Kast syndrome
                  Maffucci syndrome
         756.430  Achondroplastic dwarfism
         756.440  Other specified chondrodystrophies
                  Excludes:    Conradi's (use 756.575)
         756.445  Diastrophic dwarfism
         756.446  Metatrophic dwarfism
         756.447  Thanatophoric dwarfism
```

R = Rev. 6/07                                    A - 71
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# = on the MACDP Excl List

```
756.450  Metaphyseal dysostosis
756.460  Spondyloepiphyseal dysplasia
756.470  Exostosis
         Excludes:    Gardner syndrome (see 759.630)
756.480  Other specified chondrodystrophy
756.490  Unspecified chondrodystrophy
         Excludes:    lipochondrodystrophy (use 277.510)
```

**756.5  Osteodystrophies**

```
756.500  Osteogenesis imperfecta
756.505  Osteopsathyrosis
756.506  Fragilitas ossium
756.510  Polyostotic fibrous dysplasia
         Albright-McCune-Sternberg syndrome
756.520  Chondroectodermal dysplasia
756.525  Ellis-van Creveld syndrome
756.530  Infantile cortical hyperostosis
         Caffey syndrome
756.540  Osteopetrosis
         Albers-Schonberg syndrome
         marble bones
756.550  Progressive diaphyseal dysplasia
         Engelmann syndrome
         Camurati-Engelmann disease
756.560  Osteopoikilosis
756.570  Multiple epiphyseal dysplasia
756.575  Conradi syndrome
         chondrodysplasia punctata
         Excludes:    warfarin embryopathy
756.580  Other specified osteodystrophies
756.590  Unspecified osteodystrophies
```

**756.6  Anomalies of diaphragm**

```
756.600  Absence of diaphragm
756.610  Congenital diaphragmatic hernia
756.615  Diaphragmatic hernia (Bochdalek)
756.616  Diaphragmatic hernia (Morgagni)
756.617  Hemidiaphragm
756.620  Eventration of diaphragm
756.680  Other specified anomalies of diaphragm
756.690  Unspecified anomalies of diaphragm
```

**756.7  Anomalies of abdominal wall**

```
756.700  Exomphalos, omphalocele
756.710  Gastroschisis
         Excludes:    umbilical hernia (553.100)
756.720  Prune belly syndrome
756.790  Other and unspecified anomalies of abdominal wall
756.795  Epigastric hernia
```

**756.8  Other specified anomalies of muscle, tendon, fascia and connective tissue**

```
756.800  Poland syndrome or anomaly
756.810  Other absent or hypoplastic muscle
```

R = Rev. 6/07
N = Rev. 5/07                              A - 72
T = Rev. 6/04
* = code created by CDC
# = on the MACDP Excl List

Includes:    absent pectoralis major
                    Excludes:    prune belly syndrome (use 756.720)
          756.820  Absent tendon
          756.830  Nail-patella syndrome
          756.840  Amyotrophia congenita
          756.850  Ehlers-Danlos syndrome
          756.860  Congenital torticollis
                   (see also 754.100, anomalies of sternocleidomastoid muscle)
          756.880  Other specified anomalies of muscle, tendon, fascia and
                   connective tissue
                    Includes:    myopathy, congenital NOS

## 756.9  Unspecified anomalies of musculoskeletal system

          756.900  Unspecified anomalies of muscle
          756.910  Unspecified anomalies of tendon
          756.920  Unspecified anomalies of bone
          756.930  Unspecified anomalies of cartilage
          756.940  Unspecified anomalies of connective tissue
          756.990  Unspecified anomalies of musculoskeletal system

R = Rev. 6/07
N = Rev. 5/07                              A - 73
T = Rev. 6/04
* = code created by CDC
# = on the MACDP Excl List

**757    Congenital Anomalies of the Integument**

757.000   Hereditary edema of legs
          Hereditary trophedema
          Milroy's disease

**757.1  Ichthyosis congenita**

757.100   Harlequin fetus
757.110   Collodion baby
757.115   Bullous type
757.120   Sjogren-Larsson syndrome
757.190   Other and unspecified
757.195   Ichthyosis vulgaris
757.196   X-linked ichthyosis
757.197   Ichthyosiform erythroderma

**757.2  Dermatoglyphic anomalies**

#  757.200   Abnormal palmar creases
             Includes:    simian creases, transverse palmar creases

**757.3  Other specified anomalies of skin**
     Excludes:pigmented mole (216.900)
             hemangioma (see 228.0)

757.300   Specified syndromes, not elsewhere classified, involving skin
          anomalies
#  757.310   Skin tags
             Includes:    anal tags
             Excludes:    preauricular tag (see 744.110)
                          vaginal tags (see 752.480)
757.320   Urticaria pigmentosa
757.330   Epidermolysis bullosa
757.340   Ectodermal dysplasia
             Excludes:    Ellis-van Creveld syndrome (756.525)
757.345   X-linked type ectodermal dysplasia
757.346   Other specified ectodermal dysplasias
757.350   Incontinentia pigmenti
757.360   Xeroderma pigmentosum
757.370   Cutis laxa hyperelastica
#  757.380   Nevus, not elsewhere classifiable
             Includes:    port wine stain or nevus flammeus
T            Excludes:    hairy nevus (use 216.920)
                           Sturge-Weber syndrome (use 759.610)
#  757.385   Birthmark, NOS
#  757.386   Mongolian blue spot
#  757.390   Other specified anomalies of skin
             Includes:  cafe au lait spots
                        hyperpigmented areas
                        skin cysts
                        hypoplastic dermal patterns
757.395   Absence of skin

**757.4 Specified anomalies of hair**

Excludes:kinky hair syndrome (use 759.870)

757.400    Congenital alopecia
        Excludes:    ectodermal dysplasia (use 757.340)
757.410    Beaded hair
        Monilethrix
757.420    Twisted hair
        Pili torti
757.430    Taenzer's hair
\# 757.450    Persistent or excessive lanugo
        Includes:    hirsutism
757.480    Other specified anomalies of hair

**757.5 Specified anomalies of nails**

757.500    Congenital anonychia
        Absent nails
757.510    Enlarged or hypertrophic nails
757.515    Onychauxis
757.516    Pachyonychia
757.520    Congenital koilonychia
757.530    Congenital leukonychia
757.540    Club nail
757.580    Other specified anomalies of nails
757.585    Hypoplastic (small) fingernails and/or toenails

**757.6 Specified anomalies of breast**

757.600    Absent breast with absent nipple
757.610    Hypoplastic breast with hypoplastic nipple
757.620    Accessory (ectopic) breast with nipple
757.630    Absent nipple
T   # 757.640    Small nipple (hypoplastic)
        Always code if ≥36 weeks gestation
   #        If <36 weeks gestation, code only if another reportable defect
        is present
\# 757.650    Accessory (ectopic) nipple, supernumerary
\# 757.680    Other specified anomalies of breast
        Widely spaced nipples
        Excludes:    inverted nipples (never a defect)

**757.8 Other specified anomalies of the integument**

757.800    Includes:    scalp defects
        For specified anomalies of skin see 757.390
        For specified anomalies of hair see 757.480
        For specified anomalies of nails see 757.580

**757.9 Unspecified anomalies of the integument**

757.900    Unspecified anomalies of skin
757.910    Unspecified anomalies of hair, NOS
757.920    Unspecified anomalies of nail, NOS
757.990    Unspecified anomalies of the integument, NOS

### 758    Chromosomal Anomalies

### 758.0  Down syndrome
Clinical Down syndrome karyotype identified as:

|   |   |   |
|---|---|---|
| T | 758.000 | Down syndrome, karyotype trisomy 21, cytogenetics result in record |
| T | 758.008 | Down syndrome suspected, cytogenetics pending |
|   | 758.010 | Down syndrome, karyotype trisomy G, NOS |
|   | 758.020 | Translocation trisomy - duplication of a 21 |
|   | 758.030 | Translocation trisomy - duplication of a G, NOS |
|   | 758.040 | Mosaic Down syndrome |
| T | 758.090 | Down syndrome, NOS (i.e. chart states a diagnosis of Trisomy 21 or Downs syndrome, but no cytogenetics result in record) |
| T | 758.098 | Down syndrome suspected, cytogenetics never done |

### 758.1  Patau syndrome
Clinical Patau syndrome karyotype identified as:

|   |   |   |
|---|---|---|
| T | 758.100 | Patau syndrome, karyotype trisomy 13, cytogenetics result in record |
|   | 758.108 | Patau syndrome suspected, cytogenetics pending |
|   | 758.110 | Patau syndrome, karyotype trisomy D, NOS |
|   | 758.120 | Translocation trisomy - duplication of a 13 |
|   | 758.130 | Translocation trisomy - duplication of a D, NOS |
| T | 758.190 | Patau syndrome, NOS (i.e. chart states a diagnosis of Trisomy 13 or Patau syndrome, but no cytogenetics result in record) |
| T | 758.198 | Patau syndrome suspected, cytogenetics pending |

### 758.2  Edwards syndrome
Clinical Edwards syndrome karyotype identified as:

|   |   |   |
|---|---|---|
| T | 758.200 | Edwards syndrome, karyotype trisomy 18, cytogenetics result in record |
| T | 758.208 | Edwards syndrome suspected, cytogenetics pending |
|   | 758.210 | Edwards syndrome, karyotype trisomy E, NOS |
|   | 758.220 | Translocation trisomy - duplication of an 18 |
|   | 758.230 | Translocation trisomy - duplication of an E, NOS |
|   | 758.290 | Edwards syndrome, NOS (i.e. chart states a diagnosis of |
| T |  | Trisomy 18 or Edwards syndrome, but no cytogenetics result in record) |
|   | 758.295 | Edwards phenotype - normal karyotype |
| T | 758.298 | Edwards syndrome suspected, cytogenetics pending |

**758.3  Autosomal deletion syndromes**

    758.300  Antimongolism syndrome
             Clinical antimongolism syndrome:
               karyotype - partial or total deletion of:
                   21
                   G, NOS
               NOS
    758.310  Cri du chat syndrome
             Clinical Cri du chat syndrome:
               karyotype - deletion of:
                   5
                   B, NOS
               NOS
    758.320  Wolff-Hirschorn syndrome
             Clinical Wolff-Hirschorn syndrome:
               karyotype - deletion of:
                   4
                   B, NOS
               NOS
    758.330  Deletion of long arm of 13
             deletion of long arm of D, NOS
    758.340  Deletion of long arm of E
             deletion of long arm of 17 or 18
    758.350  Deletion of short arm of E
             deletion of short arm of 17 or 18
    758.360  Monosomy G mosaicism
    758.370  Deletion in band 11 of long arm of 22 (22q11 deletions)
             Note: Code added for use with births on or after 4/1/2001
    758.380  Other loss of autosomal material
    758.390  Unspecified autosomal deletion syndromes

**758.4  Balanced autosomal translocation in normal individual**

    758.400  Balanced autosomal translocation in normal individual

**758.5  Other conditions due to autosomal anomalies**

    758.500  Trisomy 8
    758.510  Other trisomy C syndromes
             Trisomy: 6, 7, 9, 10, 11, 12, or C, NOS
    758.520  Other total trisomy syndromes
             Trisomy 22
             Trisomy, NOS
    758.530  Partial trisomy syndromes
    758.540  Other translocations
             Excludes:    balanced translocation in normal
                           individual (use 758.400)
    758.580  Other specified anomalies of autosomes, NOS
             Includes:    marker autosome
    758.585  Polyploidy
    758.586  Triploidy
    758.590  Unspecified anomalies of autosomes

**758.6  Gonadal Dysgenesis**
        Excludes:pure gonadal dysgenesis (752.720)
                 Noonan syndrome (759.800)

        758.600   Turner's phenotype, karyotype 45, X [XO]
        758.610   Turner's phenotype, variant karyotypes
                  karyotype characterized by:
                  isochromosome
                  mosaic, including XO
                  partial X deletion
                  ring chromosome
                  Excludes:    Turner's phenotype, karyotype normal XX
                                (use 759.800, Noonan syndrome)
        758.690   Turner syndrome, karyotype unspecified, NOS
                  Bonneville-Ullrich syndrome, NOS


**758.7  Klinefelter syndrome**

        758.700   Klinefelter's phenotype, karyotype 47, XXY
        758.710   Klinefelter's phenotype, other karyotype with additional
                  X chromosomes
                  XX
                  XXXY
                  XXYY
                  XXXXY
        758.790   Klinefelter syndrome, NOS


**758.8  Other conditions due to sex chromosome anomalies**

        758.800   Mosaic XO/XY, 45X/46XY
                  Excludes: with Turner's phenotype (758.610)
        758.810   Mosaic XO/XX
                  Excludes: with Turner's phenotype (758.610)
        758.820   Mosaic XY/XXY,46XY/47XXY
                  Excludes:    Klinefelter's phenotype (758.710)
        758.830   Mosaic including XXXXY,49XXXXY
                  Excludes:    with Klinefelter's phenotype (use 758.710)
        758.840   XYY, male, 47XYY
                  mosaic XYY male
        758.850   XXX female,47XXX
        758.860   Additional sex chromosomes, NOS
        758.880   Other specified sex chromosome anomaly
                  Includes:  fragile X
        758.890   Unspecified sex chromosome anomaly


**758.9  Conditions due to anomaly of unspecified chromosomes**

        758.900   Mosaicism, NOS
        758.910   Additional chromosome(s), NOS
        758.920   Deletion of chromosome(s), NOS
        758.930   Duplication of chromosome(s), NOS
        758.990   Unspecified anomaly of chromosome(s)

R = Rev. 6/07                          A - 78
N = Rev. 5/07
T = Rev. 6/04
* = code created by CDC
# = on the MACDP Excl List

**759    Other and Unspecified Congenital Anomalies**

**759.0  Anomalies of spleen**

```
    759.000  Absence of spleen
             asplenia
    759.005  Ivemark syndrome
    759.010  Hypoplasia of spleen
  # 759.020  Hyperplasia of spleen
             splenomegaly
             hepatosplenomegaly (also use code 751.620)
    759.030  Misshapen spleen
    759.040  Accessory spleen
    759.050  Ectopic spleen
    759.080  Other specified anomalies of spleen
    759.090  Unspecified anomalies of spleen
```

**759.1  Anomalies of adrenal gland**

```
    759.100  Absence of adrenal gland
    759.110  Hypoplasia of adrenal gland
    759.120  Accessory adrenal gland
    759.130  Ectopic adrenal gland
    759.180  Other specified anomaly of adrenal gland
             Excludes:   congenital adrenal hyperplasia
                              (use 255.200)
    759.190  Unspecified anomalies of adrenal gland
```

**759.2  Anomalies of other endocrine glands**

```
    759.200  Anomalies of pituitary gland
    759.210  Anomalies of thyroid gland
    759.220  Thyroglossal duct anomalies
             thyroglossal cyst
    759.230  Anomalies of parathyroid gland
  # 759.240  Anomalies of thymus
             thymic hypertrophy
             absent thymus
    759.280  Other specified anomalies of endocrine gland
    759.290  Unspecified anomaly of endocrine gland
```

**759.3  Situs inversus**

```
    759.300  Dextrocardia with complete situs inversus
    759.310  Situs inversus with levocardia
    759.320  Situs inversus thoracis
    759.330  Situs inversus abdominis
    759.340  Kartagener syndrome (triad)
    759.390  Unspecified situs inversus
             Excludes: dextrocardia (746.800) not
                         associated with complete situs inversus
```

**759.4  Conjoined twins**

```
    759.400  Dicephalus
             two heads
    759.410  Craniopagus
```

```
                head-joined twins
      759.420   Thoracopagus
                thorax-joined twins
      759.430   Xiphopagus
                xiphoid- and pelvis-joined twins
      759.440   Pygopagus
                buttock-joined twins
      759.480   Other specified conjoined twins
      759.490   Unspecified conjoined twins
```

**759.5  Tuberous sclerosis**

```
      759.500   Tuberous sclerosis
                Bourneville's disease
                epiloia
```

**759.6  Other hamartoses, not elsewhere classified**

```
      759.600   Peutz-Jeghers syndrome
      759.610   Encephalocutaneous angiomatosis
                Kalischer's disease
                Sturge-Weber syndrome
      759.620   Von Hippel-Lindau syndrome
      759.630   Gardner syndrome
      759.680   Other specified hamartomas
      759.690   Unspecified hamartomas
```

**759.7  Multiple congenital anomalies,**

```
      759.700   Multiple congenital anomalies,
                anomaly, multiple, NOS
                deformity, multiple, NOS
```

**759.8  Other specified anomalies and syndromes**

```
      759.800   Cong malformation syndromes affecting facial appearance
                cyclops
                Noonan syndrome
                oral-facial-digital (OFD) syndrome, type I
                Orofaciodigital syndrome, type II (Mohr syndrome)
                Waardenburg syndrome
                whistling face syndrome
      759.820   Cong malformation syndromes associated with short stature
                Amsterdam dwarf (Cornelia de Lange syndrome)
                Cockayne syndrome
                Laurence-Moon-Biedl syndrome
                Russell-Silver syndrome
                Seckel syndrome
                Smith-Lemli-Opitz syndrome
      759.840   Cong malformation syndromes involving limbs
                Carpenter syndrome
                Holt-Oram syndrome
                Klippel-Trenaunay-Weber syndrome
                Rubinstein-Taybi syndrome
                sirenomelia
                thrombocytopenia-absent radius (TAR) syndrome
      759.860   Cong malformation syndromes with other skeletal changes
```

```
                    Marfan syndrome
                    Stickler syndrome
          759.870   Cong malformation syndromes with metabolic disturbances
                    Alport syndrome
                    Beckwith (Wiedemann-Beckwith) syndrome
                    leprechaunism
                    Menkes syndrome (kinky hair syndrome)
                    Prader-Willi syndrome
                    Zellweger syndrome
          759.890   Other specified anomalies
                    Includes:    hemihypertrophy
                                 Meckel-Gruber syndrome
```

**759.9  Congenital anomaly, unspecified**

```
      #  759.900   Anomalies of umbilicus
                    low-lying umbilicus
                    umbilical cord atrophy
         759.910   Embryopathia, NEC
         759.990   Congenital anomaly, NOS
```

# Other Specified Codes Used in Metro Atlanta Congenital Defects Program

List ordered alphabetically

|       |       | 524.000 | Abnormalities of jaw size |
|-------|-------|---------|---------------------------|

```
          524.000  Abnormalities of jaw size
                   micrognathia
                   macrognathia  T
          255.200  Adrenogenital syndrome
      #   270.200  Albinism
      #   277.620  Alpha-1 antitrypsin deficiency
  T   #   658.800  Amniotic bands (constricting bands, amniotic cyst)
      #   270.600  Arginosuccinic aciduria
      #   778.000  Ascites, congenital

      216        Benign neoplasm of skin
        T  (NOTE: All neoplasms should be coded ONLY if another reportable code
           is present)
                   Includes: blue nevus       pigmented nevus
                             papilloma         dermatofibroma
                             syringoadenoma    hydrocystoma
        *                  * dermoid cyst      syringoma
                   Excludes: skin of female genital organs (use 221.000),
                             skin of male genital organs (use 222.000)
  T   #   216.200  Benign neoplasm of skin, ear and external auditory canal
                   Includes:   auricle ear
                               external meatus
                               auricular canal
                               external canal
                               pinna
                   Excludes:   cartilage of ear
  T   #   216.100  Benign neoplasm of skin, eyelid, including canthus
                   Excludes:   cartilage of eyelid
  T   #   216.000  Benign neoplasm of skin, lip
                   Excludes:   vermillion border of lip
  T   #   216.700  Benign neoplasm of skin, lower limb, hip
  T   #   216.300  Benign neoplasm of skin, other and unspecified parts of face
                   Includes:    cheek, external nose, external eyebrow
                                temple
  T   #   216.800  Benign neoplasm of skin, other specified sites of skin
                   Excludes:   epibulbar dermoid cyst (use 743.810)
  T   #   216.400  Benign neoplasm of skin, scalp and skin of neck
  T   #   216.900  Benign neoplasm of skin, site unspecified
      #   216.500  Benign neoplasm of skin, trunk, except scrotum
                   Includes:    axillary fold
                                perianal skin
                                skin of:    chest  wall,  abdominal  wall,  groin,
                                            buttock, anus, perineum, back, umbilicus,
                                            breast
                   Excludes:    anal canal
                                anus, NOS
                                skin of scrotum
```

R = Rev. 6/07
N = Rev. 5/07                          A - 82
T = Rev. 6/04
* = code created by CDC
# = on the MACDP Excl List

# Continued: Other Specified Codes Used in Metro Atlanta Congenital Defects Program

```
T    #  216.600   Benign neoplasm of skin, upper limb, shoulder
     #  221.000   Benign skin neoplasm of female genital organs
     #  222.000   Benign skin neoplasm of male genital organs
        453.000   Budd-Chiari, occlusion of hepatic vein
        427.900   Cardiac arrhythmias, NEC. Never code premature atrial
                  contractions, PACs.
     #  330.100   Cerebral lipidoses
                  Includes:   Tay-Sachs disease, gangliosidosis
        363.200   Chorioretinitis
        279.200   Combined immunodeficiency syndrome
        771.280   Congenital infection, other specified
                  Excludes:    human immunodeficiency virus (HIV)
                               infection and acquired immunodeficiency
                               syndrome (AIDS)
     #  277.000   Cystic fibrosis
                  No mention of meconium ileus
     #  277.010   Cystic fibrosis
                  With mention of meconium ileus
        228.100   Cystic hygroma
                  Lymphangioma, any site
        771.100   Cytomegalovirus (CMV) (in utero infections only)
        253.820   Diencephalic syndrome
        279.110   DiGeorge syndrome
        277.400   Disorders of bilirubin excretion
        425.300   Endocardial fibroelastosis
        553.200   Epigastric hernia
     #  767.600   Erb's palsy
     #  368.000   Esotropia
     #  378.000   Exotropia
     #  351.000   Facial palsy
        331.890   Familial degenerative CNS disease
        760.710   Fetal alcohol syndrome
        760.718   Fetal alcohol syndrome, probable
                  Includes:  "facies"
        760.750   Fetal hydantoin (Dilantin) syndrome
     #  282.200   Glucose-6-phosphate dehydrogenase (G-6-PD) deficiency
     #  271.000   Glycogen storage diseases
T       216.920   Hairy nevus
T   228.0  Hemangioma
                  Include if greater than 4-inches diameter, if multiple
                  hemangiomas, or if cavernous hemangioma

        228.040   Hemangioma, intra-abdominal (Always code regardless of size, type or
                  number)
        228.020   Hemangioma, intracranial (Always code regardless of size, type or
                  number)
        228.090   Hemangioma, of other sites (Always code regardless of size, type or
                  number)
     #  228.000   Hemangioma, of unspecified site. Always code if multiple hemangiomas
                  of any size are present, if one or more cavernous hemangiomas of any
                  size are present, or if a single hemangioma measuring ≥ 4cm in
                  diameter or desribed as large, huge, or of medical significance is
                  present.
```

228.030   Hemangioma, retinal (Always code regardless of size, type or number)

## Continued: Other Specified Codes Used in Metro Atlanta Congenital Defects Program

```
#  228.010  Hemangioma, skin & subcutaneous, NOS Always code if multiple
                hemangiomas of any size are present, if one or more cavernous
                hemangiomas of any size are present, or if a single hemangioma
                measuring ≥ 4cm in diameter or desribed as large, huge, or of medical
                significance is present.

#  286.000  Hemophilia (all types)
   774.490  Hepatitis, neonatal, NOS
   774.480  Hepatitis, neonatal, other specified
#  282.100  Hereditary elliptocytosis
#  282.000  Hereditary spherocytosis

   771.220  Herpes simplex (in utero infections only)
                Includes:    encephalitis
                             meningoencephalitis
   202.300  Histiocytosis, malignant
   277.510  Hurler syndrome
                Includes:    lipochondrodystrophy
#  778.600  Hydrocele, congenital
#  270.700  Hyperglycinemia
#  251.200  Hypoglycemia, idiopathic
#  252.100  Hypoparathyroidism, congenital
#  275.330  Hypophosphatemic rickets
   253.280  Hypopituitarism, congenital
 # 243.990  Hypothyroidism, congenital (Exclude even if other
                defects are present only if the record specifies
                hypothyroidism of prematurity. Other types of
                hypothyroidism or hypothyroidism NOS should continue
                to be on the routine exclusion list.)
   345.600  Infantile spasms, congenital
#  550.000  Inguinal hernia or patent processus vaginalis never
    -550.900  code in infants if <36 weeks gestation regardless of
                the presence of a reportable defect.
                NOTE: for those ≥36 weeks:
                Code in males only if another reportable defect is present;
                Code in females, always code even if found in isolation
   208.000  Leukemia, congenital, NOS
214    Lipoma

   214.300  Lipoma, intra-abdominal organs
   214.200  Lipoma, intrathoracic organs
   214.810  Lipoma, lumbar or sacral lipoma
                    paraspinal lipoma
   214.100  Lipoma, other skin and subcutaneous tissue
   214.800  Lipoma, other specified sites
   214.000  Lipoma, skin and subcutaneous tissue of face
   214.400  Lipoma, spermatic cord
   214.900  Lipoma, unspecified site
#  457.800  Lymphatics - other specified disorders of (including chylothorax)
   524.000  Macrognathia
#  270.300  Maple syrup urine disease
#  777.600  Meconium peritonitis
#  777.100  Meconium plug syndrome
   524.000  Micrognathia
   352.600  Moebius syndrome
```

```
    #  520.600  Natal teeth
       239.200  Neck cyst
       774.490  Neonatal hepatitis, NOS
```

## Continued: Other Specified Codes Used in Metro Atlanta Congenital Defects Program

```
       774.480  Neonatal hepatitis, other specified
       159.800  Neoplasms of the abdomen, other specified
       191.000  Neoplasms of the CNS
                Includes:   medulloblastoma, gliomas
       171.800  Neoplasms of the connective tissue
                Includes:   Ewing's sarcoma
                            fibrosarcoma
       155.000  Neoplasms of the liver
                Includes:   hepatoblastoma
                hemangio-epithelioma
       162.800  Neoplasms of the lung
       186.000  Neoplasms of the testes
       194.000  Neuroblastoma
       237.700  Neurofibromatosis
    #  379.500  Nystagmus
    #  270.100  Phenylketonuria (PKU)
    *  524.080  Pierre Robin sequence
    #  685.100  Pilonidal sinus (sacrodermal), sacral sinus, sacral dimple
    #  277.630  Pseudocholinesterase enzyme deficiency
    #  284.000  Red cell aplasia
       362.600  Retinal degeneration, peripheral
       362.700  Retinitis pigmentosa
       190.500  Retinoblastoma
       771.000  Rubella, congenital (in utero infections only)
    #  685.100  Sacral dimple
T   #  216.910  Sebaceous cyst
    #  282.600  Sickle cell anemia
    #  090.000  Syphilis, congenital (in utero infections only)
       238.030  Teratoma, abdomen
       238.010  Teratoma, head and face
       238.020  Teratoma, neck
       238.000  Teratoma, NOS
       238.080  Teratoma, other specified
       238.040  Teratoma, sacral, coccygeal
       257.800  Testicular feminization syndrome
       771.090  TORCH infection, unspecified (in utero infections only)
    #  608.200  Torsion of the testes or spermatic cord
       771.210  Toxoplasmosis (in utero infections only)
    #  553.100  Umbilical hernia
    #  286.400  von Willebrand disease
       335.000  Werdnig-Hoffman disease
       189.000  Wilms tumor (nephroblastoma)
       426.705  Wolfe-Parkinson-White syndrome, congenital
```

# Other Specified Codes Used in Metro Atlanta Congenital Defects Program

List ordered by 6-digit code number

```
#  090.000   Syphilis, congenital (in utero infections only)
   155.000   Neoplasms of the liver
             Includes:    hepatoblastoma
                          hmangio-epithelioma
   159.800   Neoplasms of the abdomen
   162.800   Neoplasms of the lung
   171.800   Neoplasms of connective tissue
             Includes:    Ewing's sarcoma
                          fibrosarcoma
   186.000   Neoplasms of the testes
   189.000   Wilms tumor (nephroblastoma)
   190.500   Retinoblastoma
   191.000   Neoplasms of the CNS
             Includes:    gliomas
                          mdulloblastoma
   194.000   Neuroblastoma
   202.300   Histiocytosis, malignant
   208.000   Leukemia, congenital, NOS

   214       Lipoma
   214.000   Lipoma, skin and subcutaneous tissue of face
   214.100   Lipoma, other skin and subcutaneous tissue
   214.200   Lipoma, intrathoracic organs
   214.300   Lipoma, intra-abdominal organs
   214.400   Lipoma, spermatic cord
   214.800   Lipoma, other specified sites
   214.810   Lipoma, lumbar or sacral lipoma
             paraspinal lipoma
   214.900   Lipoma, unspecified site

T  216       Benign neoplasm of skin
             (NOTE: All benign neoplasms should be coded ONLY if
              another reportable code is present)
             Includes:    blue nevus   pigmented nevus
                          papilloma    dermatofibroma
                          syringoadenoma
                          *dermoid cyst
                          hydrocystoma
                          syringoma
             Excludes:    skin of female genital organs (use 221.000),
                          skin of male genital organs (use 222.000)
#  216.000   Skin of lip
             Excludes:    vermillion border of lip
#  216.100   Eyelid, including canthus
             Excludes:    cartilage of eyelid
#  216.200   Ear and external auditory canal
             Includes:    auricle ear
                          external meatus
                          auricular canal
                          external canal
                          pinna
             Excludes:    cartilage of ear
```

R = Rev. 6/07
N = Rev. 5/07
T = Rev. 6/04
* = code created by CDC
# = on the MACDP Excl List

```
# 216.300  Skin of other and unspecified parts of face
            Includes:     cheek, external nose,
                          external eyebrow  temple
```

**Continued: Other Specified Codes Used in Metro Atlanta Congenital Defects Program**

```
      # 216.400  Scalp and skin of neck
      # 216.500  Skin of trunk, except scrotum
                         Includes:     axillary fold
                                       perianal skin
                       skin of:        chest wall
                                       abdominal wall
                                       groin
                                       buttock
                                       anus
                                       perineum
                                       back
                                       umbilicus
                                       breast
                 Excludes:     anal canal
                               anus, NOS
                               skin of scrotum
      # 216.600  Skin of upper limb, shoulder
      # 216.700  Skin of lower limb, hip
      # 216.800  Other specified sites of skin
                 Excludes:    epibulbar dermoid cyst (use 743.810)
      # 216.900  Site unspecified
      # 216.910  Sebaceous cyst
        216.920  Hairy nevus
      # 221.000  Benign skin neoplasm of female genital organs
      # 222.000  Benign skin neoplasm of male genital organs


T    228.0      Hemangioma
                 Include if greater than 4-inches diameter, if multiple
                 hemangiomas, or if cavernous hemangioma
      # 228.000  Hemangioma, of unspecified site
                 Always code if multiple hemangiomas of any size are present, if one
                 or more cavernous hemangiomas of any size are present, or if a single
                 hemangioma measuring ≥ 4cm in diameter or desribed as large, huge, or
                 of medical significance is present.
      # 228.010  Hemangioma, skin & subcutaneous, NOS
                 Always code if multiple hemangiomas of any size are present, if one
                 or more cavernous hemangiomas of any size are present, or if a single
                 hemangioma measuring ≥ 4cm in diameter or desribed as large, huge, or
                 of medical significance is present.
        228.020  Hemangioma, intracranial(Always code regardless of size, type or
                 number)
        228.030  Hemangioma, retinal (Always code regardless of size, type or number)
        228.040  Hemangioma, intra-abdominal(Always code regardless of size, type or
                 number)
        228.090  Hemangioma, of other sites(Always code regardless of size, type or
                 number)
        228.100  Cystic hygroma
                 Lymphangioma, any site
        237.700  Neurofibromatosis
        238.000  Teratoma, NOS
        238.010  Teratoma, head and face
        238.020  Teratoma, neck
        238.030  Teratoma, abdomen
        238.040  Teratoma, sacral, coccygeal
```

R = Rev. 6/07
N = Rev. 5/07                                        A - 89
T = Rev. 6/04
* = code created by CDC
# = on the MACDP Excl List

```
238.080   Teratoma, other specified
239.200   Neck cyst
```

R = Rev. 6/07
N = Rev. 5/07                           A - 90
T = Rev. 6/04
* = code created by CDC
# = on the MACDP Excl List

# Continued: Other Specified Codes Used in Metro Atlanta Congenital Defects Program

```
 #  243.990  Hypothyroidism, congenital
               (Exclude even if other defects are present only if the record
                specifies hypothyroidism of prematurity <36 weeks. Include other
                types of hypothyroidism and hypothyroidism NOS only when another
                reportable defect is present regardless of gestational age)
 #  251.200  Hypoglycemia, idiopathic
 #  252.100  Hypoparathyroidism, congenital
    253.280  Hypopituitarism, congenital
    253.820  Diencephalic syndrome
    255.200  Adrenogenital syndrome (adrenal hyperplasia)
    257.800  Testicular feminization syndrome
 #  270.100  Phenylketonuria (PKU)
 #  270.200  Albinism
 #  270.300  Maple syrup urine disease
 #  270.600  Arginosuccinic aciduria
 #  270.700  Hyperglycinemia
 #  271.000  Glycogen storage diseases
 #  275.330  Hypophosphatemic rickets
 #  277.000  Cystic fibrosis with no mention of meconium ileus
 #  277.010  Cystic fibrosis with mention of meconium ileus
    277.400  Disorders of bilirubin excretion
    277.510  Hurler syndrome
               Includes: lipochondrodystrophy
 #  277.620  Alpha-1 antitrypsin deficiency
 #  277.630  Pseudocholinesterase enzyme deficiency
    279.110  DiGeorge syndrome
    279.200  Combined immunodeficiency syndrome
 #  282.000  Hereditary spherocytosis
 #  282.100  Hereditary elliptocytosis
 #  282.200  Glucose-6-phosphate dehydrogenase (G-6-PD) deficiency
 #  282.600  Sickle cell anemia
 #  284.000  Red cell aplasia
 #  286.000  Hemophilia (all types)
 #  286.400  von Willebrand disease
 #  330.100  Cerebral lipidoses
               Includes:    Tay-Sachs disease
                            gangliosidosis
    331.890  Familial degenerative CNS disease
    335.000  Werdnig-Hoffman disease
    345.600  Infantile spasms, congenital
 #  351.000  Facial palsy
    352.600  Moebius syndrome
    362.600  Retinal degeneration, peripheral
    362.700  Retinitis pigmentosa
    363.200  Chorioretinitis
 #  368.000  Esotropia
 #  378.000  Exotropia
 #  379.500  Nystagmus
    425.300  Endocardial fibroelastosis
    426.705  Congenital Wolfe-Parkinson-White syndrome
    427.900  Cardiac arrhythmias, NEC. Never code premature atrial
               contractions, PACs.
    453.000  Budd-Chiari, occlusion of hepatic vein
 #  457.800  Other specified disorders of lymphatics (including chylothorax)
```

## Continued: Other Specified Codes Used in Metro Atlanta Congenital Defects Program

```
      # 520.600  Natal teeth
        524.000  Abnormalities of jaw size
                 micrognathia
                 macrognathia
      * 524.080  Pierre Robin sequence
     # 550.000-  Inguinal hernia or patent processus vaginalis never
       550.900   code in infants if <36 weeks gestation regardless of the
                 presence of a reportable defect.
                 NOTE: for those ≥36 weeks:
                   Code in males only if another reportable defect is present;
                   in females, always code even if found in isolation
      # 553.100  Umbilical hernia
        553.200  Epigastric hernia
      # 608.200  Torsion of testes or spermatic cord
   T  # 658.800  Amniotic bands (constricting bands, amniotic cyst)
      # 685.100  Pilonidal sinus (sacrodermal), sacral sinus, sacral dimple
        760.710  Fetal alcohol syndrome
        760.718  Probable fetal alcohol syndrome
                 Includes:    "facies"
        760.750  Fetal hydantoin (Dilantin) syndrome
      # 767.600  Erb's palsy

    771           Congenital infections (in utero infections only)
                  Excludes: congenital syphilis (use 090.000)

        771.000  Rubella, congenital
        771.090  TORCH infection, unspecified
        771.100  Cytomegalovirus (CMV)
        771.210  Toxoplasmosis
        771.220  Herpes simplex
                 Includes:    encephalitis
                              meningoencephalitis
        771.280  Congenital infection, other specified
                 Excludes:    human immunodeficiency virus (HIV) infection and
                              acquired immunodeficiency syndrome (AIDS)
        774.480  Hepatitis, neonatal, other specified
        774.490  Hepatitis, neonatal, NOS
      # 777.100  Meconium plug syndrome
      # 777.600  Meconium peritonitis
      # 778.000  Ascites, congenital
      # 778.600  Hydrocele, congenital
```

R = Rev. 6/07
N = Rev. 5/07
T = Rev. 6/04
* = code created by CDC
# = on the MACDP Excl List

**EXCLUSION LIST** for the MACDP
Nonreportable birth defects

**Conditions Never to be Reported**

The following newborn and infant conditions include those descriptions considered to
be excludable or nonreportable conditions in the MACDP.  This includes certain
biochemical disorders not considered part of the present MACDP case definition.

**Alphabetical** list of conditions that are <u>never</u> considered to be defects.

**Description**

|   |   |
|---|---|
|  | Anal fissure |
|  | Atrial contractions, premature |
|  | Breast hypertrophy |
|  | Bronchopulmonary dysplasia (Wilson-Mikity syndrome) |
|  | Cephalohematoma |
|  | Chalasia (gastroesophageal reflux) |
|  | CNS hemorrhage |
|  | Conjunctivitis |
|  | Diastasis recti |
|  | Epulis |
|  | Gastroesophageal reflux |
|  | Gum cysts - Includes epulis, ranula, mucocele |
|  | Hydrocephalus secondary to intraventricular hemorrhage (IVH) or CNS bleed |
|  | Hip click, with no follow-up or therapy |
| T | Heart murmur |
|  | Hyaline membrane disease |
| T | Intestinal obstruction - requires chart review to determine if cause of obstruction is a reportable defect. If so, code only the cause. |
| T | Intussusception - requires chart review to determine if cause of intussusception is a reportable defect. If so, code only the cause. |
|  | Inverted nipples |
|  | Laryngotracheomalacia or tracheomalacia |
|  | Meconium stained skin or nails |
|  | Mucocele |
|  | Neonatal acne |
|  | Overriding (overlapping) sutures |
|  | Petechiae |
|  | Phimosis |
|  | Pneumothorax |
|  | Premature atrial contractions |
|  | Protruding tongue |
|  | Ranula |
|  | Redundant foreskin |
|  | Retractile testes |
|  | Tracheomalacia |
| T | Volvulus - requires chart review to determine if cause of volvulus is a reportable defect. If so, code only the cause. |
|  | Wilson-Mikity syndrome |

R = Rev. 6/07
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T = Rev. 6/04
* = code created by CDC
# = on the MACDP Excl List

**EXCLUSION LIST** for the MACDP
Nonreportable birth defects

## Conditions Which may be Included Under Certain Conditions

The following newborn and infant conditions include those descriptions considered to be excludable or nonreportable conditions in the MACDP,  but which may be included under certain circumstances.

The following rules apply to coding these conditions:

   A.   If a condition or defect listed appears in a chart, singly or in any combination with other defects listed only on the Exclusion List, do not fill out the case record form.
   B.   If one of these conditions listed accompanies a reportable birth defect (from the 6-digit code manual and not on the exclusion list), then use the listed 6-digit code and record all defects (including those from this list) from the hospital record onto the case abstraction form.

**Alphabetical** list of conditions requiring no record abstraction to be performed unless associated with a reportable defect.  The addition or revision dates of the changes in the list of conditions requiring no record abstraction are shown.

| Revised/<br>Changed<br>Date | | | Code | Description |
|---|---|---|---|---|
| | | | 744.100 | Accessory auricle |
| | | | 757.650 | Accessory nipple (supernumerary nipple, or skin tag) |
| | | | 270.200 | Albinism |
| | | | 277.620 | Alpha 1-antitrypsin deficiency |
| | T | | 658.800 | Amniotic bands (constricting bands, amniotic cyst) |
| | | | 757.310 | Anal tags |
| 10/1/92 | T | | 746.400 | Aortic valve insufficiency or regurgitation, congenital – Code cases designated as 'mild', minimal', 'trivial', or 'physiologic' only if another reportable defect is present. Code all other degrees of insufficiency or regurgitation, including those where the degree is not specified, regardless of whether another reportable defect is present. |
| | | | 270.600 | Argininosuccinic aciduria |
| | T | | 778.000 | Ascites or anasarca, congenital. Includes: hydrops fetalis |
| | | | 744.220 | Bat ear |
| | T | # | 216.200 | Benign neoplasm of skin, ear and external auditory canal Includes:   auricle ear<br>              external meatus<br>              auricular canal<br>              external canal<br>              pinna<br>Excludes:  cartilage of ear |
| | T | # | 216.100 | Benign neoplasm of skin, eyelid, including canthus<br>Excludes:  cartilage of eyelid |
| | T | # | 216.000 | Benign neoplasm of skin, lip<br>Excludes:  vermillion border of lip |

R = Rev. 6/07
N = Rev. 5/07
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# = on the MACDP Excl List

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```
          T   #  216.700    Benign neoplasm of skin, lower limb, hip
          T   #  216.300    Benign neoplasm of skin, other and unspecified parts of face
                               Includes:  cheek, external nose, external eyebrow, temple
```

**EXCLUSION LIST** for the MACDP
Nonreportable birth defects

## Alphabetical - Conditions Which may be Included Under Certain Conditions

**Revised/**
**Changed**
**Date**                    **Code**        **Description**

```
          T   #  216.800    Benign neoplasm of skin, other specified sites of skin
                               Excludes:  epibulbar dermoid cyst (use 743.810)
          T   #  216.400    Benign neoplasm of skin, scalp and skin of neck
          T   #  216.900    Benign neoplasm of skin, site unspecified
              #  216.500    Benign neoplasm of skin, trunk, except scrotum
                             Includes:  axillary fold
                             perianal skin
                             skin of:   chest wall, abdominal wall, groin, buttock, anus,
                                           perineum, back, umbilicus, breast
                             Excludes:  anal canal, anus, NOS skin of scrotum
              #  216.600    Benign neoplasm of skin, upper limb, shoulder
                 221.000    Benign skin neoplasm of female genital organs
                 222.000    Benign skin neoplasm of male genital organs
                 754.020    Bent nose, deviation of nasal septum
                 744.820    Big lips
                 757.385    Birth mark, NOS
                 743.450    Blue sclera - if <36 weeks gestation, code only if
                            another reportable defect is present. Always code if
                            ≥36 weeks gestation.
                 743.800    Brushfield spots
                 757.390    Cafe au lait spots
                 746.860    Cardiomegaly, congenital NOS
                 744.230    Cauliflower ear
                 330.100    Cerebral lipidoses (e.g., Tay-Sachs, gangliosidoses,
                            etc.)
                 756.200    Cervical rib
                 755.500    Clinodactyly (incurving of fifth finger)
1/1/93           752.520    Cryptorchidism (see undescended testicle)
                 277.010    Cystic fibrosis, with mention of meconium ileus
                 277.000    Cystic fibrosis, with no mention of meconium ileus
                 744.280    Darwin's tubercle
1/1/96    T      754.030    Dolichocephaly - if <36 weeks gestation, code only if
                            another reportable defect is present. Always code if
                            ≥36 weeks gestation.
1/1/93           743.800    Downward eye slant (antimongoloid)
                 744.110    Ear tags, preauricular
                 744.120    Ear tags, other
                 744.230    Elfin ear, absent or decreased ear cartilage - if <36
                            weeks gestation, code only if another reportable
                            defect is present.
                 743.800    Epicanthal folds
                 767.600    Erb's palsy
                 368.000    Esotropia
                 378.000    Exotropia
```

R = Rev. 6/07
N = Rev. 5/07
T = Rev. 6/04
\* = code created by CDC
\# = on the MACDP Excl List

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|   | Code | Description |
|---|------|-------------|
|   | 351.000 | Facial palsy |
|   | 757.380 | Flammeus nevus or port wine stain |
|   | 748.180 | Flat bridge of nose |
|   | 754.040 | Fontanelle (large or small) |
| T | 743.630 | Fused eyelids - never code if <25 weeks gestation unless another reportable defect is present |

**EXCLUSION LIST** for the MACDP
Nonreportable birth defects

## Alphabetical - Conditions Which may be Included Under Certain Conditions

| Revised/ Changed Date | | Code | Description |
|-----------------------|---|------|-------------|
|        |   | 752.440 | Fusion of vulva |
|        |   | 282.200 | Glucose-6-phosphate dehydrogenase (G-6-PD) deficiency |
|        |   | 271.000 | Glycogen storage disease |
|        |   | 746.990 | Heart murmur - if chart review does not confirm a heart defect within 6 months, do not code as a defect even if other codable defects are present |
|        |   | 286.000 | Hemophilia |
|        |   | 751.620 | Hepatomegaly |
|        |   | 282.100 | Hereditary elliptocytosis |
|        |   | 282.000 | Hereditary spherocytosis |
| 3/4/91 |   | 750.240 | High arched palate |
|        |   | 778.600 | Hydrocele, congenital |
|        |   | 752.480 | Hymenal tags |
|        |   | 270.700 | Hyperglycinemia |
|        |   | 251.200 | Hypoglycemia, idiopathic |
|        |   | 252.100 | Hypoparathyroidism, congenital |
|        |   | 275.330 | Hypophosphatemic rickets |
| 1/1/96 | T | 752.440 | Hypoplastic labia majora - if <36 weeks gestation, code only if another reportable defect is present. Always code if ≥36 weeks gestation. |
| 3/4/91 | T | 748.510 | Hypoplasia of lung; pulmonary hypoplasia - exclude only if an isolated defect in infants <36 weeks gestation |
|        |   | 752.810 | Hypoplastic scrotum - exclude if secondary to undescended testes |
|        | T | 243.990 | Hypothyroidism, congenital (Exclude hypothyroidism of prematurity in infants <36 weeks gestation even if other reportable defects are present. Include other types of hypothyroidism and hypothyroidism NOS when another reportable defect is present regardless of gestational age) |
|        |   | 752.430 | Imperforate hymen |
|        |   | 755.500 | Incurving fingers (clinodactyly) |
|        | T | 550.000- 550.900 550.901 550.902 | Inguinal hernia or patent processus vaginalis. Never code in infants <36 weeks gestation regardless of the presence of a reportable defect. For infants ≥36 weeks: In **males,** code only if another reportable defect is present; In **females**, always code even if found in isolation |
|        |   | 757.450 | Lanugo, excessive or persistent |
|        |   | 754.040 | Large fontanelle |

R = Rev. 6/07
N = Rev. 5/07
T = Rev. 6/04
* = code created by CDC
# = on the MACDP Excl List

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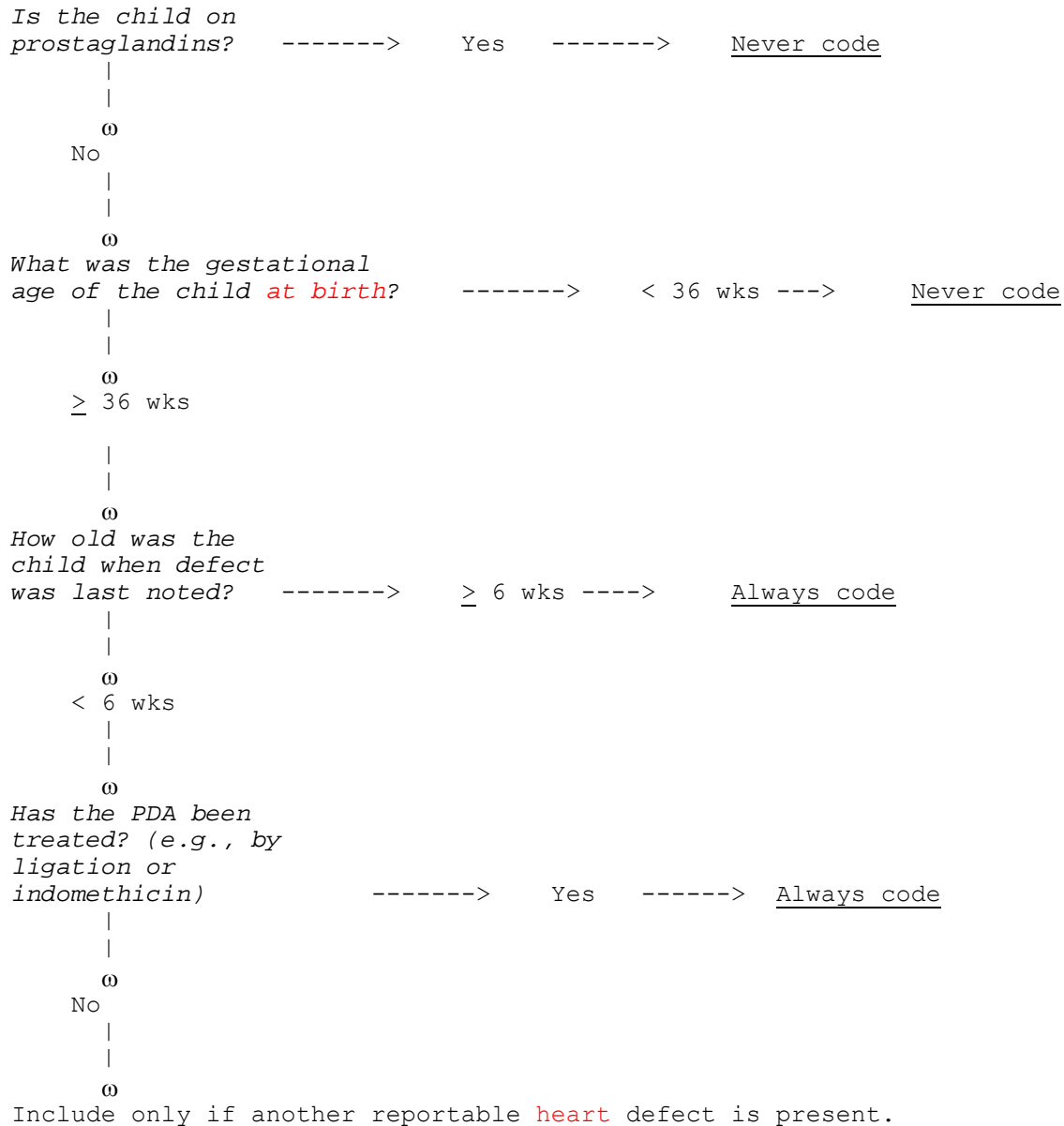|          |       | 755.500 | Long fingers and toes |
|          |       | 744.230 | Lop ear |
|          |       | 744.245 | Low set ears |
|          |       | 744.820 | Macrocheilia (big lips) |
|          |       | 270.300 | Maple syrup urine disease |
|          |       | 751.010 | Meckel's diverticulum |
|          |       | 777.600 | Meconium peritonitis |
|          |       | 777.100 | Meconium plug |
| 9/10/90  |       | 754.520 | Metatarsus varus or adductus |

**EXCLUSION LIST** for the MACDP
Nonreportable birth defects

## Alphabetical - Conditions Which may be Included Under Certain Conditions

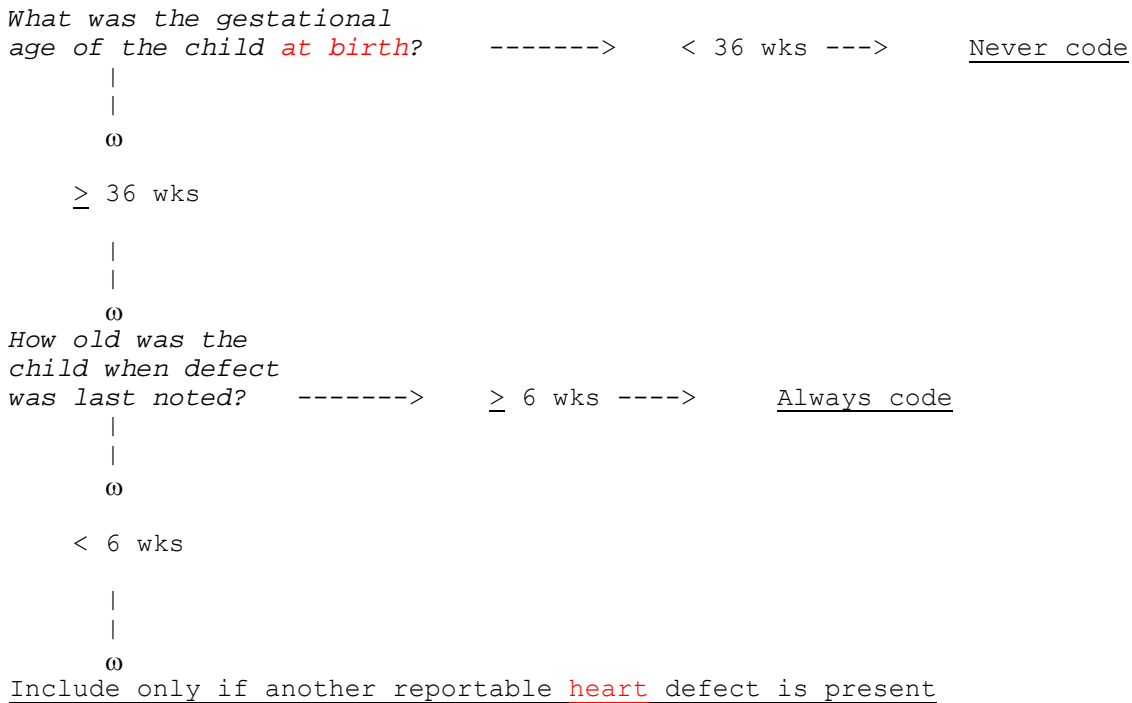| Revised/<br>Changed<br>Date |     | Code | Description |
|------|-----|------|-------------|
|      |     | 744.830 | Microcheilia (small lips) |
| 10/1/92 | T | 746.600 | Mitral valve insufficiency or regurgitation, congenital - Code cases designated as 'mild', minimal', 'trivial', or 'physiologic' only if another reportable defect is present. Code all other degrees of insufficiency or regurgitation, including those where the degree is not specified, regardless of whether another reportable defect is present. |
|      |     | 757.386 | Mongolian spots |
|      |     | 743.650 | Nasal lacrimal duct obstruction |
|      |     | 520.600 | Natal teeth |
|      |     | 745.500 | Nonclosure of foramen ovale, NOS (see PFO) |
|      |     | 379.500 | Nystagmus |
| 9/10/90 |   | 756.080 | Occiput, flat or prominent |
| 3/5/90 |   | 457.800 | Other specified disorder of lymphatics, including chylothorax |
|      |     | 755.600 | Overlapping toes |
| 10/14/92 | T | 747.000 | Patent ductus arteriosus (PDA)<br>1) Always code if ≥36 weeks of gestation and defect last noted at ≥6 weeks of age.<br>2) If ≥36 weeks gestation and defect last noted <6 weeks of age, code only if the PDA was treated )e.g. by ligation or indomethicin) or if another reportable defect is present.<br>3) Never code if <36 weeks gestation or if treated with prostaglandins regardless of gestational age. |
| 10/14/92 | T # | 745.500 | Nonclosure of foramen ovale, NOS<br>Patent foramen ovale (PFO)<br>1) Always code if ≥36 weeks of gestation and defect last noted at ≥6 weeks of age.<br>2) If ≥36 weeks gestation and defect last noted <6 weeks of age, code only if another reportable defect is present.<br>3) Never code if <36 weeks gestation regardless of presence of other defects. |
|      | T   | 753.700 | Patent urachus |
|      |     | 744.820 | Patulous lips (wide lips) |
| 8/1/93 |   | 747.325 | Peripheral pulmonic stenosis (PPS) murmur - <u>do</u> collect |

R = Rev. 6/07
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T = Rev. 6/04
* = code created by CDC
# = on the MACDP Excl List

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```
                         if PPS documented by echocardiogram
           270.100       Phenylketonuria (PKU)
           685.100       Pilonidal or sacral dimple
           744.230       Pixie-like ear
           744.230       Pointed ear
           755.006       Polydactyly in blacks (postaxial, type B), includes
                         only skin tags on hands or feet. All other types of
                         postaxial polydactyly (i.e. extra finger with bone,
                         nail, etc.) should always be coded.
           744.246       Posteriorly rotated ears
           744.410       Preauricular sinus, cyst or pit
           744.110       Preauricular tags
                    EXCLUSION LIST for the MACDP
                      Nonreportable birth defects
```

## Alphabetical - Conditions Which may be Included Under Certain Conditions

| Revised/ Changed Date | | Code | Description |
|---|---|---|---|
| | T | 747.680 | Primary pulmonary artery hypertension |
| | | 752.450 | Prominent clitoris |
| | | 277.630 | Pseudocholinesterase enzyme deficiency |
| 10/1/92 | T | 746.020 | Pulmonary valve insufficiency or regurgitation, congenital |
| | | | Code cases designated as 'mild', minimal', 'trivial', or 'physiologic' only if another reportable defect is present. Code all other degrees of insufficiency or regurgitation, including those where the degree is not specified, regardless of whether another reportable defect is present. |
| | | 750.500 | Pylorospasm (intermittent pyloric stenosis) |
| | | 751.580 | Rectal fissures |
| | | 284.000 | Red cell aplasia |
| | | 744.500 | Redundant neck skin folds |
| | | 755.616 | Rocker-bottom feet |
| | | 685.100 | Sacral dimple |
| 1/1/96 | T | 754.060 | Scaphocephaly, no mention of craniosynostosis |
| | | | Always code if ≥36 weeks gestation. |
| | # | | If <36 weeks gestation, code only if another reportable defect is present. |
| 1/1/96 | T | 216.910 | Sebaceous cysts |
| | | 744.900 | Short neck |
| | | 282.600 | Sickle cell anemia |
| | | 757.200 | Sidney line |
| | | 757.200 | Simian crease (transverse palmar crease) |
| | | 747.500 | Single umbilical artery |
| | | 757.390 | Skin cysts |
| | | 754.040 | Small fontanelle |
| | | 744.830 | Small lips |
| 1/1/96 | T | 757.640 | Small nipple (hypoplastic) |
| | | | Always code if ≥36 weeks gestation. |
| | # | | If <36 weeks gestation, code only if another reportable defect is present |
| 10/1/92 | T | 759.020 | Splenomegaly |
| 7/13/92 | | 090.000 | Syphilis, congenital |
| | | 759.240 | Thymic hypertrophy |

```
       R = Rev. 6/07
       N = Rev. 5/07
       T = Rev. 6/04
       * = code created by CDC
       # = on the MACDP Excl List
```

|  |  | 755.630 | Tibial torsion |
|---|---|---|---|
|  |  | 750.000 | Tongue-tie |
|  |  | 608.200 | Torsion of spermatic cord |
|  |  | 608.200 | Torsion of testes |
| 10/1/92 | T | 746.105 | Tricuspid valve insufficiency or regurgitation, congenital – Code cases designated as 'mild', minimal', 'trivial', or 'physiologic' only if another reportable defect is present. Code all other degrees of insufficiency or regurgitation, including those where the degree is not specified, regardless of whether another reportable defect is present. |
|  |  | 759.900 | Umbilical cord atrophy |
|  |  | 553.100 | Umbilical hernias (completely covered by skin) |

**EXCLUSION LIST** for the MACDP
Nonreportable birth defects

## Alphabetical - Conditions Which may be Included Under Certain Conditions

| Revised/ Changed Date | | Code | Description |
|---|---|---|---|
| 1/1/93 | T | 752.500- | Undescended testicle (cryptorchidism) |
|  | T | 752.520 | 1) If < 36 weeks gestation, code only if there is a medical/surgical intervention for this problem; 2) If ≥36 weeks gestation and defect last noted at <1 year of age, code only if there was a medical/surgical intervention for this problem or if another reportable defect is present 3) Always code if ≥36 weeks gestation and defect first noted at ≥1 of age. |
|  |  | 748.180 | Upturned nose |
|  |  | 743.800 | Upward eye slant (mongoloid) |
|  |  | 752.460 | Vaginal cysts |
|  |  | 752.480 | Vaginal tags |
|  |  | 286.400 | von Willebrand's disease |
| 3/14/91 | T | 755.130 | Webbed toes Code webbing of the second and third toes only if another reportable defect is present. Always code webbing of other toes regardless of whether another reportable defect is present |
|  |  | 744.500 | Webbing of neck |
|  |  | 748.180 | Wide nasal bridge |
|  |  | 755.600 | Widely spaced first and second toes |
|  |  | 757.680 | Widely spaced nipples |

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* = code created by CDC
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**EXCLUSION LIST** for the MACDP

**Numerical** list of conditions requiring <u>no</u> record abstraction <u>unless</u> associated with a reportable defect.  The addition or revision dates of the changes in the list of conditions requiring no record abstraction are shown. .

**Revised/
Changed
<u>Date</u>**                    **<u>Code</u>**          **<u>Description</u>**

7/13/92              090.000    Syphilis congenital
              216          Benign neoplasm of skin
                          (NOTE: All benign neoplasms should be coded ONLY if
                          another reportable code is present)
                          Includes:
                                blue nevus
                                pigmented nevus
                                papilloma
                                dermatofibroma
                                syringoadenoma
                                *dermoid cyst
                                hydrocystoma
                                syringoma
                       Excludes:    skin of female genital organs (use 221.000),
                                skin of male genital organs (use 222.000)
      #      216.000    Skin of lip
                          Excludes:  vermillion border of lip
      #      216.100    Eyelid, including canthus
                          Excludes:  cartilage of eyelid
      #      216.200    Ear and external auditory canal
                          Includes:  auricle ear
                                    external meatus
                                    auricular canal
                                    external canal
                                    pinna
                          Excludes:  cartilage of ear
      #      216.300    Skin of other and unspecified parts of face
                          Includes: cheek,external nose, external eyebrow, temple
      #      216.400    Scalp and skin of neck
      #      216.500    Skin of trunk, except scrotum
                          Includes:  axillary fold
                                    perianal skin
                                    skin of:   chest wall
                                              abdominal wall
                                              groin
                                              buttock
                                              anus
                                              perineum
                                              back
                                              umbilicus
                                              breast
                          Excludes:  anal canal
                                    anus, NOS
                                    skin of scrotum
      #      216.600    Skin of upper limb, shoulder
      #      216.700    Skin of lower limb, hip
      #      216.800    Other specified sites of skin
                          Excludes:  epibulbar dermoid cyst (use 743.810)

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* = code created by CDC                      A -100
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```
      #        216.900     Site unspecified
      #        216.910     Sebaceous cyst
```

**EXCLUSION LIST** for the MACDP

**Numerical** list of conditions requiring <u>no</u> record abstraction <u>unless</u> associated with a reportable defect.  The addition or revision dates of the changes in the list of conditions requiring no record abstraction are shown. .

**Revised/**
**Changed**

| Date | | Code | Description |
|---|---|---|---|
| | | 221.000 | Benign skin neoplasm of female genital organs |
| | | 222.000 | Benign skin neoplasm of male genital organs |
| | T | 243.990 | Hypothyroidism, congenital (Exclude even if other defects are present only if the record specifies hypothyroidism of prematurity <36 weeks. Include other types of hypothyroidism and hypothyroidism NOS only when another reportable defect is present regardless of gestational age. |
| | | 251.200 | Hypoglycemia, idiopathic |
| | | 252.100 | Hypoparathyroidism, congenital |
| | | 270.100 | Phenylketonuria (PKU) |
| | | 270.200 | Albinism |
| | | 270.300 | Maple syrup urine disease |
| | | 270.600 | Argininosuccinic aciduria |
| | | 270.700 | Hyperglycinemia |
| | | 271.000 | Glycogen storage diseases |
| | | 275.330 | Hypophosphatemic rickets |
| | | 277.000 | Cystic fibrosis, with no mention of meconium ileus |
| | | 277.010 | Cystic fibrosis, with mention of meconium ileus |
| | | 277.620 | Alpha 1-antitrypsin deficiency |
| | | 277.630 | Pseudocholinesterase enzyme deficiency |
| | | 282.000 | Hereditary spherocytosis |
| | | 282.100 | Hereditary elliptocytosis |
| | | 282.200 | Glucose-6-phosphate dehydrogenase (G-6-PD) deficiency |
| | | 282.600 | Sickle cell anemia |
| | | 284.000 | Red cell aplasia |
| | | 286.000 | Hemophilia |
| | | 286.400 | von Willebrand's disease |
| | | 330.100 | Cerebral lipidoses (e.g., Tay-Sachs, gangliosidoses, etc.) |
| | | 351.000 | Facial palsy |
| | | 368.000 | Esotropia |
| | | 378.000 | Exotropia |
| | | 379.500 | Nystagmus |
| 3/5/90 | | 457.800 | Other specified disorder of lymphatics, including chylothorax |
| | | 520.600 | Natal teeth |
| | T | 550.000-550.900 | Inguinal hernia or patent processus vaginalis never code in infants if <36 weeks gestation regardless of the presence of a reportable defect. <u>NOTE</u>: for those ≥36 weeks: in **males,** code only if another reportable defect is present; in **females,** always code even if found in isolation |
| | | 553.100 | Umbilical hernias (completely covered by skin) |

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|   |   | 608.200 | Torsion of spermatic cord |
|---|---|---------|---------------------------|
|   |   | 608.200 | Torsion of testes |
|   | T | 658.800 | Amniotic bands (constricting bands, amniotic cyst) |
|   |   | 685.100 | Pilonidal or sacral dimple |

**EXCLUSION LIST** for the MACDP
Nonreportable birth defects

## Numerical - Conditions Which may be Included Under Certain Conditions

| Revised/ Changed Date |   | Code | Description |
|-----------------------|---|------|-------------|
|   | T | 743.450 | Blue sclera - if <36 weeks gestation, code only if another reportable defect is present. Always code if ≥36 weeks gestation. |
|   | T | 743.630 | Fused eyelids – never code if <25 weeks gestation unless another reportable defect is present |
|   |   | 743.650 | Nasal lacrimal duct obstruction |
|   |   | 743.800 | Brushfield spots |
|   |   | 743.800 | Downward eye slant (antimongoloid) |
|   |   | 743.800 | Epicanthal folds |
|   |   | 743.800 | Upward eye slant (mongoloid) |
|   |   | 744.100 | Accessory auricle |
|   |   | 744.110 | Ear tags, preauricular |
|   |   | 744.120 | Ear tags, other |
|   |   | 744.220 | Bat ear |
|   |   | 744.230 | Cauliflower ear |
|   |   | 744.230 | Elfin ear, absent or decreased ear cartilage  If <36 weeks gestation, code only if another reportable defect is present. |
|   |   | 744.230 | Lop ear |
|   |   | 744.230 | Pixie-like ear |
|   |   | 744.230 | Pointed ear |
|   |   | 744.245 | Low set ears |
|   |   | 744.246 | Posteriorly rotated ears |
|   |   | 744.280 | Darwin's tubercle |
|   |   | 744.410 | Preauricular sinus, cyst or pit |
|   |   | 744.500 | Redundant neck skin folds |
|   |   | 744.500 | Webbing of neck |
|   |   | 744.820 | Macrocheilia (big lips) |
|   |   | 744.820 | Patulous lips (wide lips) |
|   |   | 744.830 | Microcheilia (small lips) |
|   |   | 744.900 | Short neck |
|   |   | 745.500 | Nonclosure of foramen ovale, NOS (see PFO) |
| 10/14/92 | T | 745.500 | Patent foramen ovale (PFO) 1) Always code if ≥36 weeks of gestation and defect last noted at ≥6 weeks of age. 2) If ≥36 weeks gestation and defect last noted <6 weeks of age, code only if another reportable defect is present. 3) Never code if <36 weeks gestation regardless of presence of other defects. |
| 10/1/92 | T | 746.020 | Pulmonary valve insufficiency or regurgitation, congenital - Code cases designated as 'mild', minimal', 'trivial', or 'physiologic' only if another reportable defect is |

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present. Code all other degrees of insufficiency or
regurgitation, including those where the degree is not
specified, regardless of whether another reportable
defect is present.


**EXCLUSION LIST** for the MACDP
Nonreportable birth defects

## Numerical - Conditions Which may be Included Under Certain Conditions

| Revised/ Changed Date | | Code | Description |
|---|---|---|---|
| 10/1/92 | T | 746.105 | Tricuspid valve insufficiency or regurgitation, congenital - Code cases designated as 'mild', minimal', 'trivial', or 'physiologic' only if another reportable defect is present. Code all other degrees of insufficiency or regurgitation, including those where the degree is not specified, regardless of whether another reportable defect is present. |
| 10/1/92 | T | 746.400 | Aortic valve insufficiency or regurgitation, congenital - Code cases designated as 'mild', minimal', 'trivial', or 'physiologic' only if another reportable defect is present. Code all other degrees of insufficiency or regurgitation, including those where the degree is not specified, regardless of whether another reportable defect is present. |
| 10/1/92 | T | 746.600 | Mitral valve insufficiency or regurgitation, congenital - Code cases designated as 'mild', minimal', 'trivial', or 'physiologic' only if another reportable defect is present. Code all other degrees of insufficiency or regurgitation, including those where the degree is not specified, regardless of whether another reportable defect is present. |
| | | 746.860 | Cardiomegaly, congenital NOS |
| | | 746.990 | Heart murmur - if chart review does not confirm a heart defect within 6 months, do not code as a defect even if other codable defects are present |
| 10/14/92 | T | 747.000 | Patent ductus arteriosus (PDA) 1) Always code if ≥36 weeks of gestation and defect last noted at ≥6 weeks of age. 2) If ≥36 weeks gestation and defect last noted <6 weeks of age, code only if the PDA was treated )e.g. by ligation or indomethicin) or if another reportable defect is present. 3) Never code if <36 weeks gestation or if treated with prostaglandins regardless of gestational age. |
| 8/1/93 | | 747.325 | Peripheral pulmonic stenosis (PPS) murmur - do collect if PPS documented by echocardiogram |
| | | 747.500 | Single umbilical artery |
| | | 747.680 | Primary pulmonary artery hypertension |

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| | | 778.000 | Ascites or anasarca. Includes: hydrops fetalis |
| | | 748.180 | Flat bridge of nose |
| | | 748.180 | Upturned nose |
| | | 748.180 | Wide nasal bridge |
| 3/4/91 | T | 748.510 | Hypoplasia of lung; pulmonary hypoplasia - exclude if isolated defect in infants <36 weeks gestation. |
| | | 750.000 | Tongue-tie |
| 3/4/91 | | 750.240 | High arched palate |
| | | 750.500 | Pylorospasm (intermittent pyloric stenosis) |
| | | 751.010 | Meckel's diverticulum |

**EXCLUSION LIST** for the MACDP
Nonreportable birth defects

## Numerical - Conditions Which may be Included Under Certain Conditions

**Revised/
Changed**

| | | Date | Code | Description |
|---|---|---|---|---|
| | | | 751.580 | Rectal fissures |
| | | | 751.620 | Hepatomegaly |
| | | | 752.430 | Imperforate hymen |
| | | | 752.440 | Fusion of vulva |
| 1/1/96 | T | | 752.440 | Hypoplastic labia majora - if <36 weeks gestation, code only if another reportable defect is present. Always code if ≥36 weeks gestation. |
| 3/4/91 | | | 752.450 | Prominent clitoris |
| | | | 752.460 | Vaginal cysts |
| | | | 752.480 | Vaginal tags |
| | | | 752.480 | Hymenal tags |
| 1/1/93 | T | | 752.500- | Undescended testicle (cryptorchidism) |
| | T | | 752.520 | 1) If < 36 weeks gestation, code only if there is a medical/surgical intervention for this problem; 2) If ≥36 weeks gestation and defect last noted at <1 year of age, code only if there was a medical/surgical intervention for this problem or if another reportable defect is present 3) Always code if ≥36 weeks gestation and defect first noted at ≥1 of age. |
| 1/1/93 | | | 752.520 | Cryptorchidism (see undescended testicle) |
| | | | 752.810 | Hypoplastic scrotum - exclude if secondary to undescended testes |
| | | | 753.700 | Patent urachus |
| | | | 754.020 | Bent nose, deviation of nasal septum |
| 1/1/96 | T | | 754.030 | Dolichocephaly - if <36 weeks gestation, code only if another reportable defect is present. Always code if ≥36 weeks gestation. |
| 1/1/93 | | | 754.040 | Fontanelle (large or small) |
| 1/1/96 | | | 754.060 | Scaphocephaly, no mention of craniosynostosis If <36 weeks gestation, code only if another reportable defect is present. Always code if ≥36 weeks gestation. |
| 1/1/93 | | | 754.520 | Metatarsus varus or adductus |
| | | | 755.006 | Polydactyly in blacks (postaxial, type B), includes only skin tags on hands or feet. All other types of postaxial polydactyly (i.e. extra finger with bone, nail, etc.) should always be coded. |

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| 3/14/91 | T | 755.130 | Webbed toes |
|---------|---|---------|-------------|

Code webbing of the second and third toes only if another reportable defect is present. Always code webbing of other toes regardless of whether another reportable defect is present

| | | 755.500 | Clinodactyly (incurving of fifth finger) |
|--|--|---------|------------------------------------------|
| | | 755.500 | Long fingers and toes |
| | | 755.600 | Overlapping toes |
| | | 755.600 | Widely spaced first and second toes |
| | | 755.616 | Rocker-bottom feet |
| | | 755.630 | Tibial torsion |
| | | 756.080 | Occiput, flat or prominent |
| | | 756.200 | Cervical rib |

**EXCLUSION LIST** for the MACDP
Nonreportable birth defects

## Numerical - Conditions Which may be Included Under Certain Conditions

| Revised/ Changed Date | | Code | Description |
|-----------------------|--|------|-------------|
| | | 757.200 | Sidney line |
| | | 757.200 | Simian crease (transverse palmar crease) |
| | | 757.310 | Anal tags |
| | | 757.380 | Flammeus nevus or port wine stain |
| | | 757.385 | Birth mark, NOS |
| | | 757.386 | Mongolian spots |
| | | 757.390 | Cafe au lait spots |
| | | 757.390 | Skin cysts |
| | | 757.450 | Lanugo, excessive or persistent |
| 1/1/96 | T | 757.640 | Small nipple (hypoplastic) |

If <36 weeks gestation, code only if another reportable defect is present.
Always code if ≥36 weeks gestation.

| 9/10/90 | | 757.650 | Accessory nipple (supernumerary nipple, or skin tag) |
|---------|--|---------|------------------------------------------------------|
| | | 757.680 | Widely spaced nipples |
| | | 759.020 | Splenomegaly |
| | | 759.240 | Thymic hypertrophy |
| | | 759.900 | Umbilical cord atrophy |
| | | 767.600 | Erb's palsy |
| | | 777.100 | Meconium plug |
| | | 777.600 | Meconium peritonitis |
| | | 778.000 | Ascites or anasarca, congenital |
| | | 778.600 | Hydrocele, congenital |

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# MACDP Decision Tree for Determining Whether to Include Patent Ductus Arteriosus (PDA)

```
Is the child on
prostaglandins?    ------->    Yes   ------->    Never code
        |
        |
        ω
     No
        |
        |
        ω
What was the gestational
age of the child at birth?    ------->    < 36 wks --->    Never code
        |
        |
        ω
     > 36 wks


        |
        |
        ω
How old was the
child when defect
was last noted?    ------->    > 6 wks ---->    Always code
        |
        |
        ω
     < 6 wks
        |
        |
        ω
Has the PDA been
treated? (e.g., by
ligation or
indomethicin)             ------->    Yes   ------>  Always code
        |
        |
        ω
     No
        |
        |
        ω
Include only if another reportable heart defect is present.
```

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# MACDP Decision Tree for Determining Whether to Include Patent Foramen Ovale (PFO)

```
What was the gestational
age of the child at birth?    ------->    < 36 wks --->    Never code
     |
     |
     ω

   > 36 wks
_

     |
     |
     ω
How old was the
child when defect
was last noted?   ------->    > 6 wks ---->    Always code
     |                       _
     |
     ω

   < 6 wks


     |
     |
     ω
Include only if another reportable heart defect is present
```

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N = Rev. 5/07
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* = code created by CDC
# = on the MACDP Excl List

# MACDP Decision Tree for Determining Whether to Include Peripheral Pulmonary Stenosis (PPS)

```
What was the gestational
age of the child at birth?      ------->    < 36 wks --->     Never code
     |
     |
     ω

   > 36 wks

     |
     |
     ω
How old was the
child when defect
was last noted?    ------->    > 6 wks ---->     Always code
     |
     |
     ω

   < 6 wks

     |
     |
     ω
Include only if another reportable heart defect is present
```

May 22, 1996

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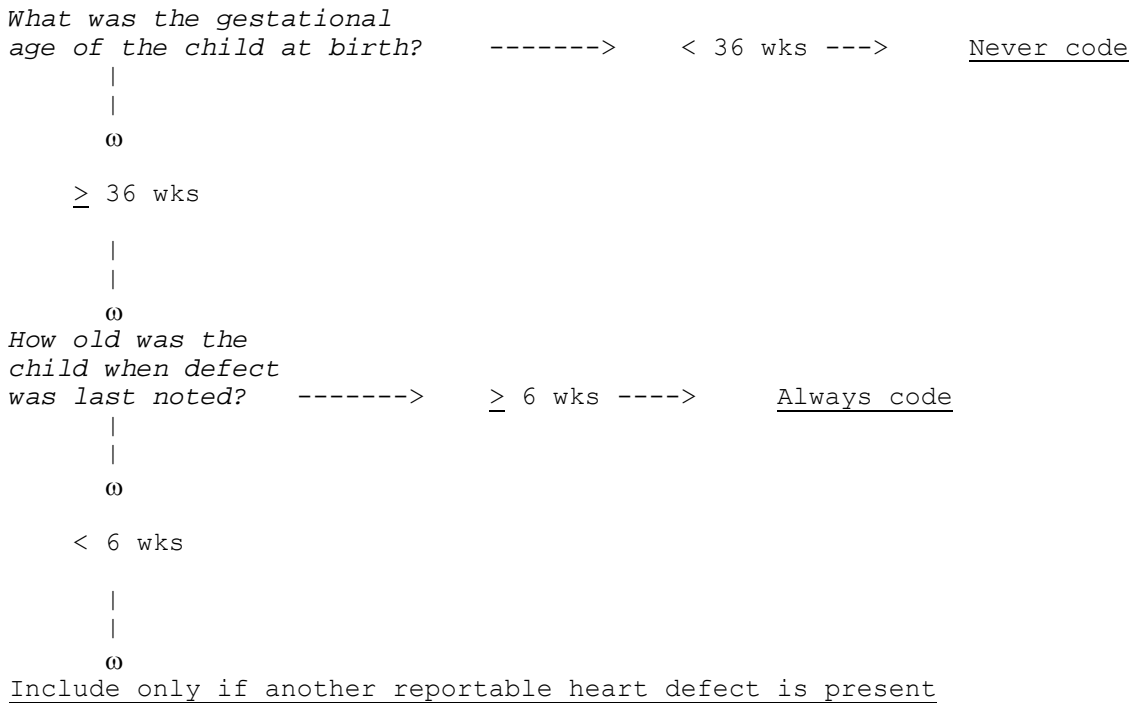
*Chapter 6*

*Case Ascertainment Methods*

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# Appendices

# 6.1 Introduction

The National Birth Defects Prevention Network (NBDPN) is committed to improving the quality, accuracy, completeness, comparability, and timeliness of birth defects surveillance data. Information on the prevalence of birth defects reported by surveillance systems can vary considerably due to differences in case definition, method of case ascertainment, and the types of data sources used.

This chapter describes two major approaches to birth defects surveillance: active case ascertainment and passive case ascertainment. The ***active case ascertainment*** approach is the intensive level of case identification that involves staff finding cases at strategic data sources. Ascertainment is usually very complete, and each diagnosis in the database is confirmed. In the ***passive case ascertainment*** approach the surveillance program receives case reports of birth defects from data sources. The completeness of ascertainment is highly dependent on the number and types of data sources used by the surveillance program and on the consistency of case reporting from the data sources. Since case reports usually are not confirmed by staff in a passive case ascertainment program, it is particularly important for these programs to implement quality assurance procedures aggressively.

Although the two surveillance approaches are operationally different, it is possible to achieve comparable levels of data quality. Programs should evaluate their surveillance approaches regularly for accuracy, completeness, and timeliness and should be creative in identifying strategic means of quality improvement.

In this chapter we first introduce some relevant terminology (Section 6.2). We then discuss general surveillance development (Section 6.3) and introduce approaches to case identification (Section 6.4). In Sections 6.5 and 6.6 we present in some detail information on the two main approaches to case identification (active and passive case ascertainment, respectively). The remaining sections cover additional topics in case ascertainment, including data sources (Section 6.7), sources of information that may be available at a given data source (Section 6.8), and issues relating to infant risk factors and case identification (Section 6.9). References cited in this chapter may be found in Section 6.10.

Appendices to this chapter provide additional detail on the following important data sources for birth defects surveillance: vital records (Appendix 6.1), hospital data sets (Appendix 6.2), hospital and patient services logs (Appendix 6.3), and genetic services (Appendix 6.4).

# 6.2  Terminology

| | |
|---|---|
| **Surveillance (public health)** | The ongoing, systematic collection, analysis, and interpretation of health data essential to the planning, implementation and evaluation of public health practice, closely integrated with the timely dissemination of these data to those who need to know (Centers for Disease Control and Prevention, 1988). |
| **Monitoring** | The performance and analysis of routine measurements using statistical methods aimed at detecting changes in the environment or health status of populations (Last, 1995). |
| **Registry** | A system of ongoing registrations, such that cases of a disease or other health-relevant condition are defined in a population and can be related to a population base. Birth and death registration systems are examples. Some disease registries, like the cancer registry, closely resemble public health surveillance systems and have epidemiologic value (Last, 1995). |
| **Case ascertainment or identification** | The process of identifying – from existing sources and using defined case definitions – embryos, fetuses, neonates, infants, and children who have a birth defect. |
| **Active case ascertainment** | A surveillance approach to case identification that is based on surveillance staff being engaged intensively in all activities related to finding and confirming potential birth defects cases. Surveillance staff seek out data sources and conduct systematic investigations of pertinent sources of information to identify potential cases of birth defects. Data collection sites include hospital medical records, diagnostic indices, unit logs, pathology departments, and specialty sites. |
| **Passive case ascertainment** | A surveillance approach to case identification whereby birth defects programs receive case reports from data sources. Passive case ascertainment programs receive case reports from one or many different data sources and may accommodate multiple reporting formats including hard copy, electronic, and web-based reports, as well as administrative data sets. There may be variability in the completeness and accuracy of case ascertainment in programs that use this type of case ascertainment. |
| **Population-based surveillance** | Surveillance that identifies a population under study, usually defined by geopolitical boundaries, and establishes the denominator from which cases come. A data source that is population based covers an entire population within a defined area. Some examples of population-based data sources are: vital records (birth, death, and fetal death), statewide newborn genetic screening programs, and statewide newborn hearing screening programs. |

| | |
|---|---|
| **Data source** | Any facility, site, or entity that has cases or potential cases of birth defects or other pertinent medical information. This includes a hospital, clinic, physician's office, laboratory, prenatal diagnosis center, as well as administrative databases. |
| **Reportable disease** | A disease, laboratory result, or health condition of public health significance that requires notification of its occurrence to a public health agency. Authorizing legislation or regulations usually define which conditions are reportable, which data sources are required to report, timelines for reporting, and what demographic information is required, at a minimum, in a case report. |
| **Reporting source** | A data source that is required, by law, to report or allow access to cases of birth defects and other pertinent medical conditions to the birth defects program. |
| **Administrative data set** | A data set or database that is created to fulfill operational or managerial objectives. Many are developed as information management systems with multiple functions. Examples include hospital discharge data, Medicaid data, vital records master index, clinical management information systems, health care billing and insurance claims systems. |
| **Unit** | A component, section, or department within a data source that serves a specific function or performs a specific activity. Examples include health information management department, labor and delivery unit, neonatal intensive care unit, and pathology department |
| **Data collection** | The process of gathering information, which includes receiving, retrieving, accessing, abstracting, and extracting information from information sources. |
| **Medical records review (information review)** | The process of reading, identifying, interpreting, and translating documentation per specific program objectives. Medical records review precedes abstracting. |
| **Abstracting** | The process of recording information, identified when reviewing documentation in a medical record or other information source, and entering the information into data fields in a specified format. Information may be recorded on hard copy forms or through formatted data entry computer screens. |
| **Disease coding** | The process of assigning a standardized disease code (e.g., ICD-9-CM or 6-digit CDC code) to medical information. |
| **Case abstract or case record** | The documentation file(s) containing complete information about the birth defects case. |

# 6.3  General Surveillance Development

Birth defects surveillance systems should be developed to facilitate the essential activities of data collection, data analysis, data evaluation, and information dissemination consistent with a program's established goals and objectives. The general guidelines below can be applied to developing a new system or improving an existing system. We are indebted to Mausner and Bahn (1974) and Teutsch and Churchill (2000) for much of the information in this section.

In the following sections we discuss planning and documenting the system (Section 6.3.1), identifying data sources (Section 6.3.2), obtaining knowledge about individual data sources (Section 6.3.3), implementing data quality procedures (Section 6.4.3), and evaluating surveillance method and analytical capability (Section 6.3.5).

## 6.3.1  Plan and Document

A birth defects surveillance program must be organized and have supporting documentation before beginning operations. The program can begin to process case reports once the logistics of case identification and data collection are established with data sources. Therefore, it is important to engage surveillance staff, data sources, stakeholders, advisors, and others affected by program operations early in the planning process.

The program should:

➤ **Understand** the legal authority and restrictions that shape surveillance operations, including processes for changing or amending legislation (see Chapter 2 on Legislation).

➤ **Develop** a mission statement and define the surveillance program's goals and objectives. Determine what outcome measurements are desired by the program. For example, the program may want to describe the distribution of birth defects in their population, calculate rates and perform statistical analyses, and identify children who require services. Ideally, the development of goals, objectives, and outcome measurements will be done in collaboration with stakeholders and with internal and external advisory groups.

➤ **Define** the parameters of case definition for the surveillance program, including residency, pregnancy outcomes, eligible diagnoses, and age range. Define the minimum criteria for an eligible case report (see Chapter 3 on Case Definition).

➤ **Define** the method of case identification that will be used. Usually, a program will develop an infrastructure to support functions of active or passive case ascertainment. It is essential to document procedures, protocols, decision items, and methods of data collection (the program's surveillance approach). Records review and data collection procedures should be defined precisely.

➤ **Determine** the data variables needed to fulfill program goals and objectives. Define the minimum information that must be collected and address other information that would be beneficial to the surveillance program (see Chapter 4 on Data Variables).

➤ **Document** protocol and procedures regarding the privacy of the individual and the confidentiality of health information.

- ➢ **Design** forms for reporting, data collection, and abstracting that are adaptable to computer technology. This could include web-based reporting and forms that provide for easy data entry or scanning and that support abstracting medical records in the field (see Chapter 9 on Data Management and Security).

- ➢ **Develop** a database that has record linkage capability and that also functions as an information management system. The database should be flexible, adaptable, and able to accept electronic transfer of data files, web-based case reports, and case record abstracts from multiple sources. The database should support identification of all sources of information through which a diagnosis is identified or reported. It is also useful to be able to track and monitor medical records requests and perform other information management functions (see Chapter 9 on Data Management and Security).

## 6.3.2 Identify Data Sources

A key component in surveillance is identifying data sources for case ascertainment. A program needs to understand and evaluate the traits, characteristics, and operating procedures of all data sources. This is particularly important if there are potential sources of bias or underreporting associated with the way cases may be identified at a source.

The program should:

- ➢ **Identify** all potential data sources able to provide information that will help to fulfill the program's mission (e.g., hospitals, genetics and specialty clinics, cytogenetics laboratories, administrative data sets, vital records).

- ➢ **Determine** which data sources are included in any legislation mandating reporting and any additional sources for voluntary reporting. Consideration should be given to recommending legislative changes if program objectives change or are expanded, or if important data sources are omitted from mandatory reporting. For example, when adding prenatal diagnosis surveillance to program operations, it may be necessary to amend legislative language to include new data sources or facilities.

## 6.3.3 Obtain Knowledge about Individual Data Sources

For each reporting data source the program should:

- ➢ **Know** the data source's mission or goals.

- ➢ **Identify** professional or legal mandates governing operations of the data source that may affect access to, or quality of, data from that source.

- ➢ **Describe** the population served by the data source.

- ➢ **Chart** the flow of information that is relevant to the surveillance program through the unit and/or data source. This is a good way to determine how the information is collected originally and whether or not the information is forwarded to a central repository (e.g., centralized computer file, medical records department, administrative database).

- ➢ **Maintain** an up-to-date directory of names and contact information for relevant people at the data source (e.g., medical records personnel).

➢ **Utilize** multiple data sources. Surveillance programs should use multiple data sources, both for case identification and data collection. It is important for the surveillance program to realize that one source rarely fills all of a surveillance system's needs for case record accuracy, completeness, and timeliness.

➢ **Develop** record linkage procedures to facilitate matching all reports to the correct case record. This is especially important when programs utilize multiple data sources (see Chapter 9 on Data Management and Security).

## 6.3.4  Implement Data Quality Procedures

Surveillance programs should evaluate data for completeness, accuracy, timeliness, and comparability to other birth defects programs. At a minimum, programs should develop quality assurance procedures (manual and/or computerized) to identify potential issues in data quality. This includes accuracy, completeness, and timeliness. Additionally, programs should maintain documentation on data collection, data abstraction, and medical records review procedures. This will reduce the risk of multiple interpretations that lead to an inconsistent application of procedures and interpretation of medical information. For further details, see Chapter 7 on Data Quality Management.

## 6.3.5  Evaluate Surveillance Method and Analytical Capability

Surveillance programs should evaluate the surveillance approach and determine whether the surveillance system is meeting program objectives. Additionally, outcome measurements should be evaluated. NBDPN recommends the guidelines offered in the document *Updated Guidelines for Evaluating Public Health Surveillance Systems* (Centers for Disease Control and Prevention, 2001).

# 6.4 Birth Defects Surveillance – Approaches to Case Identification

Cases of birth defects are generally identified in one of two ways: through 'active case ascertainment' (i.e., staff conduct case finding) or through 'passive case ascertainment' (i.e., case reports are received by the program). While some surveillance systems use both kinds of ascertainment approaches for case identification, program activities are generally structured around one or the other approach.

Birth defects rates are directly related to the method of case identification and type of surveillance approach. Table 6.1 presents birth defects rates based on various surveillance approaches (Edmonds, 1997).

**Table 6.1  Birth Defects Rates by Surveillance Approach**

| Data Source | % of Babies Reported with Birth Defects |
|---|---|
| Birth Certificates in 1996 | 1.5 |
| Newborn Hospital Discharge Data (Florida) | 4.3-7.1 |
| Mandatory Hospital Reporting (New York) | 3.4 |
| Linked Data Sources (North Carolina) | 4.7 |
| Active Hospital Surveillance (Atlanta 1992-1996) | 2.6 |
| Physical Exam of Infants (Collaborative Perinatal Project) | 8.3 |

Although a physical examination of each infant provides the most complete assessment of birth defects among births, and therefore the highest prevalence, few programs can sustain this type of intensive case ascertainment. At the other extreme, the prevalence at birth of defects is clearly underreported when only birth certificates are used in case ascertainment. The NBDPN promotes case ascertainment approaches that provide a more complete description of birth defects prevalence in the US.

Whereas the previous section on general surveillance development (Section 6.3) provides a foundation for surveillance systems, the following two sections (Sections 6.5 and 6.6) discuss unique issues that arise in using either active or passive case ascertainment approaches in the identification of birth defect cases. We are indebted to Lynberg and Edmonds (1992) for much of the information in Sections 6.5 and 6.6.

# 6.5  Active Case Ascertainment

With active case ascertainment, cases of birth defects are identified at data sources by surveillance staff. The case-finding process includes identifying potential birth defects cases, reviewing and abstracting information from medical records, and conducting follow-up in order to complete abstracts or verify information. Programs take measures to ensure complete case ascertainment by using multiple data sources and multiple units within data sources. Case-finding activities may vary depending on the program's resources and objectives. A program's resources, as well as program goals and objectives, should be used to determine the intensity of case finding. Regardless of the case-finding methods used, active surveillance programs should provide detailed instructions on the case-finding process, document procedures for collecting information and completing case abstracts, nurture relationships between the program and its data sources, evaluate the quality and effectiveness of all steps in the case-finding process, and implement quality improvement methods.

In the sections below we discuss characteristics of active case ascertainment (Section 6.5.1), a recommended approach for active case ascertainment (Section 6.5.2), data quality issues in active case ascertainment (Section 6.5.3), evaluation (Section 6.5.4), and tips and hints for active case ascertainment (Section 6.5.5).

## 6.5.1  Characteristics of Active Case Ascertainment

➤ *Surveillance staff identify birth defects cases by visiting data sources.* Staff should follow a thorough and systematic set of investigative methods so that all potential birth defects cases are identified.

➤ *Surveillance staff are trained to find birth defects cases.* Staff learn how to find (or ***cull***) cases in hospitals, medical facilities, clinics, laboratories (e.g., cytogenetics laboratories, genetics clinics, prenatal diagnostic centers), and in medical records that relate to each potential case (e.g., prenatal, maternal delivery, newborn, infant, pediatric).

➤ *Staff are trained to gather information from information sources and medical records.* This includes following abstracting procedures and documentation guidelines. Staff are trained in birth defects coding and learn how to conduct follow-up.

➤ *Multiple information sources are used to obtain data.* All potential data sources should be part of the case-finding investigative process, and some are essential (e.g., birth hospitals, unit logs in birth hospitals). Surveillance systems should evaluate the effectiveness of case finding at each data source.

➤ *Case abstract forms are detailed and comprehensive and usually include a number of variables pertaining to the pregnancy, delivery, and outcome.* Information on the mother and infant is often collected in detail, including medical and prenatal care history, complications of pregnancy or delivery, reproductive history, physical examinations, postnatal procedures, and birth defects diagnosis.

➤ *Clinical reviewers, usually physicians, are trained to confirm, qualify, and evaluate the diagnostic information collected* by the surveillance abstractors.

➤ *Active case-finding surveillance should result in accurate and complete identification of birth defects cases.* The data are of high quality due to extensive staff training. The data collected are comprehensive and result in a detailed case abstract.

# 6.5.2 Approach to Active Case Ascertainment

Active surveillance is based on surveillance staff investigating data sources and finding potential birth defects cases. Although other activities may be part of the active surveillance approach, case finding is the primary task. There are various approaches to the case-finding process. Some programs have staff review all pertinent data sources and information reports, while others limit case finding to the most important information sources. Some use existing databases or lists of potential cases that are generated by the data source. Because case finding is labor intensive, most programs evaluate case-finding activities and determine ways to identify cases effectively and efficiently, yet still be relatively sure that case ascertainment is complete. It is necessary to take into account the legal or legislative issues that govern program activities.

Essential program activities for active case ascertainment include those listed below.

➢ **Identify** program objectives. It is important to develop or enhance the case-finding approach based on the purpose and objectives of the surveillance system. For example, if information is used to refer children to services, then the case-finding process should be designed to collect identifying and contact information early enough in the process to make the referral in a timely manner.

➢ **Develop** a flow chart of the case-finding process. Identify the data sources that are consistently used for case finding. At a minimum the program must conduct extensive case finding at birth and major pediatric hospitals. Within the data sources, the program should identify which units and departments will always be used. Important units and departments to consider are labor and delivery, nursery, surgery, and pathology (see list of data sources in Section 6.7). Some programs use the medical records department to generate a list of diagnoses (i.e., disease codes) from the disease index.

➢ **Define** the type of information to look for and collect during the case-finding process. Information gathered may be sketchy, incomplete, and general. This is especially true when gathering information from unit logs. The case-finding process may also include gathering information for the conditions of low birth weight, prematurity, and other conditions that may potentially lead to a case.

➢ **Define** the frequency of case-finding activities (i.e., visiting sources of information and completing abstracts). Frequency and consistency of case-finding activities affect the timeliness of the surveillance database. For example, if the program identifies a child who needs to be referred for services, it is usually important for the referral to be made in a timely manner. Timeliness can be measured by setting goals for the maximal length of time between birth and referral.

➢ **Conduct** case finding (culling). This is the systematic and ongoing process of identifying birth defects cases. Potential cases at the data source are found by surveillance staff through one or more procedures: (1) reviewing information at unit logs within a data source and creating a list of medical records to be pulled by the health information department within the data source; and/or (2) requesting a line listing of potential cases from the data source or unit, usually by identifying the cases by ICD codes (e.g., hospital index); or (3) reviewing the medical records for every delivery, termination, miscarriage, etc. occurring at the data source.

➢ **Conduct** medical records reviews. Potential cases of birth defects identified by the case-finding process are further investigated through medical records reviews. Requests for medical records are provided to staff of medical records departments at hospitals (or other sites), who pull the charts and make them available to surveillance system staff. Surveillance staff, who determine if the child or fetus meets the eligibility criteria for inclusion as a case, review the medical records. Multiple medical records may be reviewed during this process. These may include: maternal medical records during prenatal care, hospital admits during the pregnancy, and the delivery record. Medical records for a child include the newborn delivery record and any medical (hospital) records generated after the birth.

➢ **Abstract** information. As medical records are reviewed, surveillance staff abstract (record) the required information and record it on the case abstract form. Trained surveillance staff follow program guidelines and procedures for completing the data elements on the case abstract, confirming a diagnosis, and conducting follow-up to find cases at data sources and within units at data sources. Although a surveillance program develops its own set of abstracting guidelines and procedures, these should be based on established guidelines when available. In some programs, the abstractor assigns the disease code. In others, assigning the disease code occurs separately from abstracting.

➢ **Perform** a clinical review. Some programs have an expert in medical diagnosis issues review the case abstract after it is complete. The abstract is evaluated for incomplete data variables (i.e., data fields), accuracy of the medical information, and accuracy of the disease code assigned. Some clinical reviews result in the further classification of the case with a summary diagnosis, as an isolated or syndromic case, or other classification.

## 6.5.3  Data Quality Issues in Active Case Ascertainment

Active case finding requires surveillance staff to review and collect information from medical records. Staff are involved directly in verifying and confirming medical information and determining whether further follow-up or investigation is needed. In these programs, the burden of maintaining the quality of the database rests with the surveillance staff. It is essential to understand the challenges to data quality that occur in active surveillance and to implement strategies to identify and to correct them (see also Chapter 7 on Data Quality Management).

➢ *Field work (case finding, record review, abstracting) should be evaluated for accuracy, incomplete data variables, and consistency.* Desired outcome benchmarks in each of these areas should be identified and improvements implemented and tracked.

➢ *Data sources and individual units should be evaluated with respect to the staff resources expended and the results obtained.* Since case finding is labor intensive, programs should streamline and improve operations whenever possible. The value of the output of each unit or department utilized should be evaluated against the staff resources used. The program should determine whether unnecessary medical records are being reviewed and identify which non-anomaly ICD codes are most effective in identifying potential cases.

➢ *The surveillance database should be evaluated for timeliness.* This includes measuring how current the database is in relation to calculating disease rates. Although programs may collect information on individual birth defects cases over many months or years, they should set benchmarks for finalizing an individual case record or meeting a level of productivity by specified times.

## 6.5.4 Active Case Ascertainment Surveillance Evaluation

Evaluation of active ascertainment surveillance methods should occur at two levels. Both levels directly impact data quality and the program's ability to meet goals and objectives. One level targets case identification and data collection. Examples of areas that should be evaluated are:

➤ Completeness

➤ Accuracy

➤ Timeliness

➤ Measurability

Programs should develop outcome measurements that will improve data quality and are important to meet program needs and surveillance objectives.  See Chapter 7 on Data Quality Management for a more detailed discussion of this topic.

The other level focuses on the surveillance system itself. For a comprehensive approach to evaluating surveillance systems refer to CDC's *Updated Guidelines for Evaluating Surveillance Systems* (Centers for Disease Control and Prevention, 2001).

## 6.5.5 Tips and Hints in Active Case Ascertainment

➤ *Establish precise guidelines and criteria for data requests to data sources.* The process of active case ascertainment includes requesting information from data sources. Specific criteria or data variable parameters should be provided (e.g., ad hoc reports generated from an existing database, extracted information from databases).

➤ *Visit tertiary care (e.g., major pediatric site) hospitals first.* These sites usually have the most complete diagnostic information on a birth defects case. Surveillance staff can follow the case back to the birth hospital for the remaining information. Often a delivery that occurs in a rural hospital is transferred to the tertiary care facility.

➤ *Coordinate the schedule of site visits with the data sources* to minimize inconvenience for them.

➤ *Form relationships with staff in medical record departments (directors, coders, those coordinating release of information, record retrievers), birth registrar at the hospital, and hospital unit staff.* Discuss the purposes of the surveillance program with them and describe the work that surveillance staff perform at their sites.

➤ *Know key information technology or data processing staff at the data source.* These individuals often can access and retrieve specific pieces of information collected at the data source or within a component unit.

➤ *Use caution with ICD lists or an ICD disease index generated by medical records (or information technology) staff.* Hospital medical records coders are required to adhere to a set of federal guidelines when assigning a disease code to the medical record. Surveillance staff use a set of abstracting guidelines developed by the program (or NBDPN). Measure the benefit of using a disease code from an index against the output gained and resources used. For example, evaluate the results of a medical records review after using disease codes from an index.

➤ *Use laptops.* Design computer screens to assist in the case-finding process. Direct data entry during information gathering is more efficient, and likely more accurate, than recording information on paper forms and then entering it into the database.

➢ *Be conscious of HIPAA,* especially as this relates to the privacy and security rules that covered entities (i.e., health care facilities) are required to follow. Be knowledgeable about public health exemptions in HIPAA. Provide reassuring documentation to sites as appropriate (see Chapter 2 on Legislation).

# 6.6  Passive Case Ascertainment

With passive case ascertainment, case reports are submitted by data sources to the surveillance program. The level of interaction between programs and reporting sources varies, as do the methods of reporting. Some programs create birth defects case reporting forms and instruct reporting sources on how to complete them. Other programs merge or extract pertinent information into the surveillance program's database from a data source's existing database. Many use a combination of reporting methods to develop as complete an identification of birth defects cases as possible within the resources available. Regardless of the methods used, operating a surveillance system that receives case reports from data sources requires the program to identify and use multiple data reporting sources, provide detailed instructions to case reporting sources, nurture the relationship between the program and the reporting data source, and evaluate the quality of the case reports received.

In the sections below we discuss characteristics of passive case ascertainment (Section 6.6.1), a recommended approach for passive case ascertainment (Section 6.6.2), data quality issues in passive case ascertainment (Section 6.6.3), evaluation (Section 6.6.4), and tips and hints for passive case ascertainment (Section 6.6.5).

## 6.6.1  Characteristics of Passive Case Ascertainment

➢ *Birth defects cases are reported by data sources* to the surveillance program.

➢ *Medical information is received by the program as a case report* and is generally accepted as reported (i.e., the program does not confirm every case report for accuracy or comprehensiveness of diagnostic information).

➢ *The operational procedures used by each data source influence the accuracy, totality, definition, and timeliness of reported diagnoses.* This, in turn, influences the quality of the data in the surveillance program's database.

➢ *Information is usually reported from multiple reporting sources.* Data sources often serve different purposes for a program. Many can be used as major sources of clinical information (e.g., hospital reports, hospital discharge index, cytogenetics laboratories). Some are used as a source of demographic and statistical information (e.g., vital records). Others are used primarily for tracking or follow-up (e.g., genetics clinics, pathology and autopsy reports, specialty treatment clinics, and developmental centers). See the list of data sources in Section 6.7.

➢ *The database is developed to accommodate various reporting formats.* Information may be submitted in many ways and formats, including web-based reporting, electronic transfer and digital format, computerized reports, and hard copy reporting forms. Medical information may be reported in text format or in ICD code.

➢ *Record-matching procedures are used since data are collected from multiple sources and existing databases.* Case report information is extracted from administrative databases (e.g., hospital discharge data set, Medicaid data, vital records) and from existing databases within a facility (e.g., laboratories, specialty clinics, prenatal diagnostic centers).

# 6.6.2  Approach to Passive Case Ascertainment

Passive case ascertainment is based on case reports submitted to the program by data reporting sources. Reporting sources may include mandatory hospital reporting and physician reporting and/or administrative databases (e.g., hospital discharge data set, Medicaid data, vital records). Completeness in the surveillance system is improved by using multiple data sources, especially when data sources are selected to fill a gap in case identification (e.g., fetal death certificates, pathology and autopsy reports). Customized reporting forms may be used, or a program may elect to use other methods for receiving case reports. All legal or legislative issues that govern program operations must be taken into account.

Essential program activities for passive case ascertainment include those listed below.

➢ **Establish** the type and scope of passive case ascertainment that defines program operations, including whether surveillance includes fetal deaths. Some programs have limited disease reporting guidelines and a smaller set of data sources that are required to report. Some programs may have more liberal disease reporting guidelines but, due to limited resources, have to limit the scope of program operations. Generally, programs that use multiple data sources will have more complete case ascertainment than those that use only one or two data sources. If programs use the birth certificate as a data source for case reports, they should use another data source for case identification.

➢ **Identify** the case identification data sources. These include birth and major pediatric hospitals. If fetal death is an outcome that is ascertained, it is important to use the fetal death certificate, and possibly cytogenetics laboratories, as a source of case identification. See the list of data sources in Section 6.7.

➢ **Define** case reporting requirements precisely for each data source. This includes identifying the required or minimum data variables that should be reported. Some data sources will only report the required minimum data variables, while others, like an administrative database, may be able to furnish the program with additional pieces of information. Refer to Chapter 4 on Data Variables.

➢ **Develop** data reporting methods and procedures for each data source, including data format, timeliness, or reporting schedules. When possible, encourage electronic or web-based reporting. Data sources are usually more consistent in reporting when the burden of submitting the case report is minimized.

➢ **Develop** record linkage capability. It is important not only to accommodate multiple case reporting formats, but also to use the efficiencies of technology in processing case reports from administrative and existing databases and linking them to case records in the program's database.

➢ **Develop** procedures for abstracting information from medical records. This includes using the NBDPN Abstractor' Instructions (see Chapter 3 on Case Definition, Appendix 3.2), assigning disease codes, recording other pertinent information, and entering data into the database. Passive case ascertainment programs should review medical records as part of data quality evaluations. Additionally, medical records reviews are often conducted for other focused surveillance functions. For example, some programs that perform statistical monitoring regularly review medical records to confirm a diagnosis. Other times it is important for surveillance staff to review medical records to confirm a diagnosis during a community investigation or when investigating a suspected cluster.

# 6.6.3 Data Quality Issues in Passive Case Ascertainment

In passive case ascertainment, reporting sources submit case reports to the surveillance system. The reports are accepted without prior confirmation or verification of the information. Therefore, evaluations for quality must be conducted, especially regarding key program outcomes such as completeness, accuracy, and timeliness. Evaluations are often done by reviewing medical records and comparing results between the review and the reported diagnosis. A result of the evaluation process should include quality assurance procedures to identify future problems and methods to track improvement (see also Chapter 7 on Data Quality Management).

➢ *The quality of a reported diagnosis should be evaluated for accuracy and comprehensiveness.* Errors and differences in reporting will occur, resulting in underreporting, overreporting, and inaccurate reporting. By "rating" the quality of a reported diagnosis, data sources can also be evaluated. Results can be used to adjust quality control and assurance procedures and direct strategic programmatic decisions.

➢ *The surveillance database should be evaluated for timeliness.* This includes measuring how 'current' the database is in relation to the program's ability to calculate disease rates. Track timeliness of reporting per data source and identify reporting time lags. For example, watch reporting trends to identify whether some calendar months or quarters are problematic for some data sources. Evaluate the surveillance program's data processing procedures for time lags.

➢ *The disease coding classification system should be evaluated to identify weaknesses, limitations, and problematic codes.* This is especially important for data sources that report cases in ICD code format, which can happen with a data source such as an administrative or existing database. Additionally, although federal coding guidelines are used to direct a hospital or clinic medical records coder in assigning a disease code, the interpretation of medical documentation in the chart is often the reason for a particular code assignment. A good way for a surveillance program to identify potential code problems is to understand some of the conditions that may surface during the newborn time period. For example, a problematic code could be 748.0, choanal atresia or stenosis, since some newborns do experience difficulty in breathing in the first few hours of life. Additionally, situations that might cause a misuse of codes are low birth weight and prematurity (see Chapter 3 on Case Definition, Appendix 3.3). To gain experience in understanding these issues, medical records should be reviewed and results evaluated.

➢ *The surveillance database should be evaluated for fluctuations in counts and rates of specific diagnoses.* It is possible that an increase in a rate may be due to a change in procedure at a data source. Passive case ascertainment systems must understand that procedures and processes at the data source affect the quality of information in the surveillance database.

➢ *The surveillance program should develop benchmarks for desired outcome measurements and develop strategies for how to improve the outcome results.* For example, a critical data source that is consistently lagging in reporting might be the focus of a strategic plan to improve timeliness.

## 6.6.4  Passive Case Ascertainment Surveillance Evaluation

Evaluation of passive case ascertainment surveillance methods should occur at two levels. Both levels directly impact data quality and the program's ability to meet goals and objectives.

One level targets data reporting sources, case identification, and data collection. Examples of areas that should be evaluated are:

- ➢ Completeness
- ➢ Accuracy
- ➢ Timeliness
- ➢ Measurability

Programs should develop outcome measurements that will improve data quality and are important to meet program needs and surveillance objectives. See Chapter 7 on Data Quality Management for a more detailed discussion of this topic.

The other level focuses on the surveillance system itself. For a comprehensive approach to evaluating surveillance systems refer to CDC's *Updated Guidelines for Evaluating Surveillance Systems* (Centers for Disease Control and Prevention, 2001).

## 6.6.5  Tips and Hints in Passive Case Ascertainment

- ➢ *Use record linkage to link to the vital record early in the data collection process.* The vital records data source is excellent for establishing a unique case in the database and one that readily identifies the residency of the pregnancy outcome. Additionally, the birth and fetal death certificates fulfill many data collection variables for pregnancy outcome, maternal, and pregnancy information, as well as other statistical information (see Chapter 4 on Data Variables). See the detailed description of the vital records data source in Appendix 6.1.

- ➢ *Identify high-quality data sources that report a confirmed diagnosis.* A diagnosis from a high-quality source is an efficient way to improve the accuracy of the database. It also offsets the need to conduct a medical records review for quality evaluations for the specific diagnosis.

- ➢ *Ensure cooperation and compliance of data sources as critical factors in passive case ascertainment.* Ease the burden on data sources by encouraging electronic, computerized, and web-based reporting formats for submitting case reports. Offer technical assistance to sites. Many data sources already have the information the surveillance system needs in a database. It is usually easier to sustain consistent, timely, and compliant reporting using a computer program to extract information, rather than expecting staff at the data source to complete a case report.

- ➢ *Be flexible when discussing reporting methods and reporting requirements with a data source.* All data sources may not be able to provide all of the desired 'minimum' data fields easily. Evaluate the contribution, including efficiencies, the data source can make to the surveillance system and adjust reporting requirements accordingly. Identify which sources can usually be depended upon to report the majority of demographic information.

- ➢ *Be knowledgeable about the information flow through respective hospitals and sites.* Understand medical records content and documentation practices, including how the ICD code classification is used. Passive case ascertainment systems should be proactive in understanding where to go and who to contact to clarify issues when problems arise.

➢ *Consider conducting 'case finding' at a data source as an alternative to receiving the case report.* Although 'case finding' is not part of the passive surveillance approach, this method should be considered for data sources that may not have an efficient or reliable method of reporting (e.g., outpatient specialty clinics), that may not be able to report in a thorough manner (e.g., autopsy/pathology), or that are not required to report (i.e., voluntary reporting).

➢ *Communicate with data sources on how birth defects data are used.* Identify the users of the data (the customers) and some of the products produced using surveillance information. Reporting sources like to be recognized for the contributions they make (i.e., reporting cases) and appreciate knowing that the data they provide are used and serve important and valuable purposes.

➢ *Be active and creative in managing the quality of the database.* It is possible to develop program strategies that not only promote the efficiencies of passive case ascertainment but also improve the important outcome measurements of accuracy, completeness, and timeliness.

➢ *Be conscious of HIPAA,* especially as this relates to the privacy and security rules that covered entities (i.e., health care facilities) are required to follow. Be knowledgeable about the public health exemptions in HIPAA. Provide reassuring documentation to sites as appropriate (see Chapter 2 on Legislation).

# 6.7  Data and Case Identification Sources

Information on birth defects cases can be obtained from many sources, each of which has strengths and limitations. Rarely is one source able to provide all of the information necessary to complete a case record. Some, like birth and pediatric hospitals, are ideal for identifying a large number of cases. However, it is important not to overlook data sources like cytogenetics laboratories and specialty outpatient clinics, since they may identify cases previously unknown to a birth defects program. The challenge for birth defects surveillance programs is to evaluate and select data sources that meet the objectives of the program and that can be accessed using available resources. Most data sources can be useful for both active and passive case identification. Differences arise between the two case ascertainment approaches in how the information is gathered and collected. Some data sources are more conducive to active case ascertainment since the only way to access the information is to physically gather it. Some of the major data sources – including vital records, hospital discharge data, hospital unit logs, and genetics clinics – are described in further detail in Appendices 6.1, 6.2, 6.3, and 6.4.

**Vital Records** (see Appendix 6.1 for detailed description)

- ➢ Birth certificates
- ➢ Fetal death certificates
- ➢ Elective termination reports
- ➢ Death certificates

**Hospital Information** (see Appendix 6.2 for detailed description)

- ➢ Hospital discharge data set
- ➢ Hospital disease index

**Hospital Unit Logs, including** (see Appendix 6.3 for detailed description):

- ➢ Labor and delivery
- ➢ Surgery
- ➢ Nursery
- ➢ Neonatal Intensive Care Unit (NICU)

**Hospital Departments, including:**

- ➢ Pathology
  *Forensic (autopsy) pathology*
  *Surgical pathology*

- ➢ Surgery
  *Inpatient and outpatient/ambulatory*

> ➢ Specialty and outpatient clinics
> *Obstetrics*
> *Prenatal*
> *Perinatology*
> *Laboratory*
> *Pediatric medicine*

## Prenatal and Obstetrics Centers

➢ Birthing centers

➢ Obstetrics services

➢ Planned Parenthood, and other women's care clinics

➢ Prenatal diagnosis and high-level ultrasound referral sites

➢ Prenatal genetics counseling services

## Specialty Clinics

➢ Genetics (see Appendix 6.4 for detailed description)

➢ Oral-facial, craniofacial

➢ Meningomyelocele

➢ Cardiology

➢ Pulmonary/respiratory

➢ Musculo-skeletal

➢ Developmental and growth

➢ Audiology and speech

➢ Early intervention

➢ Neuro-developmental

➢ Ophthalmology

## Laboratories

➢ Cytogenetics

➢ Prenatal diagnosis

➢ Metabolic

## Physicians

➢ Pediatricians

➢ Obstetricians

➢ Specialists

**Health Care Professionals**

&#10148; Audiologists

&#10148; Developmental therapists

**Administrative Databases**

&#10148; Statewide hospital discharge data set (see Appendix 6.2 for detailed description)

&#10148; Medicaid data

&#10148; HMO data sets

**Other Sources of Information**

&#10148; University-based medical clinics

&#10148; Newborn hearing screening program

&#10148; Newborn genetic screening program

&#10148; Coroners and medical examiners

&#10148; Child fatality/mortality review programs

&#10148; Public health maternal and child health programs

*Public health clinics, including developmental clinics*

&#10148; School records

# 6.8  Sources of Information in a Data Source

In this section we discuss the various sources of information that may be available at a given data source. In Section 6.8.1 we provide a general introduction to the medical record, followed by a more detailed discussion of the various types of documentation within a medical record in Section 6.8.2. Other sources of information discussed include maternal delivery medical records (Section 6.8.3); prenatal medical records (Section 6.8.4); cytogenetic laboratory reports (Section 6.8.5); and autopsy, pathology, and laboratory reports (Section 6.8.6).

## 6.8.1  Medical Records

By law, all health care facilities are required to maintain some form of medical record on every patient for every service encounter that occurs in the facility. A medical record provides documentation on the course of treatment and progress of the patient at the facility for each admission or service encounter. The medical record may also include information from other health care facilities that may be pertinent to the treatment at that facility. For additional information on the professional practices and standards for medical records and other issues related to health information management, please consult the American Health Information Management Association (http://www.ahima.org).

Medical records differ according to type of health care facility. Medical records maintained by a private health care provider, genetic counseling facility, hospital, or cytogenetics laboratory are likely to differ in the documentation included in the record and how the records are organized. The medical records that birth defects program staff are most likely to work with are those maintained by hospitals, particularly birth and tertiary care pediatric hospitals, and specialty clinics.

The documentation required in a hospital medical record is usually defined by state legislation. Additionally, accreditation organizations maintain standards regarding required documentation (e.g., the Joint Commission on the Accreditation of Healthcare Organizations). Therefore, although medical records from different hospitals in a given state may be compiled and stored differently, the required content is the same. This is useful to know, especially if documentation appears to be deficient.

Since the early 1990s, the 'traditional' medical record has been undergoing change. Today, it is not unusual for the content of medical records to be a combination of hard copy, electronic, and computerized formats. Therefore, surveillance staff should be aware that the hard copy medical record that is traditionally stored and managed by hospital medical records departments may not appear to be 'complete' with respect to documentation. Some documentation may be in computer files or on electronic storage files (e.g., CD-ROM, microfiche, microfilm).

A hospital medical record is generated for every admission and service encounter, and each record follows the guidelines for standard documentation. Some exceptions to this rule may apply in certain pregnancy outcomes. Programs should consult with hospitals and delivery sites for their procedures for outcomes other than live birth. The following are offered as possible scenarios:

- ➢ *Live birth.* The infant and mother will each have individual medical records.

- ➢ *Live birth with neonatal demise shortly after birth.* The infant *may* have a newborn medical record. However, most useful information will be in the mother's delivery medical record (e.g., if autopsy or cytogenetics laboratory work is done, the results may be placed in the mother's chart).

➢ *Fetal death.* The fetus *may* have a medical record. However, most of the useful information will be in the mother's delivery medical record (e.g, if autopsy on cytogenics laboratory work is done, the results may be placed in the mother's chart).

➢ *Elective termination.* A medical record will be created only for the mother. Sometimes the admission at the hospital (or other site) will be as an outpatient.

There are other locations and places where births and other pregnancy outcomes can occur (e.g., in transit, in clinics, at home). Most, but not all, of these sites will generate a delivery medical record at least to fulfill federal and state requirements to complete a vital record. The depth of the information may be incomplete or inconclusive; therefore, additional investigative effort is usually required.

## 6.8.2  Type of Documentation in the Medical Record

Surveillance staff should be aware of the typical documents found in a medical record. This is true for staff conducting active case finding, as well as for staff conducting a medical records data quality audit for passive case ascertainment. Surveillance staff should consult with individual sites regarding records content requirements and how the documents are stored at the site (i.e., hard copy or computer file). The following are offered as examples:

➢ *Face sheet.* Contains demographic information, facility-specific information (e.g., medical record number, attending physicians, primary care provider, insurance).

➢ *History and physical.* Information is gathered and an exam is conducted at admission, at birth, and at various periods during the hospital stay (depending on the length of stay).

➢ *Discharge summary.* A document that is completed by a physician after a patient leaves the hospital. The summary pertains to a specific hospital stay and includes: admission diagnoses; pertinent medical history prior to the admission and problems, progress, and treatment during the hospital stay; a list of discharge diagnoses; and recommendations for follow-up, such as future visits to specialists and medications to be taken. At some hospitals a discharge summary may not be required for a very brief length of stay (e.g., less than 48 hours). Sometimes discharge summary information is recorded in the progress notes.

➢ *Consultations.* Specialists such as neurologists, geneticists, or cardiologists also see the patient and provide diagnostic clarification.

➢ *Progress notes.* Health care providers (e.g., physicians and nurses) document treatment and plans.

➢ *Diagnostic reports.* Any procedure, whether invasive or non-invasive, requires documentation. This includes: diagnostic tests, laboratory analysis, surgery, cytogenetics, pathology, and autopsy. Sometimes, the final report will not be in the medical record (e.g., it may be in an electronic file or on file in a department of the respective site). Some results will be referred to in the discharge summary, progress notes, or consultation, while others may not be completed for several weeks (e.g., autopsy cytogenetics).

# 6.8.3  Maternal Delivery Medical Record

In addition to standard documentation required in hospital medical records, the mother's delivery medical record contains unique pieces of information that are important for case ascertainment.

➢ *Labor and delivery summary.* Many hospitals use a standardized form to record important aspects of the outcome (e.g., time, weight, pregnancy risk factors).

➢ *Prenatal medical records.* Although the private obstetrician maintains these, some documents may be inserted in the mother's delivery record (or located in other places in the mother's hospital medical record). These include copies of the course of pregnancy management and results of prenatal diagnostic procedures, such as ultrasounds, amniocentesis, and cytogenetics analyses, particularly if a birth defect is detected prenatally.

➢ *Pathology and laboratory reports.* Pathological analysis is important in the case of fetal demise. Laboratory reports are important when there are suspected infectious disease or toxicology concerns in the mother. For example, there may be concerns about an exposure that could be passed along to the infant through breast milk.

➢ *Autopsy.* If an autopsy is performed on a fetal demise or neonatal death, the report is often inserted in the mother's medical record or may need to be tracked to the appropriate department.

# 6.8.4  Prenatal Medical Record

Currently, prenatal care may result in a woman having multiple medical records generated over the course of the pregnancy.

➢ *Obstetrician's prenatal care medical record.* This record contains documentation of how the pregnancy is managed. The content of this medical record is very similar to a hospital-based medical record; thus, it is important for birth defects surveillance. Sometimes the prenatal care medical record is inserted into the maternal delivery medical record.

- *Prenatal care forms.* These are often in a standardized format and facilitate complete recording of information (e.g., laboratory work, family history, risk factors, genetic screens, and tests).

- *Flow charts of care.* Prenatal visits, care and treatment, and patient discussions are documented, although often written by hand.

- *Diagnostic tests.* The record may contain diagnostic tests, laboratory results, genetic counseling reports, consultations, and referrals to diagnostic centers.

Prenatal diagnosis is growing in importance for birth defects surveillance. There is a long history of chromosomal diagnoses that are detected prenatally through the procedures of amniocentesis and chorionic villus sampling. Many more diagnoses can now be detected through the use of high-level ultrasound. Technology and diagnostic methods will continue to advance in the area of prenatal diagnosis.

➢ *Referral prenatal diagnostics and diagnosticians.* Referral centers specialize in high-risk pregnancy and have high-level diagnostic capabilities. Depending on the course of a high-risk pregnancy, the referral physician (diagnostician) may assume primary management of the pregnancy and may attend the delivery. However, usually, the referral diagnostic site and diagnostician do not follow the patient throughout the pregnancy. Medical records generated at the referral diagnostic sites may contain pertinent information from the primary obstetrician's

office, including demographic information, index prenatal care history, medical history, risk factors, and reasons for referral. They also contain unique information for case ascertainment. Sometimes the referral prenatal diagnostics are inserted into the obstetrician's prenatal care medical record.

- *Diagnostic and laboratory results.* The medical record includes the results and discussion of the results.

- *Genetic counseling.* Documentation in this report includes significant family history, discussion of prenatal diagnosis, and discussion of prognosis.

## 6.8.5  Cytogenetic Laboratory Reports

Cytogenetic analysis may be performed at the hospital (in-house) or at freestanding laboratories. Programs are encouraged to use cytogenetic laboratories as data sources that consistently report cases. It is important for birth defects program staff to have some knowledge of basic genetics and the chromosomal terminology they are likely to encounter in medical records. For additional information on cytogenetics terminology (and corresponding abbreviations and symbols) refer to the reference manual, *International System for Human Cytogenetic Nomenclature (ISCN)* (Mitelman, 1995).

The report on cytogenetic findings is created by the lab that did the analysis. The report usually identifies:

➢ Name of patient

➢ Date of birth

➢ Referring facility and/or physician

➢ Reason for referral (or suspected diagnosis)

➢ Result/karyotype

➢ Narrative regarding the analysis

Rarely does the report provide an address for the patient. This presents a challenge for a surveillance program that regularly receives case reports directly from the cytogenetic laboratory, since the laboratory may also perform analyses for patients from several states. Surveillance programs should develop quality control procedures that address this and other challenging issues when working with cytogenetic laboratories. One possible approach is to develop a list of the locations of the referring facilities and/or physicians.

The original report of the result of a cytogenetic analysis (or other test) is the property of the laboratory that performed the analysis. A copy of the report may or may not be sent to the referring facility or physician (or included in the referring facility's medical record). The results may be communicated orally or referenced in a medical record. The surveillance program should develop abstracting procedures for accepting a referenced cytogenetics analysis and for determining when it is necessary to locate the initial source of medical information.

There is a growing trend for hospitals to use out-of-state laboratories. Surveillance programs should investigate the feasibility, including legal authority, of using and contacting out-of-state laboratories.

# 6.8.6 Autopsy, Pathology, and Laboratory Reports

Pathology laboratories are usually associated with hospitals, while autopsies may be performed in selected hospitals or through coroner's offices. Autopsy and pathology reports are usually placed in the patient's medical record, but the autopsy report may be completed long after death (some states have 45- to 60-day time frames for completion of autopsies). Therefore, the autopsy report may not be filed with the admission medical record; it may be in the outpatient or 'other' section of the record. It is important to note that there are two completion status categories for autopsy findings or reports: provisional and final. Surveillance staff should place the highest level of diagnostic certainty on the final report.

Anatomical pathology laboratories usually produce high-quality case reports due to the exacting nature of the procedures performed during autopsy. An important exception to this is when the specimen is destroyed, macerated, or otherwise compromised, as is the case with many fetal deaths. When this happens, the autopsy and tissue analysis may be of limited value for birth defects case identification. Still, the autopsy report or tissue analysis will often provide the most definitive information on structural defects. Additionally, the type of tissue sample can provide useful information regarding the time frame of the pregnancy. Therefore, it is important to track and examine these reports.

Autopsy and pathology laboratories may have information management systems, manual or computerized, specific to the laboratory. Diagnostic information is usually accessible since these laboratories catalog their findings for forensic investigations, historical and legal archives, case studies, and medical board reviews.

Surveillance programs should understand that there might be varying degrees of quality in autopsy reports. Much depends on the expertise of a given pathologist or coroner, the majority of whom are not fetal and pediatric anatomical pathologists, the experts in this area. In some states these pathologists, and the hospitals or sites where they work, act as referral centers for specialized autopsies. Programs should consult with the respective pathologists and sites to better understand referral patterns in a given state and to evaluate the level of expertise available in this specialized area.

# 6.9  Infant Risk Factors in Case Identification

A condition that affects an individual's chance of having a particular outcome is called a ***risk factor***. Various maternal and pregnancy exposures and conditions have been associated with an increased risk for birth defects. Birth defects programs can use these risk factors to identify potential cases, either through including their ICD-9 codes on the discharge lists obtained from medical records departments, through reviewing logs for any entries citing these risk factors in addition to birth defects, or through identifying vital records with particular birth weights, etc.

However, even though certain factors are associated with increased birth defects risk, the majority of infants and fetuses with these risk factors will not have a birth defect. Thus, a large number of records will be reviewed that do not turn out to be cases.

Moreover, the list of risk factors that may be used as case-finding sources can become very large. It is possible that a large portion of the potential inclusion population will have at least one of the risk factors used as a case-finding source. Most risk factors only result in a small to moderate increase in birth defect risk, so the majority of records reviewed on this basis will not yield eligible cases. Such risk factor lists are developed from experience, logic, and research. Programs that use risk factors should evaluate the yield in their case identification approach and determine whether using risk factors as case-finding sources is useful to the program over time.

In the short term, the use of risk factors as screens for identifying potential cases of birth defects may be a valuable effort when the program is involved in a concentrated focus on a specific outcome, exposure, medical condition, or cluster investigation.

Surveillance staff may encounter various postnatal complications during the review of data sources and units. This information is most likely found in the infant's medical record, and often in progress summaries. In the situation of a fetal demise or stillbirth, the information is usually found in the maternal delivery chart.

The list below provides some examples of risk factors that may be useful as case-finding sources. Surveillance staff should use pediatric references to become familiar with newborn conditions and evaluate which conditions are appropriate to use for case finding. Passive case ascertainment programs should also evaluate the effectiveness of using risk factors. The majority of the items listed below are identified in data fields on the vital record (birth and fetal death, death certificates) and easily accessible to both active and passive case ascertainment surveillance systems.

Examples of infant risk factors include:

➢ Infants who weigh less than 2,500 grams (5 lbs, 8 oz) or are < 36 weeks gestational age

➢ Fetal and neonatal deaths

➢ Infants with a history of asphyxia at birth (Apgar score at 5 minutes less than 7)

➢ Infants admitted to neonatal intensive care or special care nurseries

➢ Multiple births

➢ Infants with respiratory distress

➢ Infants with heart murmurs

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*Appendix 6.1*

*Data Source Described in Detail – Vital Records*

# Appendix 6.1
# Data Source Described in Detail – Vital Records

**Source or Site**

> ➢ Birth certificates
>
> ➢ Fetal death certificates
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> ➢ Elective termination reports
>
> ➢ Death certificates

Birth, death, and fetal death certificates provide a standardized way of reporting vital events that occur in a politically defined unit, a state. Vital records include facts about an individual and the specific circumstances regarding the reported event. Vital records are particularly important in that they fulfill two significant functions: they provide a mechanism for registering the occurrence of vital events, and they provide a mechanism for collecting demographic, social, and health information regarding the person in a standardized way. Integral to these functions is the fact that they are population based.

**Legal or Professional Mandates**

Federal law mandates birth and death registration. The lead federal agency is the National Center for Health Statistics (NCHS), Centers for Disease Control and Prevention (CDC). NCHS maintains the national birth and death registration system and is the recipient of vital records data from the states and territories. Recording births and deaths is the responsibility of the individual states and territories. The procedures and regulations regarding the reporting of these vital events are established by the individual states and territories. NCHS provides guidelines and recommendations for standardization of the information collected by birth and death certificates by promulgating standard certificates. Although federal law does not mandate the reporting of fetal deaths, there is an NCHS-recommended standard fetal death certificate. See http://www.cdc.gov/nchs for further information.

**Mission or Objective**

Provides a population-based statistical database of all births and deaths that occur in the United States.

**Scope or Breadth**

The birth, death, and fetal death certificates provide for registration of a defined vital event at a point in time. There are established criteria for what constitutes a live birth, but there is evidence to suggest that those criteria are not always followed. Registration of fetal deaths is usually defined on the basis of gestational age, with $\geq 20$ weeks as the cut-off used by most states. Some states require the reporting of all fetal deaths, regardless of gestational age, and there is recognized underreporting of early fetal deaths.

## Operational Structure

> *Data.* NCHS recommends standard data elements on birth and death (and fetal death) certificates. States are required to complete a minimum data set for national reporting and may add other data elements to their certificates. The birth certificate is usually revised and updated every decade. In 2003, the final drafts of a new version of the certificate are being reviewed. Please refer to http://www.cdc.gov/nchs for further information.

> *Certification.* State statutes, regulations, and procedures stipulate who is responsible for certifying a live birth, death, or fetal death. The designated person is required to certify date, time, and place of birth/death as well as other priority areas on the certificate. The completion of death certificates has additional protocols, procedures, and instructions because of the many circumstances that may surround a death.

> *Filing the certificate.* State statutes, regulations, and procedures stipulate time requirements for filing. Although the timing varies among states, the certificate is usually filed with the state registrar's office within 5 to 10 days of the event. Many states now have methods for entering and filing birth certificates electronically. The timing for filing a fetal death certificate depends on state guidelines. Although filing a death certificate is required within a specified time period, it may not be complete at filing, as some data elements may be missing due to autopsy, coroner investigation, or other legal proceeding. These data may or may not be added subsequently and the certificate revised.

> *Unique identification of an individual event.* Each state has a numbering system that uniquely identifies the respective event.

> *Storing the information.* Most states have a centralized database specifically designed to collect, amend, transmit, retrieve, sort, print, and analyze vital records information.

> *Reciprocity.* Agreements with bordering states ensure reporting of life events occurring in neighboring states to the state of residence.

## Types of Information Collected

> NCHS and other interested parties have developed a set of standardized data elements or **minimum data variables** that are required to be reported, as well as a set of **recommended data variables** and recommended standard certificates. Of importance is the unique identifying information per person, per event.

> The birth certificate and fetal death certificate are each divided into two sections: legal and statistical. The **legal section** contains the unique identifying information about the person, date, time, place, and type of life event. It is this portion of the certificate that registers the vital event. The information in the legal section is certified, and this is the part of the certificate that is issued to individuals when proof of the life event is required. The **statistical section** – labeled "Information for medical and health purposes only" – contains demographic, prenatal care, pregnancy risk factors, and medical conditions of the mother and of the newborn, including congenital anomalies. The statistical part is not released to the public, and many states do not keep the statistical part attached to the legal certificate. The statistical information is usually data entered and maintained in a database.

> The death certificate is a certified legal document, and it is available to authorized individuals in its entirety.

**Accessibility and Retrievability**

States transmit vital records information to NCHS electronically. State laws and regulations stipulate how the information is made available for other users at the state level. Due to the confidentiality of the information, states protect the medical and health information on vital records from unwarranted or indiscriminate disclosure. Most states have legal safeguards in place to further protect the information.

 ➢ The information contained in the birth, death, and fetal death master index computer file is usually available to authorized public health programs. Sometimes confidentiality or security agreements are required.

 ➢ Many states copy the legal sections of the hard copy certificate into a permanent electronic storage format (e.g., microfiche, film, CD-ROM). The storage format is cataloged for easy information retrieval.

**Strengths as a Data Source**

 ➢ *Timeliness.* Electronic filing allows information to be available to users as soon as the reports are filed in the state database. This may be as early as 30 days after the event.

 ➢ *Population base.* Provides statistical and denominator data.

 ➢ *Unique identification.* States assign a unique ID to each person, per vital event.

 ➢ *Legality of case report.* State laws require that some information must be certified for all births and deaths. Additional attention to legal procedures is required for death registration.

 ➢ *Comprehensiveness as a data source.* Over 97 percent of all births occur in a hospital or birthing facility. Out-of-hospital births are also registered because of the necessity for a child to have a birth certificate. There may be some underreporting of early infant deaths, and there is marked underreporting of fetal deaths at early gestational ages.

 ➢ *Existing data set and one that is accessible over time.* There is historical depth to vital records, but there have been major changes in format, content, and coding over time.

 ➢ *Record linkage.* Useful in combination with other data for building the case record. The use of unique identifying information permits matching and linking with other data sources. Many states routinely link vital records to each other, for example a death certificate with the birth certificate, providing a linked birth-infant death file.

 ➢ *Risk factor screening tool.* Some data elements can be used to identify potential birth defects cases. Examples include: low birth weight, prematurity, low Apgar scores, neonatal death, multiple births.

 ➢ *Intervention.* The availability of information in a timely manner is conducive to rapid intervention or investigation.

**Weaknesses as a Data Source**

 ➢ *Data quality.* Much of the medical information on the certificate has been shown not to be reliable.

 ➢ *Case ascertainment.* The birth certificate has been shown to underreport birth defects. As shown in Section 6.4, rates from this source are 1.5 percent, compared to 3 to 4 percent for hospital reporting and from using linked data sources.

**Liaisons and Partnerships**

➤ *Vital records/registrar's office.* These are staff that are involved in managing the activities involved in filing the certificate. These staff often go to hospitals to train personnel in the procedures and methods of filling out the certificate. Other activities include amending a certificate, maintaining the centralized database, and cross-referencing other vital record certificates.

➤ *Hospital.* These are staff that are involved in providing information for completing the certificate. Includes medical records services, neonatal nursing, labor and delivery unit staff.

**Hints and Tips**

➤ *Neonatal and infant death.* A death certificate is issued upon death for any infant who was live born, regardless of duration of the pregnancy. These individuals will have a birth and a death certificate. There is no distinction in death certificates for 'neonatal' or 'infant' deaths. Many vital records divisions cross reference the birth and death certificate numbers to make sure that a birth certificate is issued if a neonatal or infant death is reported. Sometimes, the facility will overlook filing a birth certificate for an early neonatal death. Sometimes a fetal death certificate is filed as well as a birth certificate and/or a death certificate. In these situations further investigation should occur to determine the actual vital status at birth.

➤ *The timing for filing birth and death certificates is similar.* However, often the birth certificate is processed by vital records more quickly since many hospitals use the electronic birth certificate. It is important for birth defects programs to be aware of these timing issues if they refer children to services, especially if they refer children based on low birth weight, prematurity, and other severe conditions. Regardless of how quickly a case report is sent to the surveillance program, it is a wise practice to allow a period of time to elapse before referring a child with severe conditions. A time period to consider before referring a child to services is 60 to 90 days past the date of birth.

➤ *Fetal death certificate.* This certificate is usually issued for any pregnancy that results in a non-live outcome at the end of a pregnancy that is $\geq$ 20 weeks gestational age. What constitutes 'live' is subject to legal definition, and most states have clear guidelines in state statutes for what is considered a 'live birth'. Some states accept any sign of life (e.g., a pulse), regardless of the intent for the delivery (e.g., elective termination). Surveillance systems need to understand the definition of 'live birth' in their state. There may be instances when an Apgar score is a very low number (e.g., 1) at the first minute, and 0 for the fifth minute. Some states might count this as a live birth or a termination, depending on the age of the fetus and intent of the delivery. Some states have guidelines that exclude filing a fetal death certificate if the intent of the pregnancy delivery is for a termination, regardless of the gestational age.

➤ *Termination reports.* Some states collect statistical information on terminations. Often there is no identifying information; however, a birth defect may be listed as a reason for the termination. In most instances these reports do not have sufficient identifying information to link to an individual. Additionally, although some states require the filing of these reports, compliance is notably poor, such that there is an underreporting of these events and conditions.

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*Appendix 6.2*

*Data Source Described in Detail – Hospital Data Sets*

# Appendix 6.2
# Data Source Described in Detail – Hospital Data Sets

**Source or Site**

 ➢ Hospital discharge data set

 ➢ Hospital admissions reporting system

 ➢ Hospital disease index

Discharge information is collected by the data source in a standardized format on individuals admitted for hospital-based services. This usually includes inpatient stays and outpatient surgery but may also include services performed in outpatient hospital clinics and emergency rooms.

**Legal or Professional Mandates**

 ➢ *Federal law.* The Health Information Portability and Accountability Act of 1996 (HIPAA) legislation defines electronic health care transactions, health information privacy and security standards, electronic signature codes, transaction standards and code sets, and unique health identifiers.

 ➢ *Other professional mandates* dovetail with federal requirements (e.g., Joint Commission on the Accreditation of Healthcare Organizations, American Hospital Association).

**Mission or Objectives**

Discharge data are collected for a wide range of possible uses. These include population-mix studies, market share analysis, hospital charges comparisons, length-of-stay studies, disease-specific and clinical information-specific case volumes, health care delivery access analysis, and crude and severity-adjusted death rate analysis. Discharge data are also used indirectly for financial analysis and billing.

**Scope or Breadth**

These data result from ongoing data collection and include all inpatient encounters. Some hospital data sets may also include outpatient encounters. The age of population served is defined by the mission of the site (e.g., a children's hospital may serve patients up to age 20 years). A discharge data set may consist of information from one hospital or may be a large statewide discharge data set of all hospitals. A record is created for each defined admission for hospital service. Discharge data sets are defined by a period of time (e.g., year) and are maintained so that they can be accessed over time.

**Operational Structure**

Information for the data set is collected from many places in the hospital, incorporated into the individual's medical record, and compiled in a standardized format. Health information management or medical records departments are responsible for processing the information that results in the data record for each patient encounter and in ensuring that the medical record contains the required documentation (content).

**Type of Information Collected**

Information included in this type of data set usually does not include patient names or Social Security numbers. The data elements collected, however, can lead one to a specific medical record. These data sets usually include: hospital identifier, patient medical record number, admission and discharge dates, patient type, patient date of birth, patient gender, patient's residential location (e.g., zip code, county), insurance source, charges, physician type, diagnosis and procedure codes in ICD format, and length of stay. Other information may be collected depending on the objectives of the data set.

**Accessibility and Retrievability**

Hospital discharge data sets are computerized and are used to generate routine reports and to respond to ad hoc queries. Some hospitals submit their discharge data to a larger organization that collects data from each hospital and compiles the information into a single statewide hospital discharge data set.

**Strengths as a Data Source/Site**

> *Existing database.* Data are easily accessible, retrievable, and available in a computerized format.

> *Specific information.* Specific data fields can be identified and extracted from the data base.

> *Cross-referencing.* Available data fields provide information that can be used to locate the medical record.

> *Disease classification system.* Information on discharge diagnoses and procedures is collected in a coded and standardized format, currently ICD-9-CM.

> *Timeliness.* Data are usually available rapidly, within 6 months of discharge. Internet technology has increased accessibility and improved timeliness of data from this source for some states.

> *Consistency of the data set.* Data fields are filled in as required for billing and for federal reimbursements.

> *Follow-up.* Hospitals have unique medical record numbers for patients, facilitating tracking and monitoring of cases.

> *Screening tool.* Specific data fields, especially ICD-9-CM disease and procedure codes, can be selected for further investigation.

**Weakness as a Data Source/Site**

> *Discharge set bias.* The discharge data set is an administrative database. Information is collected and compiled using procedures that suit a particular health facility or meet other legal requirements. It is a services-, planning-, and financial-based data set.

> *Population base.* The service area and patient population for most hospitals are not well defined. Therefore, the relationship of the hospital's patients to a larger group of persons is difficult to quantify.

> *Disease classification system.* Some disease categories and codes for birth defects are not specific and are limited in scope.

> *Accuracy and clarity of diagnosis.* Federal and professional standards are used to govern interpreting medical record documentation, which includes identifying a diagnosis and assigning a representative disease code. Suspected and rule-out conditions may be coded as a final

diagnosis at discharge, leading to overreporting. A diagnosis may not be recorded for many reasons. Underreporting may occur if not all of the diagnoses documented in the patient's medical record are coded.

➢ *Personal identifiers.* Externally recognizable personal identifiers usually are not available. Data elements can be used to locate medical records. Some states have adopted legislation to permit the reporting of identifying information directly in the discharge data set for specific reportable conditions (e.g., Colorado adopted regulations to permit named reporting from hospital discharge data).

➢ *Maternal information.* Information on the mother is not recorded on the discharge data record for a newborn infant or child.

➢ *No medical record is generated.* In some circumstances a medical record is not created. For stillbirths and even some neonatal deaths, a medical record may not be created for the infant. Information pertaining to the delivery outcome, including autopsy and laboratory reports, will be in the mother's delivery medical record. However, the mother's chart cannot be coded to reflect an infant's medical conditions. Therefore, in these circumstances a birth defect diagnosis will be missed. Surveillance staff should use other data sources, such as the vital record, to identify a case where a medical record might not be created.

**Liaisons and Partnerships**

➢ *Data processing unit.* Hospital staff in a data processing unit manage the computerized information that is collected from various departments in the hospital. These persons can assist surveillance staff by accessing birth defects information that is stored in computer format.

➢ *State hospital associations.* Some state hospital associations may serve the function of producing the statewide hospital discharge data set. They have a vested interest in providing customer service to a hospital by compiling aggregate statewide hospital data. Often these associations are also actively involved with the major users of the discharge data set (e.g., health departments, epidemiology programs, health planners).

➢ *Health information management and medical records departments.* The hospital's medical records staff are responsible for managing the information contained within a medical record. In addition to assembling the medical record and ensuring that it contains the required documentation, skilled personnel – coders – assign the disease classification codes and abstract pertinent information for administrative purposes (e.g., billing and the discharge data set). Since surveillance staff often use the disease classification codes to identify cases, it is helpful to maintain open communication with medical records departments regarding questions about hospital coding rules and other issues that might affect data quality.

**Additional Comments**

The hospital discharge data set is facing significant changes due to evolving federal regulations, including HIPAA and the conversion of the disease classification from the ICD-9-CM system to ICD-10-CM. HIPAA requirements address electronic transmission of data, standard data elements, and privacy and security issues. ICD-10 is a larger and more complex disease classification system, one that will affect the general taxonomy used for coding purposes.

# References

American Hospital Association (AHA). http://www.hospitalconnect.com/

Health Information Portability and Accountability Act of 1996.

Joint Commission on Accreditation of Healthcare Organizations (JCAHO). http://www.jcaho.org/

*Appendix 6.3*

*Data Source Described in Detail – Hospital and Patient Services Logs*

# Appendix 6.3
## Data Source Described in Detail – Hospital and Patient Services Logs

**Source or Site**

- ➢ Hospital unit logs
- ➢ Patient services logs (in non-hospital settings)

Hospital units operate within a hospital or clinic and serve specific operational functions. Traditional units relevant to birth defects case ascertainment include Neonatal Intensive Care, Critical Cardiac Care, Labor and Delivery, and the Newborn Nursery. In some hospitals, units are their own departments, like Pathology and Surgery. A ***unit log*** is the documentation that provides information in general terms on the patients who used (or were admitted to) the unit.

**Legal or Professional Mandates**

- ➢ *Legal – state statute*. Hospital-based unit logs are operated in accordance with hospital licensing and accreditation.
- ➢ *Legal – state statute*. Non-hospital-based unit logs (e.g., birthing centers, prenatal diagnosis referral centers, genetics clinics), are usually operated in accordance with licensing guidelines.

**Mission or Objective**

Determined by site. Logs are used to record specific events or health system encounters in a particular hospital department or facility setting. Logs may also account for equipment use. The log represents an inventory of events or activities.

**Scope or Breadth**

Logs are point-in-time accounts of events. The unit log accounts for each entry or use of services into the specific area. Most logs identify an entrance time, and an exit time, as well as other information specific to unit requirements.

**Operational Structure**

Determined by site. Logs are designed to be read easily and to provide sufficient information to establish why the patient was in the unit or department.

**Type of Information Collected**

Determined by the site. Generally, logs are used by surveillance programs as a case identification screening tool. Most logs provide enough cross-referencing information to support follow-through or tracking. This includes name, date of birth, medical record or other identification number, and current date and time. Additionally, information is collected specific to the purpose of the encounter. Examples include:

- *Labor and delivery log.* Prenatal information, maternal risk issues, prenatal diagnosis, and event or outcome measurements.

- *Neonatal Intensive Care Unit (NICU) log.* Event/outcome measurements, perinatal medical issues, diagnosis, other risk factors.

- *Surgery log.* Preoperative diagnosis, possible risk factors.

- *Prenatal diagnostic center log.* Prenatal information, referring physician, referring diagnosis, procedure, medical risk factors.

## Accessibility and Retrievability

Logs are used as management tools within individual facility units. Therefore, information is gathered for and used by the unit and, possibly, by the facility. While some information may be collected and entered into a database, most logs consist of paper copy record books or reports.

## Strengths as a Data Source

- *Timeliness.* Information is recorded in real time, as events occur. Rapid identification of potential cases is possible.

- *Consistency in recording information.* The population base is well defined for each particular unit since each service encounter is recorded. For example, if a surgical procedure was performed at the site, a surgical log will record the episode.

- *Case identification screening tool.* Generally, enough information is recorded so that surveillance staff can identify potential cases for further investigation.

## Weaknesses as a Data Source

- *Effort to retrieve the information.* Generally, logs are kept in hard copy format and are based on a handwritten recording of events. Review of the information can be effort intensive.

- *Accuracy and clarity of clinical information.* Information recorded may be inaccurate or incomplete with respect to diagnoses or medical conditions. For example, a prenatal ultrasound log may state 'referred for cardiac irregularity'.

- *Documentation in the log.* Information recorded on a log may be of limited use for case identification. Sites establish criteria for log documentation to meet internal or ward management objectives, not for disease coding. As such, the information is most relevant for immediate patient management rather than as a tool in medical diagnosis and treatment.

- *Different logs within the data source* may provide conflicting information on the same patient. Surveillance staff should develop management tools to keep track of information recorded from different logs.

## Liaisons and Partnerships

- *Unit staff.* These persons are usually front-line staff who work in the unit and have a use for the information that is recorded.

➢ *Office staff*. These are the persons at the unit who are usually responsible for compiling statistics for the unit and who monitor occupancy. They may be able to assist the surveillance staff in identifying efficient ways to access log information. For example, they may be able to generate a computer listing of the log or provide a photocopy of the log sheet.

**Issues to Consider**

*Surveillance program time and efficiency issues*. Unit logs usually require surveillance staff to spend time identifying potential cases on the log and following up by reviewing medical records. Case identification screening criteria and the quality of information included in a log are significant factors to consider when evaluating the amount of time spent on finding cases using this source. Inefficiencies result when follow-up medical records reviews result in too many non-cases. Time and effort evaluations should be conducted for the case identification processes involved in using unit logs.

Unit logs serve as a management tool for individual components of a facility. Therefore, a potential birth defects case may show up on multiple logs. It is useful to compare the information recorded at each unit within the data source and to develop a surveillance management tool that tracks case-finding activity. Such a tool will minimize staff time spent requesting and reviewing a medical record multiple times.

**References**

None.

*Appendix 6.4*

*Data Source Described in Detail – Genetic Services*

# Appendix 6.4
## Data Source Described in Detail – Genetic Services

**Source or Site**

> ➢ Regional/state genetics networks
>
> ➢ Hospital-based genetics clinics
>
> ➢ University-based genetics clinics
>
> ➢ Provider-based genetics clinics

Geneticists and dysmorphologists are skilled at evaluating a constellation of findings, providing differential diagnoses, and determining the definite medical condition. They use diagnostic procedures such as chromosomal analysis and genetic testing, as well as drawing from their personal experiences and extensive literature in evaluating a patient.

The information from this data source is of high quality.

**Legal or Professional Mandates**

> ➢ *Legal.* State statutes for hospital-based clinics. These are operated in accordance with hospital licensing and accreditation.
>
> ➢ *Legal contract.* Specified in individual contracts or collaborative agreements.
>
> ➢ *Professional.* Certification and professional credential as required.

**Mission or Objective**

Genetic diagnostic and counseling services, therapeutic management of genetic diseases.

**Scope or Breadth**

Clinics may include prenatal, pediatric, and/or general population. Some may be specialized by disease category (e.g., Down syndrome, cystic fibrosis). Some providers include diagnostic and research laboratories, clinical research centers, and off-site clinics.

**Operational Structure**

Genetics clinics may be set up as a referral site (i.e., to provide a diagnosis back to the referring physician), for services (i.e., for ongoing treatment and consultation), or for research or study (i.e., database).

**Type of Information Collected**

Depends on the focus of the encounter (i.e., prenatal, pediatric, and counseling). As a rule, genetics clinics collect a core set of information for each patient, including demographic data and family medical history. A detailed physical exam and diagnosis, if known, as well as a case summary, is also usually available.

Copies of outpatient diagnostic tests and procedures may also be found. Clinics may use multiple disease classification systems depending on the diagnosis (e.g., ICD-9-CM, ISCN or *International System for Human Cytogenetic Nomenclature*, [Mitelman, 1995]) and/or use proprietary coding systems (e.g., POSSUM, Mendelian Inheritance in Man). Clinic charts may also include letters and notes from other physicians, results of research studies, or diagnostic testing that borders on research.

**Accessibility and Retrievability**

Usually the medical charts for clients/patients are available at the clinic site for review and abstraction. Many clinics collect information in database format for insurance purposes, clinic needs, and network-wide data collection. Due to the nature of the information gathered, the data often are retained permanently. However, state statutes should be consulted for statute of limitations for health information.

**Strengths as a Data Source**

➢ *Accuracy*. High quality. The status of a diagnosis is qualified (i.e., the definite, rule out, possible). Although some patients never get a definitive diagnosis, the differential diagnosis is usually provided.

➢ *Level of detail*. High quality. Specific information on syndromes (identification and description of dysmorphic features) and chromosomal anomalies is often provided.

➢ *Case identification*. Specialty clinics, like those for genetics, are important outpatient data sources. Previously unknown cases may be identified for the surveillance program.

➢ *Case identification or screening*. This is a useful source for prenatal diagnosis cases. Clinics may provide diagnosis and/or genetic counseling services.

➢ *Retrievability*. Most pertinent information is entered into an electronic file (i.e., a database). This facilitates requesting specific pieces of information that can be extracted in electronic format.

**Weaknesses as a Data Source**

➢ *Population base*. May not be well defined.

➢ *Incomplete information*. Nature of the clinic business or the clinic encounter determines whether the complete diagnostic picture is available (i.e., the case may be referred for cytogenetics laboratory confirmation only).

➢ *Timeliness of diagnosis*. Some diagnoses are not confirmed until multiple diagnostic procedures have been conducted. Some syndromes take a long time to be diagnosed definitively.

➢ *Follow-up*. Often a case is referred for consultation and is lost to future tracking. This is important if the diagnosis is reported to the surveillance program as possible or rule out and is in the continuing or discovery phase.

**Liaisons and Partnerships**

➤ *Genetic counselors.* Clinics are often staffed by genetic counselors who contribute documentation concerning a patient's evaluation. They are often accessible to surveillance staff if a medical records review or other follow-up is needed.

➤ *Database managers and other office administrators at clinic sites.* Clinical information is often abstracted from documentation in the medical record for billing, research, or other clinic use. These persons can assist the surveillance staff in identifying efficient reporting and case identification methods.

➤ *Network system managers.* Regional genetics information may be collected and compiled in a database. Like hospital discharge data, regional genetics information is collected from participating clinics in a standardized format and compiled in a centralized format. Surveillance staff can utilize the efficiency of accessing a centralized database and bypass having to collect the case reports from individual clinics. Of importance is the fact that data from these sources are unlikely to include personal identifiers

**Issues to Consider**

➤ *Scope of information collected.* Genetics clinics may collect information and provide a diagnosis that extends beyond the types of defects included in a birth defects surveillance system. Passive case ascertainment systems should be precise in specifying the diagnoses that are included in the program's case definition and which are reportable. Active case ascertainment programs could improve efficiencies by developing a more precise list of diagnoses and medical conditions that can be used to screen for potential birth defects cases in the database or log of the clinic.

➤ *Confidentiality issues.* Genetics information may be protected by additional federal or state statutes. The surveillance system should research applicable legislation, and if necessary, strengthen security procedures and processes in the surveillance system.

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# Chapter 7

# Data Quality Management

# Table Contents

# Appendix

# 7.1  Introduction

The credibility of a birth defects surveillance program is built on a foundation of high-quality data. Information and results that are derived from surveillance data should be accurate, complete, and timely. Data quality influences the results of descriptive epidemiologic studies and, therefore, their interpretation. Data quality also affects the extent to which information can be utilized for planning, prevention, and intervention.

In this chapter, we will discuss some of the issues that affect the quality of data in surveillance systems and suggest methods for quality improvement. In Section 7.2 we present criteria designed to produce high-quality data. In Section 7.3 we introduce some relevant terminology. In Section 7.4 we discuss the relationship between data sources and quality, and in Section 7.5 we outline the distinctions between timeliness on the one hand versus thoroughness and completeness on the other. Sections 7.6 and 7.7 present various aspects of quality control and quality assurance, stressing the differences between the two. The importance of computer technology in support of quality improvement is particularly highlighted in Section 7.7. Nine specific quality improvements methods are discussed in detail in Section 7.8. References cited in this chapter may be found in Section 7.9.

This chapter contains a Data Sources Descriptive Assessment Tool that may help surveillance staff systematically evaluate the various data sources available to them (Appendix 7.1).

# 7.2 Criteria for High-Quality Data

High-quality data have a positive cascading affect on a surveillance program's outcome measurements – such as accuracy, completeness, and timeliness – which, in turn, can be monitored as a means to improve program performance. The term *quality* has many definitions and interpretations depending on use and intent. Philip Crosby, a total quality management expert, defines quality as "the conformance to agreed and fully understood requirements" (Dale and Bunney, 1999). In the surveillance field, this translates into the identification of a target, bench mark, or goal that defines the requirements against which results are measured.

Some experts in surveillance have suggested that the most important measurement indicators (or criteria) related to high-quality data are described by the mnemonic TACOMA (NAACCR, 2000). Data must be **T**imely, **A**ccurate, **C**omplete, **O**riented, **M**easurable, and **A**pplicable. The relative importance of these factors should be weighed and balanced, individually and in total, against the program's objectives and resources.

In the next section we define the terms on which the TACOMA mnemonic is based – timeliness, accuracy, completeness, oriented, measurability, and applicability – as well as several additional terms important for an understanding of data quality issues.

# 7.3 Terminology

| | |
|---|---|
| **Timeliness** | The extent to which data are rapid, prompt, and responsive. For example, a birth defect case should be ascertained or reported to the program shortly after diagnosis. With rapid case identification, the program is able to provide timely prevention and intervention services, respond quickly to investigations, and monitor trends. |
| **Accuracy** | The extent to which data are exact, correct, and valid. For example, accurate diagnostic data affect a program's ability to provide reliable disease rates and to maintain data comparable to those from other programs. Diagnostic accuracy reflects the program's standard to conform to agreed-upon case definitions and requirements. |
| **Completeness** | The extent to which data are all-inclusive and comprehensive. For example, are all of the cases of birth defects that occur within the target population, within a specified time period, identified by the surveillance system? |
| **Oriented** | The extent to which data are focused, targeted, and intended. For example, programs should collect only those data that are appropriate to their goals and objectives. Programs should determine which data variables should be collected, how quickly they can be collected, and the resources available to be devoted to their collection. Having an oriented perspective parallels the 'minimum necessary' privacy standard of the Health Insurance Portability and Accountability Act or HIPAA (i.e., identify and use only what is necessary). (See Chapter 2 on Legislation for additional information on HIPAA and Chapter 4 on Data Variables recommended for consideration by birth defects surveillance programs.) |
| **Measurability** | The extent to which data are quantifiable, calculable, and objective. For example, the conformance to agreed-upon data definitions provides the foundation for quantitative evaluations. |
| **Applicability** | The extent to which information is relevant. Outcome measurements should be designed to promote data utilization. Information derived from the data should be beneficial to the target population or to public health interests. |
| **Comparability** | The extent to which the data in one data set conform with those in other data sets. For example, programs that agree to adhere to standard data definitions and case definitions produce data that can be evaluated and weighed against one another. |
| **Thoroughness** | The extent to which data collection activities are meticulous and exhaustive in completing a case abstract or case record. In other words, each data field on case abstracts and case records should be filled in. |
| **Outcome measurements** | Strategically planned results that may be quantitative or qualitative. Criteria, such as those described by TACOMA, or other defined factors, are specifically selected (and developed) to evaluate, track, and monitor a program target, goal, or benchmark. Desired outcome measurements are often developed in the planning stages of a surveillance program, for performance evaluations, and when adding new projects. Staff should identify the type or category of results to be measured in order to evaluate progress in achieving program goals and objectives (or study objectives, project targets, etc.). |

# 7.4  Data Sources and Quality

Depending on the case ascertainment approach, birth defects cases are found-at or reported-from data sources. Therefore, the importance of the role data sources play in case ascertainment and surveillance should not be underestimated.

Quality issues surface because of variations among data sources. Some data sources may provide diagnostic information, but may lack important demographic information. Some may be service-focused, such that a precise diagnosis may not be important. Others may provide in-depth information on a specialty area, but may not identify other conditions that co-occur. Still others are administrative databases.

A single data source has the potential to affect multiple outcome measurements. For these reasons, programs should evaluate each data source in order to describe its basic characteristics, as well as to identify its potential strengths and weaknesses. A descriptive assessment tool should be designed to answer specific questions about each data source in relation to surveillance requirements. An example of such a tool is provided in Appendix 7.1 (Data Sources Descriptive Assessment Tool).

Quantitative evaluations should include outcome measurements for accuracy, completeness, and timeliness. Often data sources are evaluated in combination with other quality assessments. For example, diagnostic accuracy may be evaluated by staff reviewing a medical record to confirm a diagnosis that was identified-at or reported-from a data source. In this example, the data source is part of the evaluation because it is where the diagnosis case report originates; however, other aspects of the case ascertainment process may be evaluated as well.

Examples of quantitative evaluations are provided in Section 7.8 of this chapter on Quality Improvement Methods.

The program should:

> ➢ **Use** the data quality criteria in TACOMA as a guide when identifying outcome measurements and when evaluating data sources.

> ➢ **Identify** other factors that are important to consider, including those that relate to staff resources, such as 'location of site' and 'volume of case reports' (in relation to distance traveled).

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# 7.5  Timeliness Versus Thoroughness and Completeness

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Surveillance systems generally have limited resources to use in meeting program objectives. Additionally, staff face dilemmas in terms of prioritizing resources to achieve the outcome measurements of timeliness, thoroughness, and completeness. Should a program set a goal of timely data at the risk of potentially missing cases? Or risk losing timeliness by setting a goal of the most complete surveillance database? It is important for programs to achieve a balance that suits their needs, while also being responsive to external requirements, such as guidelines for submitting data to the National Birth Defects Prevention Network (NBDPN), as discussed in Chapter 10 on Data Collaboration and Dissemination.

Timeliness improves a system's ability to be responsive for investigations, up-to-date for monitoring trends, and current for referral to services. Thoroughness is a measure of finished versus unfinished case abstracts and case records. Clearly, data fields that are empty or inconclusive are not useful for most outcome measurements. Completeness is important because descriptive epidemiology – including the calculation of birth defects rates – is more comparable, accurate, and reliable when a surveillance program is confident that all cases have been ascertained.

When prioritizing resources to balance the quality indicators of timeliness, thoroughness, and completeness an important outcome measurement recommended by NBDPN is that the surveillance database be 95% complete by two years past the date of birth or fetal demise. Some programs may have a longer time period for reporting birth defects and, therefore, have a longer time period for case ascertainment. Still, it is important that surveillance systems be sufficiently responsive so that complete and timely data can be turned into useful information.

Programs should evaluate the factors that impact timeliness, thoroughness, and completeness. Often resources can be used more efficiently and effectively by streamlining or redeveloping procedures in individual areas, such as case finding, data collection, and data processing (see Section 7.8 on Quality Improvement Methods).

Timeliness, thoroughness, and completeness are often intertwined and affect other quality assessments. For example, the quality control methods that evaluate case finding and case abstracting may include outcome measurements for timeliness and thoroughness. Data source evaluations include a timeliness measurement.

The program should:

➢ **Develop** productivity guidelines and standards.

➢ **Use** TACOMA criteria, especially 'oriented' and 'applicable', to assess the factors that challenge timeliness and completeness.

➢ **Use** computer technology to improve timeliness. For example, consider using the Internet for case reporting. Internet and electronic reporting also ease the burden of case reporting at data sources.

➢ **Monitor** timeliness.

# 7.6  Quality Control and Quality Assurance

'Quality control' (QC) and 'quality assurance' (QA) can be defined as a set of methods, activities, and procedures designed to improve the results of specific outcomes. For birth defects surveillance programs, these outcomes are related directly to surveillance functions, such as case ascertainment and data collection. Although active and passive case ascertainment systems may use different methods and procedures for improving data quality, the goal is the same, namely high-quality data.

*Quality control* is a retrospective and reactive approach to improvement that focuses on discovery and detection. Deficiencies and inaccuracies are found, resolved, and fixed so that final results or outcome measurements are accurate. As a result of QC procedures, high-quality data are created at the back end. In QC, the emphasis is on checking, investigating, containing, and adjusting (Dale and Bunney, 1999).

QC procedures may include re-case finding, re-abstracting, validity audits, timeliness monitoring, and data source evaluations. QC can also be used with data linkage, especially as this involves checking selected data fields, including birth weight, date of birth, name, etc. The results of QC procedures are used to evaluate, adjust, or correct the original data that were collected or the original circumstance that occurred.

*Quality assurance* is a proactive approach to improvement that focuses on prevention. Program functions are designed and activities are planned in advance to avoid inaccurate or deficient data. As a result of QA procedures, high-quality data are created at the front end or design stage. Often, the results of a QC method lead to QA activity. The QC method detects a deficiency, and the QA method redesigns the process to prevent its recurrence (Dale and Bunney, 1999).

QA procedures may include documentation (e.g., case finding, abstraction, medical records review, disease coding, data entry), the use of selective data sources, and the development and maintenance of the database infrastructure. Additionally, QA procedures can be implemented when specific outcome measurements require consistently high-quality data. Examples include (1) using an expert clinical reviewer to routinely evaluate case abstracts for data accuracy and thoroughness and (2) conducting medical records reviews to confirm a diagnosis prior to the data being used for projects like rapid case ascertainment, investigative inquiries, or statistical monitoring of trends. QA is cost efficient in the long run. Finding and solving problems can be time consuming and resource intensive, and unless the process is fixed, the same problems will continue to recur.

Maintaining high-quality data requires continual attention to improvement. Program performance is enhanced when quality improvement procedures are integrated into program operations and conducted in a consistent and systematic manner.

Refer to Section 7.8 (Quality Improvement Methods) in this chapter for specific examples of quality control and quality assurance applications.

The program should:

> ➢ **Maintain** documentation on program procedures, especially as these affect case ascertainment and data collection activities.

> ➢ **Record** and date decision items.

➢ **Identify** the sources of potential data quality issues and prioritize the impact of each on case ascertainment and surveillance. Some situations are provided in the 'quality issues' sections in other chapters in these guidelines.

➢ **Use** the TACOMA quality indicators to develop outcome measurements for evaluations. Of particular importance are quantitative evaluations of accuracy, completeness, and timeliness.

➢ **Design** meaningful evaluations, develop benchmarks, and track improvements. Quality assessments should be used to guide any decision to change or modify the program's practices and procedures.

➢ **Use** the results of quality control to design quality assurance procedures. Quality assurance is a self-propelling mechanism that ensures continual quality improvement.

## 7.7  Quality Control and Quality Assurance in the Surveillance Database

Computer technology provides many opportunities to implement quality control and quality assurance procedures. Computerization can promote standardization, perform queries on selected criteria, monitor timeliness, reduce duplication, and generate reports.

Quality assurance can be built into the design, development, maintenance, and expansion of the surveillance database. It is essential that the computer system address, at a minimum, the requirements of case ascertainment and data collection, data entry, information management, and statistical analysis. The system must also ensure security and privacy for the health information that is stored electronically (see Chapter 9 on Data Management and Security).

A database system should be documented thoroughly, with methods in place to track changes in procedures and processes and to identify security safeguards.

Standardization of data variables is an important quality assurance procedure. Data fields should have discrete definitions, and programs should standardize the information in a data field with unique codes or pre-formatted text. Drop-down windows can assist with this by providing choices and by placing limits on the options for the data field. Drop-down windows also prevent keying errors during data entry. Data fields can be programmed to perform logic checks for dates, time, age, gender-specific disease codes, and geographic information. Calculations can be programmed into data fields for measurements (such as weight, height, and head circumference) or can be programmed to complete a 'missing' measurement for a data field.

Software technology can also provide excellent resources for quality control. Procedures can be developed to monitor timeliness, productivity, and progress. Transaction logs can be used to monitor key activities and tasks. A posting-date field can be used to track staff entries as the case ascertainment process proceeds. Posting fields can also be used to monitor data source reporting trends, data collection activities, and data processing functions.

Any number of outcome measurements can be developed to track quality indicators, including measuring accuracy and completeness. Additionally, computer technology is uniquely suited to detect duplicate cases in the surveillance system. Information can be cross-linked on many different data fields, including name, date of birth, hospital of birth, mother's maiden name, etc.

There are almost limitless ways that computer technology can be used in quality control. The database integrates and supports surveillance activities. As such, the inputs and outputs of the database play a role in each TACOMA quality indicator. A well-designed database improves program efficiencies, outcome measurements, and data utilization (see Section 7.8 on Quality Improvement Methods).

The program should:

> ➢ **Identify** situations in case ascertainment and data collection where computer technology can be used to detect or prevent problems and to track measurements.

# 7.8  Quality Improvement Methods

Methods to measure and ensure high-quality data may vary depending on the approach to case ascertainment.

In active case ascertainment, field staff engage in the process of case identification, including gathering information and confirming a diagnosis for the case abstract. Quality control is directed at improving the way staff ascertain cases. In passive case ascertainment, the surveillance system receives case reports from data sources. Staff are not engaged in collecting the information on a case report. Additionally, a diagnosis reported on a case report is not usually confirmed prior to entry into the database. Therefore, in passive case ascertainment, quality control is directed at improving the results of the data collection process.

Although the ascertainment approaches are different, quality control and quality assurance methods can be used to achieve comparable levels of data quality across surveillance programs regardless of the ascertainment approach used.

While the list is not all inclusive, some of the methods used most frequently by birth defects surveillance programs for quality control are described below. Some are useful regardless of the case ascertainment approach and can be modified to suit the specific programmatic needs.

On the following pages we describe the following quality improvement methods in detail:

- ➢ Re-case finding
- ➢ Re-abstracting
- ➢ Validity audits and medical records reviews
- ➢ Clinical review
- ➢ Reliability and inter-rater agreement checks
- ➢ Timeliness measurements
- ➢ Data source evaluation
- ➢ Comparison/verification between multiple data sources
- ➢ Computer technology

## Improving Quality through Re-Case Finding

**Purpose**                    ***To evaluate the accuracy and comprehensiveness of the case-finding process.***

**Background**                 The case-finding process, used primarily in active case ascertainment, involves staff identifying potential birth defects cases at data sources.

**Method**                     For re-case finding, perform the same steps and functions as for case finding. Develop procedures to evaluate results from the different pathways and steps in the process. Re-case finding should be conducted on a sample of information sources. The sample should consist of an appropriate number of entries, either from a single log or from multiple logs.

**Outcome Measurements**
- *Evaluation of results between the original case-finding activity and the quality control process.* This includes calculating the false positive and false negative rates at different steps in the case-finding process. In other words, this QC procedure evaluates the decision making that results in identifying a case versus a non-case.
  - Compare the QC list and the original staff review list of potential cases found at a data source during initial case finding. This is the list that identifies which cases go on to a medical records review and which do not.
  - Compare the results of re-reviewing the medical records. This involves QC re-reviewing medical records that were selected for review during the original case-finding activity and reviewing (for the first time) some medical records that were not on the original staff review list.
  - Determine the timeliness of the case-finding process.
- *Evaluation of compliance with case-finding procedures, including assessing decision-making skills.*

**Frequency**                  It is important to develop a benchmark for re-case finding and to monitor outcome measurements periodically. The frequency with which re-case finding is conducted should be based on the demonstrated expertise and proficiency of the staff.

**Quality Assurance**
- Update case-finding procedures.
- Streamline the process to improve timeliness.

**Tips**                       The case-finding process is a critical step in case identification. Not only is it important to evaluate staff effectiveness in identifying cases (and not missing any), it is also recommended that programs evaluate program efficiencies in case finding. For example, programs should evaluate the types of conditions that are considered potential cases. An evaluation might consist of determining how many confirmed diagnoses resulted from using a 'potential condition' in the initial steps of case finding. Some programs include ICD codes (i.e., searching through a hospital's disease index) as 'potential conditions'. An evaluation might consist of evaluating the effectiveness of searching using disease codes to identify a potential case in relation to whether specific codes were predictive in identifying a true birth defects case (i.e., an abstract is created).

## Improving Quality through Re-abstracting

**Purpose**      ***To evaluate the accuracy and comprehensiveness of information that is entered on a case abstract form (hard copy or computer screen).***

**Background**      Abstracting, used in active and passive case ascertainment, is the process of gathering and recording specific information from logs, medical records, or other information sources onto standard case abstract forms or computer screens.

**Method**      For re-abstracting, gather and abstract information from the same information source and record the data using the same abstract format (e.g., hard copy or computer screen). Re-abstracting should be conducted on a sample of information sources and a range of diagnosis categories

**Outcome Measurements**

- Comparison of the results of the quality control method to the results from the original case abstract and evaluation of the differences. Evaluation of the percentage and type of false positive cases.
- Identification of types and categories of errors or deficiencies. This may include disease coding, incomplete or missing information, and data entry errors. Includes the types of data variables that are problematic.
- Evaluation of compliance with abstracting procedures and guidelines.
- Determination of the timeliness of the abstracting process.

**Frequency**      It is important to develop a benchmark for re-abstracting and to monitor outcome measurements periodically. The frequency with which re-abstracting is conducted should be based on the expertise and proficiency demonstrated by staff.

**Quality Assurance**

- Update case abstracting guidelines.
- Provide training in disease coding, as applicable.
- Incorporate additional standardization into the data entry process. For example, provide drop-down windows to select and limit choices and to prevent key stroke errors.

**Tips**      Conduct an ***abstraction form review*** to identify differences and errors on completed abstraction forms. The abstraction forms should be checked for completeness, logic, and correct coding. Additionally, it is useful to categorize the types of data variables that are problematic to abstractors. For a given time period, QC should document, for each field staff member, the total number of abstraction forms reviewed and the number that have errors, such as incomplete or illogical data and incorrect coding.

## Improving Quality through Validity Audits and Medical Records Reviews

**Purpose**            ***To evaluate the accuracy and comprehensiveness of a diagnosis that is reported by a data source or represented in a listing (e.g., hospital disease index) at a data source.***

**Background**        In programs using passive case ascertainment, birth defect cases reported by data sources are accepted without confirmation. Active case ascertainment systems may use a listing of diseases provided by data sources, in disease-coded format, as part of case finding.

**Method**             The medical record, or other medical information report, is reviewed at the site or data source that reported the diagnosis or provided the diagnosis in a listing. This method is also used in the data sources audit.

**Outcome Measurements**
- *Predictive validity.* This is the degree to which an original measurement (e.g., reported diagnosis) successfully predicts a valid or confirmed outcome of interest. In other words, it represents agreement between the case report from the data source and the medical records review performed by surveillance staff.
- *Evaluation of missed diagnoses.* In other words, how many more diagnoses were identified by the medical records review process.
- *Identification of disease-coding issues,* especially as this pertains to data sources that report birth defects in a coded format (e.g., administrative databases such as the hospital discharge data set).
- *Incorporation of an evaluation of the data source* with the validity audit.
- *Timeliness* of the review process.

**Frequency**         Passive case ascertainment systems rarely have the resources to confirm all reported cases through medical records review. Therefore, the frequency of validity audits depends on program resources, requirements, and priorities. However, it is important to develop and maintain a certain level of validity audits. Programs should develop benchmarks, set goals, monitor results, and adjust program procedures.

**Quality Assurance**
- Identify and use data sources that report a confirmed diagnosis.
- Select diagnoses for consistent, concurrent, and timely validity audits. It is recommended that the diagnoses be from the set of birth defects that are reported to NBDPN. This QA procedure is primarily for passive case ascertainment systems.
- Identify disease codes that are problematic for describing birth defects precisely. Prioritize which ones should have a consistent validity audit. This QA procedure is applicable for passive case ascertainment systems that use the ICD-9-CM coding system, and can be adapted to accommodate active case ascertainment programs that use the hospital disease index during case finding.

## Improving Quality through Validity Audits and Medical Records Reviews
### (continued)

**Tips**                    Validity checks are a quality control tool. Although used primarily by passive case
                            ascertainment systems, the tool is relevant for active ascertainment programs as
                            well (e.g., active ascertainment key data entry systems or on-line abstracting).
                            Validity checks in birth defects surveillance provide a way of evaluating the
                            accuracy of what was reported (or represented) compared with what was
                            'validated' or confirmed after an investigation or medical records review.

## Improving Quality through Clinical Review

| | |
|---|---|
| **Purpose** | ***To review the diagnoses listed on the case abstract or in the case record for accuracy and plausibility.*** |
| **Background** | Information on birth defects cases is gathered and compiled by staff in active case ascertainment. Information is reported and collected from data sources in passive case ascertainment. |
| **Method** | Case abstracts or case records are examined by a designated clinical expert. |
| **Outcome Measurements** | • Evaluation of the information recorded on the case abstract.<br>• Identification of abstracting or coding problems.<br>• Percentage and types of agreement or disagreement with clinical review result. |
| **Frequency** | Programs should develop a benchmark for volume and types of case abstracts that should be reviewed and monitor the rate of agreement. In other words, the program should determine whether all case abstracts should be reviewed, or merely a percentage. |
| **Quality Assurance** | • Update and standardize abstracting and disease coding procedures.<br>• Train staff in the deficiencies cited and evaluate compliance concurrently.<br>• Increase the volume of clinical reviews, as required. |
| **Tips** | A clinical reviewer should be proficient at disease coding since the literal text of the diagnosis needs to be translated into the most accurate disease code.<br><br>In passive case ascertainment, the medical records from all data sources that reported a diagnosis for a respective birth defect case should be available to the clinical reviewer. Document the policies and procedures for the clinical review to ensure standardization. Include instructions for assigning the disease code. |

**Improving Quality through Reliability and Inter-Rater Agreement Checks**

| | |
|---|---|
| **Purpose** | ***To evaluate rate of agreement between two or more persons for the outcomes of interest.*** |

**Background**   Results of case ascertainment and data collection should be consistent, especially when staff are required to make abstracting decisions.

**Method**
- *Dual-entry coding system (double-checking of assigned code).* At least two coders assign codes from the same list of diagnoses.
- *Dual-entry data entry*. At least two staff key information from the same case abstract into the surveillance database.
- *Dual clinical review*. At least two clinical reviewers examine the same abstracts and provide results.
- *Dual medical records reviews*. At least two staff review the same medical records and abstract information per program procedures. This may include evaluating disease code assignments. Some passive case ascertainment programs may benefit by including a clinical expert in this inter-rater reliability evaluation.

**Outcome Measurements**
- Rate of agreement
- Type of deficiencies
- Compliance with abstracting and other program procedures

**Frequency**   Programs should develop benchmarks and periodically evaluate for continued consistency.

**Quality Assurance**
- Write precise procedures.
- Develop decision-making flow charts.
- Train staff with respect to addressing any deficiencies noted.
- Develop standardized data definitions for each data element. When applicable, develop a list of acceptable responses for a data element. Use drop-down windows to facilitate selecting from a list.
- Use technology to increase the accuracy of abstracting and data entry.

**Tips**   Keep a log of decision-making items and make sure it can be referred to easily. This is important for abstracting and coding procedures. Update procedure manuals, date-stamp all changes. When disease reporting rules or procedures change, make the changes effective as of the beginning of a calendar year.

## Improving Quality through Timeliness Measurements

| | |
|---|---|
| **Purpose** | *To evaluate rapidity and readiness.* |

**Background**   All areas of case ascertainment and data collection affect how responsive the program is in meeting goals and objectives with respect to timeliness.

**Method**   The time interval between two or more points of interest is measured. Often the measurement is from one task to the next or from start to finish.

**Outcome Measurements**   Timeliness measurements can be used to evaluate and improve many areas within a surveillance program including productivity and program performance. Examples include:

- *Reporting time lags*. A measurement of the time it takes for a case report to be received-in or identified-to the birth defects program.
- *Case-finding process*. An evaluation of the time it takes to identify a case, review the medical record(s), abstract information, and complete the abstract.
- *Data processing time lags*. A measurement of how quickly information is processed for use.

**Frequency**   Timeliness can be evaluated readily. Tracking measurements can be monitored using software technology and developing date-posting fields. Queries, internal logs, and reports can facilitate this quality improvement method.

**Quality Assurance**
- Implement changes to case ascertainment procedures or processes to improve timeliness.
- Use laptops to reduce redundant steps.
- Work with data sources to improve consistency in reporting, including using electronic case reporting and Internet reporting.
- Develop computer transaction logs.

**Tips**   Evaluate the program's desired outcome measurements in relation to how long it takes to achieve them. Use the criteria in TACOMA, especially as they relate to improving timeliness. For example, the criteria 'oriented' and 'applicability' focus on selecting data variables that are important to the program. Include an evaluation of the reasons for unfinished case abstracts or case records. Data variables that consume a great deal of resources to collect should be re-evaluated for intent and usefulness.

## Improving Quality through Data Source Evaluation

**Purpose**          ***To ensure that birth defect case reporting is complete, accurate, appropriate, and within the guidelines for timely reporting.***

**Background**       Birth defects are found-at or reported-from data sources. Data sources vary in purpose, organizational structure, and scope.

**Method**           The source of the diagnostic information is evaluated for accuracy, completeness, and timeliness. This method may combine the methodology of other procedures, such as validity audits and timeliness measurements, and may also include re-case finding.

- *Accuracy.* The medical record, or other medical information report, is reviewed at the site or data source that reported the diagnosis or provided the diagnosis in a listing (see Validity Audits).
- *Accuracy.* For a large data source, such as hospital discharge data (an administrative data base), the audit may be designed to focus on a suspected hospital or unusual patterns of disease-code use.
- *Timeliness.* Time lags for reporting are evaluated per data source.
- *Completeness.* Passive case ascertainment utilizes the steps taken in active case-finding to identify all of the potential and confirmed cases of birth defects at the data source. This procedure is more difficult for passive case ascertainment to implement because of the staff resources needed to conduct comprehensive case-finding (see Chapter 6 on Case Ascertainment Methods).

**Outcome**          
**Measurements**     
- *Completion of the descriptive assessment* of the data source.
- Refer to *validity audits* and *timeliness audits*.
- *False positive rate*. What is the level of diagnostic quality from a data source?
- *Completeness rate*. What is the rate of missed individuals with birth defects? These are individual cases that were not reported-to or identified-at the data source.
- *Evaluation of data collection methods*. Is the format used for reporting cases contributing to missed case reports?

**Frequency**        Each data source should be evaluated at least once to assess a level of quality.

**Quality**          
**Assurance**        
- Use multiple data sources. One data source rarely provides comprehensive information.
- Use data sources that report a confirmed diagnosis.
- Involve the data source in discussions related to quality indicators. Develop mutually agreed-upon strategies for resolving issues.
- Encourage data sources to report cases in an electronic format, including using the Internet. This may improve timeliness and completeness. Confidentiality and privacy can be assured via encryption and other safeguards.

## Improving Quality through Data Source Evaluation
### (continued)

**Tips**       Staff from programs using passive case ascertainment often review medical records in medical records departments, and some review autopsies at pathology departments. However, these staff usually do not engage in case-finding (i.e., combing through information sources to find potential cases of birth defects). Passive ascertainment staff should engage the data source in discussions prior to a case-finding audit. It is important to involve staff at the data source in planned activities to answer their questions. A contact person at the data source should be identified to ensure minimal disruption of normal work flow once the case-finding process begins.

**Improving Quality through Comparison/Verification Between Multiple Data Sources**

| | |
|---|---|
| **Purpose** | ***To compare diagnosis, and other information, that is reported-from or identified-at different data sources.*** |

**Background**    Programs are encouraged to use multiple data sources for case ascertainment. A single data source is rarely able to provide comprehensive or accurate information.

**Method**
- Compare information that is collected from multiple data sources in order to determine what information is accurate and complete. Examples include:
  - *Confirm or invalidate a diagnosis* based on a higher level of diagnostic expertise or clinical specialty. For passive case ascertainment this could mean that a diagnosis that is reported from a high-quality data source is considered to be confirmed or valid.
  - *Clarify an incomplete or imprecise diagnosis*. Conduct follow-up to gather better information.
  - *Identify incomplete data fields* on the case abstract or case record. Some data sources may not have complete information on a birth defect case, which results in an incomplete or deficient case report.
  - *Update the case abstract or case record* with more timely information. This includes address, names, and contact information.
- Develop procedures to identify duplicate case abstracts or case records in the database. Common situations that result in duplicate case abstracts or case records are mistakes with date of birth, use of multiple or incomplete names, and adoptions.

**Outcome Measurements**
- Rate the data sources. Assign 'quality' grades for specific criteria (e.g., diagnosis quality, complete address).
- Evaluate the value-added benefit that a data source provides. For example, if two data sources identify the same cases but one source provides a higher total volume of cases, evaluate the rationale for using both data sources.

**Frequency**    The use of multiple data sources is strongly encouraged. However, a surveillance program needs to understand the potential differences in quality among data sources and adjust procedures accordingly. In active case ascertainment, the comparison and verification of information can be done in an ongoing manner. In passive case ascertainment, where each case report may not be read by staff upon receipt, a benchmark should be established and key factors evaluated. At a minimum, comparison and verification should be done annually; otherwise the volume of inconsistencies or differences may turn into a resource-intensive effort to reconcile them. Computer technology greatly enhances a program's ability to systematically conduct comparison and verification procedures.

**Improving Quality through Comparison/Verification Between Multiple Data Sources**
**(continued)**

**Quality Assurance**
- Combine or merge data that are collected or abstracted into a central case abstract or case record upon receipt. This minimizes the possibility of creating a duplicate abstract or record and reduces redundant staff work.
- Develop data linkage procedures for the large administrative, computerized data sets, such as vital records, hospital discharge data, hospital disease index, and Medicaid. Data linkage can also be developed to accommodate smaller clinic-based information systems, such as cytogenetics laboratories, genetic services, and specialty clinics. A key factor in data linkage is using standardized data variables (see Chapter 4 on Data Collection Variables).
- Develop decision-making and hierarchy models for use in comparison and verification of data elements. Programs should determine which data sources are considered a high-quality information source for specific data variables.

**Tips**　　This QC method is enhanced by using computer technology and developing a systematic approach.

**Improving Quality through Computer Technology**

**Purpose**          *To use technology in quality improvement efforts.*

**Background**   Surveillance systems are information management systems whose operations are
                 enhanced by computer technology (see Chapter 9 on Data Management and
                 Security).

**Method**
- *Quality assurance.* Use software to prevent problems and enhance
  standardization.
    - *Build in range checks* to prevent inaccurate abstracting and data entry.
      These checks can be created for any data variable with a defined parameter
      of acceptable or measurable values. Date range checks can be used for age,
      date of birth, date of fetal demise, date of death, LMP (date of last
      menstrual period).   These become the dates that other dates (e.g., date of
      case report) are compared to for rationale. Other types of range checks are
      Apgar scores, gestational age, and birth weight.
    - *Develop automated calculations and conversions* for specific data fields.
      Examples include birth weight, time and LMP .
    - *Promote the use of coded data.* Develop codes for text information. This
      method can be applied to any data variable definition that has multiple
      acceptable responses. Examples include disease, geographic, race, and
      ethnicity codes. Programs can develop code sets for data sources, specific
      sites, types of procedures, family history, physicians, etc
    - *Use drop-down windows for data fields.* This approach is useful with long
      text entries and for text that has been converted to a code.
    - *Use standard data collection variables (and data definitions)*, to
      accommodate record linkage and electronic transfers (see Chapter 4 on Data
      Collection Variables).
- *Quality control*. Develop procedures to detect, measure, and enhance
  effectiveness.
    - *Perform logic edits*. Review existing program documentation and syntax to
      ensure that the computer application is performing as intended. For
      example, when computer applications are used to convert or calculate data
      field values, make sure the results using the formula(e) are accurate.
    - *Create date-posting fields* to monitor timeliness.
    - *Develop transaction logs*. This is a method that tracks and dates additions,
      deletions, and other changes to the database.
    - *Create queries and reports* to track desired outcome measurements.
    - *Develop methods, using key data variables, to find duplicate cases* in the
      database.
    - *Develop queries to identify problem situations*. Examples include:
      – Some birth defects should not be counted due to prematurity or low birth
        weight.
      – Some ICD codes are problematic for birth defects.
    - *Develop information management systems* to improve the efficiency of
      program operations, including case ascertainment.
    - *Develop methods to improve timeliness* of case reporting. This includes
      using Internet reporting and other electronic methods, with appropriate
      security measures to protect confidentiality and privacy.

## Improving Quality through Computer Technology
### (continued)

**Outcome Measurements**   Track measurements from the QC and QA methods that are developed.

**Frequency**   Once developed, computerized quality procedures can be run on a consistent and systematic timeframe. Systems and software also facilitate flexibility for ad hoc queries and reports. Information management systems are ongoing system enhancements.

**Quality Assurance**   Design, maintain, and update to:
- Prevent problems at the source
- Promote standardization
- Improve program efficiencies, including timeliness
- Facilitate data retrieval and analysis
- Assist in tracking measurements

**Special considerations for passive case ascertainment programs**   A dilemma that primarily affects passive case ascertainment programs is how to retain the integrity of the database, while also resolving data quality problems. In other words, how do programs identify and use accurate information, especially since the majority of diagnoses in the data base are accepted as reported (i.e., not confirmed by staff)? For example, if a diagnosis is reported from a data source and is determined to be inaccurate, or incomplete, it should not be counted in statistical analysis. However, for epidemiological and evaluation purposes, this diagnostic information (and the associated information that accompanies the case report) should not be deleted from the database. A method to resolve this issue could be to develop a mechanism (perhaps a data field) that identifies or flags a diagnosis that is not accurate (valid) or should not be counted. Programs are encourage to develop methods to resolve these kinds of issues in a way that best suits the program's needs.

**Tips**
- Before building a computerized data collection system evaluate the current manual data collection instrument to determine what works and what doesn't work.
- Avoid programming an on-line data collection system based on your hard copy instrument. Once programming is completed, it is often difficult to undo.
- Prior to developing and expanding the data base, evaluate the program's needs, i.e., how the data will be used, how the data will be accessible, data transfer, etc.

# 7.9  References

The North American Association of Central Cancer Registries, Inc. (NAACCR). *Standards for Cancer Registries, Volume III: Standards for Completeness, Quality, Analysis, and Management of Data.* Springfield, IL: Registry Operations Committee of NAACCR; September 2000.

Dale B, Bunney H. *Total Quality Management Blueprint.* Malden, MA: Blackwell Publishers Inc.; 1999.

***Appendix 7.1***

***Data Sources Descriptive Assessment Tool***

# Appendix 7.1
# Data Sources Descriptive Assessment Tool

**Title of Data Source:**

| Evaluation Criteria Trait | Explanation and Description of Criteria | Specifics for This Data Source |
|---|---|---|
| | | |
| **Source or Site** | *The Source or Site is briefly defined* | |
| | | |
| **Legal or Professional Mandates** | *Legal issues or professional requirements that govern or guide operations are described* | |
| | • Statutes or regulations | |
| | • Accreditation agencies (e.g., JCAHO) | |
| | • State licensing boards | |
| | • Federal agency requirements (e.g., medical participation agreements) | |
| | | |
| **Mission or Objective** | *Purpose or reason that the Source or Site collects the information* | |
| | • How is the information used? | |
| | | |
| **Scope or Breadth** | *Time span or scope of time for the information collected* | |
| | • What time span does the Source or Site address? Point of time or follow-up capability? | |
| | | |
| **Operational Structure** | *Flow of information is described* | |
| | • Where is the information collected? | |
| | • Who collects the information? | |
| | • When is the information documented? | |
| | • How is the information documented? | |
| | • How is the information stored? | |

| Evaluation Criteria Trait | Explanation and Description of Criteria | Specifics for This Data Source |
|---|---|---|
| | | |
| **Type of Information Collected** | *Type of information collection is listed* | |
| | • Unique pieces of information that the Source or Site collects are listed | |
| **Accessibility and Retrievability** | *Location of the information* | |
| | • Accessibility to the information | |
| | • Retrievability of the information | |
| | • Data base, records merging, and other electronic applications capability | |
| | | |
| **Strengths as a Data Source/Site** | *The strengths and attributes of this Source/Site are described* | |
| | • Accuracy of the information | |
| | • Completeness of the information | |
| | • Timeliness of the information | |
| | • Efforts and resources required for case finding | |
| | • Other qualities detailed | |
| | | |
| **Weaknesses as a Data Source/ Site** | *The weaknesses and deficiencies of the Source/Site are described* | |
| | • Accuracy of the information | |
| | • Completeness of the information | |
| | • Timeliness or time lags in case reporting | |
| | • Effort and resources required to receive a case report | |
| | • Other qualities detailed | |
| | | |
| **Liaisons and Partnerships** | *Key contact people/departments are identified* | |
| | • Ways to enhance cooperation and partnerships are described | |
| | | |
| **Additional Comments** | *Issues to consider* | |
| | | |
| **References** | *Contact information* | |
| | • States with experience | |
| | • Literature references | |

# *Chapter 8*

# *Statistical Methods*

# Table of Contents

# 8.1 Introduction

Statistics are useful to surveillance programs for:

 ➢ Summarizing and comparing surveillance data

 ➢ Assessing the potential role of chance or random variability

 ➢ Controlling for the effects of extraneous factors

The objective of this chapter is to present some common statistical concepts and tools that can be applied to surveillance data. For each tool, a definition is provided, along with background information, guidelines for use, how to calculate, and an example. This is a basic introduction only; more exhaustive treatment of these topics can be found in the reference literature.

In Section 8.2 of this chapter we discuss measures of birth defect occurrence. General issues relating to prevalence are discussed in Section 8.3, with the distinctions between crude prevalence, specific prevalence, and adjusted or standardized prevalence presented in Section 8.4. Various approaches to presenting and displaying descriptive epidemiology are described in Section 8.5, while confidence intervals and their calculation are discussed in Section 8.6. Finally, in Section 8.7 we discuss means to rule out straightforward explanations for observed changes in the prevalence of a birth defect. References cited in this chapter may be found in Section 8.8.

# 8.2  Measuring Birth Defect Occurrence

In carrying out basic epidemiologic and statistical assessment of birth defects occurrence, the analyst needs to decide what to count (issues of case definition are discussed in Chapter 3) and how to use those counts in calculations. This section presents some basic concepts, clarifies definitions with respect to analysis and reporting, and presents alternatives to the standard methods used to measure birth defects occurrence, birth prevalence.

## 8.2.1  Multiple Birth Defects in the Same Child

Analyses of birth defects surveillance data should be based on cases. An infant or fetus can have multiple birth defects and can be counted as a separate case for each defect. Thus, an infant/fetus with anencephaly and cleft lip should be counted as a case of anencephaly, and again as a case of cleft lip. When using this approach, it is important to recognize that the number of different cases cannot then be added to reach a total number of infants/fetuses.

When an infant/fetus has two or more conditions coded in the same category in an analysis, count it once only. For example, if an infant has atrial septal defect and ventricular septal defect. Count the infant once in tabulations for atrial septal defect, and once in tabulations for ventricular septal defect. Additionally, count the infant only once in tabulations for cardiac defects.

## 8.2.2  Counts, Ratios, Proportions, and Rates

The most common measures of birth defect occurrence are counts, ratios, proportions, and rates.

**Counts.** *Counts* present the simple enumeration of cases. Such information can be useful for health planning purposes, where it is important to measure the burden of birth defects on existing health care resources, to assess the need for additional resources, and for cluster investigations. However, simple counts of cases are not of value as a measure of disease risk, for which rates are necessary.

**Ratios.** A *ratio* is composed of one number (the numerator) divided by another (the denominator). Ratios can be useful for comparing the number of cases in one population group with the number in another. Proportions and rates (discussed below), and prevalence (discussed in Section 8.3) are special types of ratios.

FORMULA:        A / B      or      numerator / denominator.

EXAMPLE:        The sex ratio of cleft palate cases would be represented as notated below.

$$\frac{\text{number of male cases with cleft palate}}{\text{number of female cases with cleft palate}}$$

**Proportions.** In a *proportion*, the cases in the numerator must be included in the denominator. A *percentage* is a proportion multiplied by 100. Proportions are useful for describing basic characteristics of surveillance program data. This can help with quality control.
FORMULA for a proportion:        A / (A+B)

FORMULA for a percentage:          A x 100 / (A+B)

EXAMPLES

The *proportion* of abstracted records with errors would be:

$$\frac{\text{the number of records with errors}}{\text{the total number of records}}$$

The *percentage* of abstracted records with errors would be:

$$\frac{\text{the number of records with errors}}{\text{the total number of records abstracted}} \text{ x } \quad 100$$

**Rates.** In epidemiology, *rates* express the frequency with which an event occurs (e.g., the number of new cases of disease) in a defined population in a specified period of time (Last, 1995).

FORMULA for incidence rate:

$$\frac{\text{the number of new cases of a disease during a period of time}}{\text{population at risk}}$$

As will be discussed further in Section 8.3, although some investigators and studies report 'incidence rates' when talking about birth defects occurrence, there is general consensus that the information to determine incidence is not available (Sever, 2004). Therefore 'prevalence' or 'prevalence at birth' is the more appropriate terminology.

---

# 8.3  Calculating Prevalence at Birth

---

*Prevalence* expresses the number of existing cases of disease at a point in time divided by the total population. Prevalence is useful since it allows comparison between populations of different sizes. Prevalence may be measured at any time (e.g., X cases of spina bifida of any age on June 1 2003 divided by the entire population). However, for measuring occurrence of birth defects, it is most common to use prevalence at birth or birth prevalence. That is true even though many of the cases included may not have been live births.

Ideally, incidence rates would be used instead of prevalence to measure birth defect occurrence. *Incidence rates* measure the occurrence of new events that occur in a population, so the formula for incidence of a birth defect would be:

$$\frac{\text{the number of new cases of birth defect A in an area and time period}}{\substack{\text{the number of conceptions at risk of developing defect A in} \\ \text{that area and time period}}} \quad \times \quad \text{multiplier}$$

Since the number of conceptions is unknown, as is the number of cases "lost" through spontaneous abortions, technically speaking we cannot determine incidence. Because of this, as noted above, most epidemiologists working in the area of birth defects use the term 'prevalence' to refer to birth defect occurrence. For a more complete discussion of this issue see Sever (2004).

## 8.3.1  Basic Calculation for Prevalence at Birth

Surveillance programs should measure birth defect occurrence using the following formula for birth prevalence. Note that both the numerator (number of cases) and the denominator (number of live births) always come from the same area and time period, that is, the same population. Usually the denominator is the number of live births to residents in the same geopolitical area from which the cases came during the same time period.

FORMULA for birth prevalence (expressed as cases of defect A per 10,000 live births):

$$\frac{\text{the number of cases with birth defect A in an area and time period}}{\text{the number of live births in that area and time period}} \quad \times \quad 10{,}000$$

EXAMPLE from Missouri:

$$\frac{\text{193 cases with Tetralogy of Fallot statewide delivered in 1989-1995}}{\text{532,592 live births}} \quad \times \quad 10{,}000$$

= 3.62 cases per 10,000 live births

## 8.3.2  Numerator and Denominator Issues

**Counting cases – the numerator.** For information on how to count cases for the numerator, see Section 8.2.1 on measuring birth defect occurrence (cases versus infants/fetuses).

**Pregnancy outcomes included.** Most often in analyses of birth defects surveillance data, the cases in the numerator are derived from all pregnancy outcomes collected by the program. These *may* include those listed below (see Chapter 3 on Case Definition).

➢ Live births

➢ Spontaneous fetal deaths greater than or equal to 20 weeks gestational age (GA)

➢ Spontaneous fetal deaths less than 20 weeks GA

➢ Induced terminations greater than or equal to 20 weeks GA

➢ Induced terminations less than 20 weeks GA

➢ Fetal deaths, unknown if spontaneous or induced, and/or of unknown gestational age

Sometimes analyses are restricted to certain pregnancy outcomes in comparing data from two surveillance programs that collect different pregnancy outcomes. For example, in the annual reports of EUROCAT (2002) and the International Clearinghouse for Birth Defects Monitoring Systems (2002) data are presented by live births plus stillbirths (late fetal deaths) and induced abortions (terminations of pregnancy), such that it is possible to make comparisons between prevalence based on comparable methods of case ascertainment (Sever, 2004).

The reports generated by the program should document which pregnancy outcomes are included in the numerator.

For the denominator, surveillance programs should use the total number of live births in the same area and time period from which the cases were ascertained. Although including induced and spontaneous fetal deaths would more closely approximate incidence rates calculated in other areas of epidemiology, it is impractical since these other pregnancy outcomes are often inaccurately counted compared to live births. In addition, these counts are small in comparison to the number of live births and are unlikely to affect prevalence to a large degree.

All the cases in the numerator (e.g., spontaneous and induced fetal deaths) may not come from the denominator (live births). For this and other technical reasons, birth prevalence is really a ratio and not a rate, although it is commonly referred to as a 'rate'.

**Multiplier.** The multiplier 10,000 is used for convenience, so that prevalence of most defects will have at least one digit to the left of the decimal place. Prevalence is expressed as 'X cases per 10,000 live births'. The most common multipliers for birth defects are 10,000 and 1,000.

EXAMPLE. 6.3 cases per 10,000 live births is easier for the reader to understand than 0.00063 cases (per birth).

# 8.4 General Types of Prevalence

As with rates and other measures of morbidity and mortality, there are three general types of prevalence. Prevalence can be categorized based on whether it:

➢ Applies to the whole population – ***crude prevalence***

➢ Applies to subgroups within the population – ***specific prevalence***

➢ Applies to the whole population, but adjusts for differing distribution of subgroups within the population – ***standardized or adjusted prevalence***.

Below we discuss each of these types of prevalence in turn.

## 8.4.1 Crude Prevalence

**Definition**     Prevalence calculated for the entire population without regard to possible subgroups within the population.

**When to Use**     When a single, easily calculated number summarizing the occurrence of disease in a population is desired.

**How to Use**     For birth defects, the basic calculation above is applied to the entire population. The area is usually the area covered by your birth defects surveillance program.

$$\frac{\text{The number of cases with birth defect A in an area and time period}}{\text{the number of live births in that area and time period}} \times 10,000$$

EXAMPLE

**Birth Prevalence of Down Syndrome, Texas, 1996/97 Deliveries**

| Cases | Live Births | Prevalence* |
|-------|-------------|-------------|
| 355 | 300,431 | 11.82 |

\* cases per 10,000 live births     Source: Ethen and Case, 2000.

## 8.4.2 Specific Prevalence

**Definition**     Prevalence calculated for subgroups or ***strata*** within the population, such as age groups, sex groups, or racial/ethnic groups. These then would be referred to as age-specific prevalence, sex-specific prevalence, or race/ethnicity-specific prevalence. The term 'stratified' is also used to refer to the prevalence among such subgroups.

**When to Use**     Specific prevalence is used in looking at disease occurrence in subgroups of a population. It is also used when sufficient data are available to define and categorize the population of interest. In using specific prevalence, it is important

to consider how missing values (e.g., unknown maternal ages) would affect the interpretation of the prevalence data presented.

**How to Use**        For birth defects, apply the basic prevalence calculation above to each group of interest within the population. It is necessary to have the numerator and denominator from the same group of interest. The most common groups of interest for routine birth defects reports are based on:

- Maternal age at delivery

- Maternal racial/ethnic group

- Infant sex

However, specific prevalence can be calculated for any group for which numerator and denominator data are available.

Each grouping of a variable of interest is also called a 'stratum'. For example, common strata for maternal age at delivery are:

- Less than 20 years old

- 20 – 24 years old

- 25 – 29 years old

- 30 – 34 years old

- Greater than or equal to 35 years old

Calculating maternal age-specific prevalence would then yield five values.

It is helpful to define the groups or strata in the same way vital statistics are routinely reported for the population of the area. For example, live births in Texas are commonly reported for four maternal racial/ethnic groups:

- White (non-Hispanic)

- Black

- Hispanic

- Other

Thus, those categories are used for reporting race/ethnicity-specific prevalence values.

The FORMULA for calculation is:

$$\frac{\text{the number of cases with birth defect A in group X in an area and time period}}{\text{the number of live births in group X in that area and time period}} \times 10{,}000$$

EXAMPLE

**Prevalence of Down Syndrome by Maternal Age in Years, Texas 1996/97 Deliveries**

| Maternal Age (years) | # Cases | # Live Births | Prevalence* |
|---|---|---|---|
| < 20 | 39 | 48401 | 8.06 |
| 20 – 24 | 53 | 83398 | 6.36 |
| 25 – 29 | 45 | 81442 | 5.53 |
| 30 – 34 | 84 | 57562 | 14.59 |
| 35 + | 134 | 29574 | 45.31 |

\* cases per 10,000 live births      Source: Ethen and Case, 2000.

# 8.4.3  Adjusted or Standardized Prevalence

**Definition**

Prevalence calculated for the entire population (the target population) that adjusts for (eliminates the influence of) possible differences in makeup between it and some standard or reference population. It is a summary measure that is a weighted average of the stratum-specific prevalence values.

There are two types of adjusted or standardized prevalence: direct and indirect. ***Direct adjustment*** uses specific prevalence derived directly from the target population (hence the name) and combines them using the age distribution of the standard population. The prevalence is generated that the target population would have experienced had it had the same age structure as the standard population. ***Indirect adjustment*** uses age-specific prevalence figures derived from the standard population but applies those to the age distribution of the target population. This technique produces the number of cases the target population would have experienced had it had the same age-specific prevalence as the standard population. The final result is usually expressed as a ratio of the cases observed in the target population divided by the number of cases expected based on this calculation.

**When to Use**

Adjustment is used to develop a single number summarizing the occurrence of birth defects within a population compared with some other population, removing the effect of differences between populations in the distribution of the factor adjusted for. An example would be to examine the occurrence of Down syndrome in a community near a hazardous waste site where the community has a larger proportion of older mothers than a comparison community or the state as a whole. The most common characteristics adjusted for in birth defects analyses are maternal age and maternal racial/ethnic group.

Use direct adjustment:

- When information is available on both the number of cases and the number of live births in each group/stratum/level of the factor being adjusted for (e.g., in each maternal age group); or

- To compare two or more target populations with each other (e.g., prevalence for anencephaly in 20 counties [20 target populations], standardized for maternal race-ethnicity group).

Note that to compare two or more target populations, they must be standardized using the same standard population.

Use indirect adjustment when:

- Information is not available on the number of cases in each stratum of the factor being adjusted for;

- The comparison is between a target population and a standard and not with another target population (e.g., is the prevalence of anencephaly significantly different in County X compared to the whole state?);

- Statistical precision is very important (since stratum-specific prevalence used for direct standardization can sometimes vary widely if based on few cases); or

- The results are to be presented as an observed-to-expected ratio (although a prevalence can be calculated).

**How to Use**        The following instructions are based on adjusting for maternal age groups. Each age group is called a 'stratum'. The same process would be used when adjusting for other characteristics, for example, race or ethnicity.

*Direct adjustment.* For direct adjustment follow the steps below.

1. Decide on age-group categories (strata) that can be applied to both the target and standard populations.

2. Calculate age-specific prevalence for each stratum of the *target* population. Do not use the multiplier (10,000 or 1,000) for this calculation. However, the multiplier may be used for presenting the age-specific prevalence values.

3. Multiply each prevalence by the number of live births in the same stratum of the *standard* population. This gives the number of cases expected in each stratum of the standard population, had it experienced the same age-specific prevalence as the target.

4. Add up the number of expected cases across all strata.

5. Divide the total number of expected cases by the total number of live births in the *standard* population and multiply by your multiplier (e.g., 10,000).

This is the 'directly standardized birth prevalence'.

*Indirect adjustment.* For indirect adjustment follow the steps below.

1. Decide on age-group categories (strata) that can be applied to both the target and the standard populations.

2. Calculate age-specific prevalence for each stratum of the *standard* population. Do not use the multiplier (10,000 or 1,000) for this calculation. However, the multiplier may be used for presenting the age-specific prevalence.

3. Multiply each prevalence by the number of live births in the same stratum of the *target* population. This gives the number of cases expected in each stratum of the target population, had it experienced the same age-specific

prevalence as the standard.

4. Add up the number of expected cases across all strata of the *target* population.

5. Divide the total number of observed cases in the *target* population by the calculated total number of expected cases.

This is the 'standardized birth prevalence ratio', sometimes called the 'standardized observed-to-expected ratio'. When applied to mortality, the result is called the 'standardized mortality ratio' or SMR.

EXAMPLES. The Texas Birth Defects Monitoring Division dealt with a cluster of Down syndrome in a three-county area in Texas among deliveries in 1992–1994. Down syndrome is influenced strongly by maternal age. Thus it was necessary to adjust for maternal age to see whether the excess was still apparent when possible differences in maternal ages between these three counties and the state of Texas were removed. The three counties (1992–1994 deliveries) make up the *target population*, and the entire Texas Birth Defects Registry area (1996–1997 deliveries) the *standard population*. The years 1996–1997 were used because those were the first years with data published for most of the state.

*Direct adjustment.* The crude Down syndrome prevalence for the three counties during 1992–1994 was 31.97 cases per 10,000 live births. **Steps 1-4** are presented in the following table.

| Maternal Age (years) | Target | | | Standard | |
|---|---|---|---|---|---|
| | # Cases | # Live Births | Prevalence* | # Live Births | Expected # Cases |
| | (A) | (B) | (A) / (B) = (C) | (D) | (C) x (D) |
| < 20 | 3 | 1032 | 0.00290698 | 48401 | 140.701 |
| 20 – 24 | 3 | 1666 | 0.00180072 | 83398 | 150.176 |
| 25 – 29 | 3 | 1498 | 0.00200267 | 81442 | 163.101 |
| 30 – 34 | 4 | 1028 | 0.00389105 | 57562 | 223.977 |
| 35 + | 5 | 407 | 0.01228501 | 29574 | 363.317 |
| | | | | | |
| Total | 18 | 5631 | 0.00319659 | 300377 | 1041.272 |

\* Expressed as cases per live birth. To express it the usual way, multiply by 10,000.

**Step 5**

$$\frac{\text{total number of expected cases in standard population}}{\text{total number of live births in standard population}} \times 10{,}000$$

$$= \frac{1041.272}{300377} \times 10{,}000$$

= the standardized prevalence

*Indirect adjustment.* **Steps 1-4** are presented in the following table. There are known to be 18 cases of Down syndrome in the target population, but the ages of their mothers may not be known.

| Maternal Age (years) | TARGET # Live Births | STANDARD # Cases | STANDARD # Live Births | STANDARD Prevalence * | TARGET Expected # Cases |
|---|---|---|---|---|---|
| | (E) | (F) | (G) | (F) / (G) = (H) | (E) x (H) |
| | | | | | |
| < 20 | 1032 | 39 | 48401 | 0.00080577 | 0.83155307 |
| 20 – 24 | 1666 | 53 | 83398 | 0.00063551 | 1.05875441 |
| 25 – 29 | 1498 | 45 | 81442 | 0.00055254 | 0.82770561 |
| 30 – 34 | 1028 | 84 | 57562 | 0.00145930 | 1.50015635 |
| 35 + | 407 | 134 | 29574 | 0.00453101 | 1.84411983 |
| | | | | | |
| Total | 5631 | 355 | 300377 | 0.00118185 | 6.06228927 |

* Expressed as cases per live birth. To express it the usual way, multiply by 10,000.

**Step 5**

total number of observed cases in target population  x
total number of expected cases in target population

= $\dfrac{18}{6.0623}$   x   10,000

= 2.97

= standardized birth prevalence ratio or standardized observed-to-expected ratio

# 8.5 Descriptive Epidemiology

**Background**        Surveillance data allow the description of the occurrence of birth defects in terms of the basic epidemiologic parameters of time, place, and person. In doing so, comparisons can be made among these different parameters (e.g., comparing the prevalence of anencephaly among different maternal ages).

Note that descriptive epidemiology is really just the presentation of specific prevalence where the strata are time periods (e.g., year of delivery), areas (e.g., counties), or personal characteristics (e.g., maternal age groups or maternal racial/ethnic groups). See Section 8.4.2 above on calculation of specific prevalence.

**When to Use**        To describe patterns of birth defect occurrence.

**How to Use**        Choose the most appropriate measure of birth defect occurrence. This will usually be prevalence, but in some circumstances it may be counts. For example, in planning for services, the actual number of children in a population born with a defect, such as cleft lip, may be more important than the prevalence.
Analyze the chosen measure according to the basic epidemiologic parameters of time, place, and person (Teutsch and Churchill, 2000; Seiffert, 1994). Tables, graphs, and maps are very useful in presenting data in an understandable form; suggested approaches can be found in Teutsch and Churchill (2000).

*Time.* Surveillance programs should clearly state the relevant time period of study from which cases are drawn. This is usually based on 'date of delivery'. Other options include 'estimated date of conception', 'estimated date of delivery', 'date of diagnosis', or 'date of incorporation into the surveillance database'. Common analyses of birth defect occurrence by time include:

- Prevalence by year: to look for long-term trends

- Prevalence by season or month within the year: to look for seasonal patterns

- Counts of cases by date of delivery or estimated date of conception: to look at birth defect clusters

*Place.* Reports should specify the geographic area of coverage for the prevalence presented in the report. Common analyses of birth defect occurrence by place include:

- Prevalence by state, county, or region

- Prevalence by zip code or census tract

- Spot maps of cases showing residence at delivery or conception

*Person.* Common analyses of birth defect occurrence by person include:

- Prevalence by maternal age (age of the mother at the time of delivery)

- Prevalence by maternal race/ethnicity (the most common definition is as stated by the mother)

- Prevalence by infant/fetus sex.

EXAMPLES

*Time* trend in the prevalence of gastroschisis over several years. The numerator (number of cases) and denominator (number of live births) for each year is determined. The prevalence for each year is calculated as:

<div align="center">

the number of cases with gastroschisis in Metro Atlanta in year X   x        10,000
the number of live births in Metro Atlanta in year X

</div>



Figure 7.36 - GASTROSCHISIS
TRENDS IN REPORTED BIRTH PREVALENCE AMONG LIVE BIRTHS AND STILLBIRTHS, METROPOLITAN ATLANTA CONGENITAL DEFECTS PROGRAM JAN 1968 - DEC 1991
(RATES PER 10,000 LIVE BIRTHS)
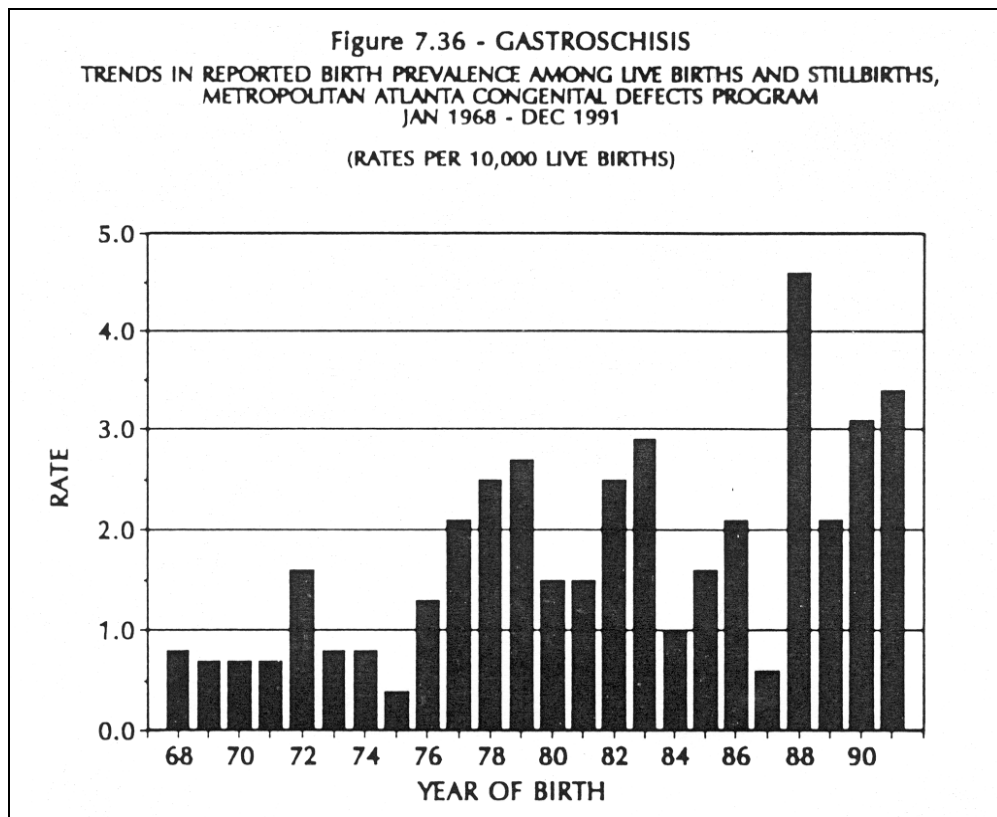
Source: Centers for Disease Control and Prevention, 1993, as cited in James et al., 1993.

Occurrence of birth defects by *place*, specifically, county prevalence of gastrointestinal defects. Numerators and denominators were collected for each county in Florida for deliveries in 1996. The results are presented in a chloropleth map (a method of mapping to display quantitative information where the areas [e.g., counties] are colored or shaded according to the value of some variable [e.g., gastroschisis prevalence]).

Source: Florida Department of Health, 1999.
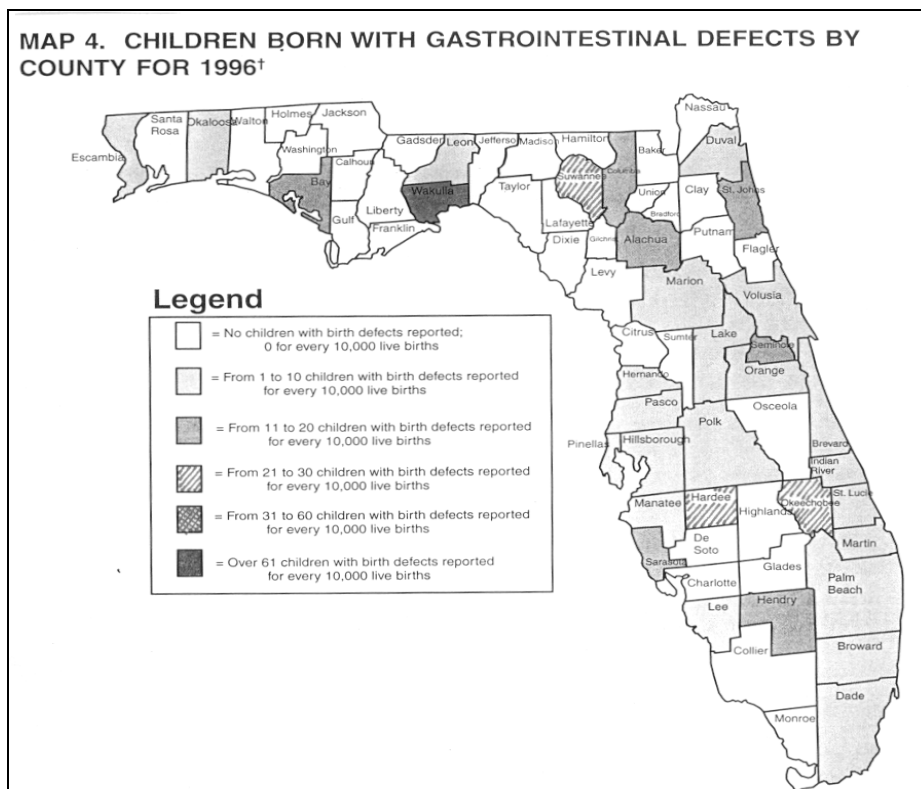
Occurrence of birth defects by *person*. Numerators and denominators were determined for five maternal age groups. Stratum-specific prevalence is calculated and presented in a table and vertical bar graph.

**Prevalence of Down syndrome by maternal age, Texas 1996-1997**

| Maternal Age (years) | Cases | Live Births | Prevalence* |
|---|---|---|---|
| < 20 | 39 | 48,401 | 8.06 |
| 20-24 | 53 | 83,398 | 6.36 |
| 25-30 | 45 | 81,442 | 5.53 |
| 30-34 | 84 | 57,562 | 14.59 |
| 35 + | 134 | 29,574 | 45.31 |

*cases per 10,000 live births
Source: Ethen and Case, 2000.
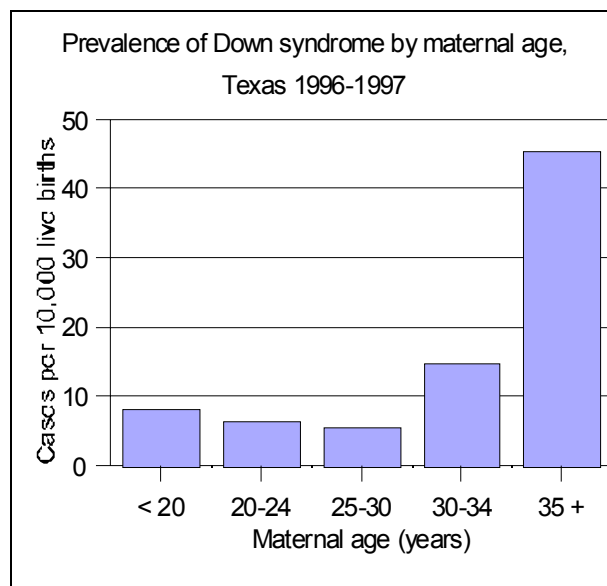


Source: Ethen and Case, 2000.

# 8.6  Confidence Intervals

In this section, we first discuss confidence intervals generally (Section 8.6.1). We then discuss the use of confidence intervals in comparing prevalence values (Section 8.6.2).

## 8.6.1  About Confidence Intervals

**Definition**       An interval around a statistic that contains the true underlying value of the statistic a certain amount of the time. For example, a 95% confidence interval for the prevalence of spina bifida will contain the true underlying value of the spina bifida prevalence 95% of the time.

The interval is bounded by an upper confidence limit and a lower confidence limit.

**Background**      The birth prevalence for a particular defect is estimated by the number of cases with the defect of interest ascertained from the population, divided by the total number of live births, and multiplied by some factor such as 10,000. This number is the best estimate of the true birth prevalence, which can never be known with certainty. To provide an idea about the precision of the estimated prevalence, a range of values is often calculated that is highly likely to contain the true prevalence. This range of likely values is called a ***confidence interval***.

A confidence interval is calculated in such a way that, if the same procedure were to be repeated a large number of times, the proportion of intervals that contain the true prevalence would equal the confidence level. So, for example, if we choose a 95% confidence value, then 95% of all those confidence intervals will contain the true, but unknown, prevalence.

The 95% value is the conventionally used confidence interval. However, sometimes people choose other values, such as 90% confidence intervals (which are narrower than 95% confidence intervals) and 99% confidence intervals (which are wider).

Estimated confidence intervals for any given level (e.g., whether 90%, 95%, or 99%) will be narrower when their prevalence values are based on more cases.

Confidence intervals only measure random error, for example, when the occurrence of a birth defect fluctuates up and down from year to year by chance. They do not address systematic error or bias.  Let's say, for example, one wanted to compare two surveillance programs. If program A does not ascertain cases from prenatal diagnosis clinics and thus consistently misses cases, while program B does ascertain cases from such clinics, a confidence interval around the prevalence of birth defects from program A should not be interpreted as accounting for the missing cases. The confidence interval merely addresses random fluctuation in the cases from program A.

**When to Use**          Confidence intervals may be calculated for any summary statistic, e.g., for
                         proportions. However, we will only discuss confidence intervals for birth
                         prevalence since that will be the most common statistic presented by birth defects
                         surveillance programs. To learn about calculating confidence intervals for
                         proportions or other types of rates, please consult one of the statistics books listed
                         in the reference section (e.g., Dawson and Trapp, 2001; Fleiss et al., 2003;
                         Snedecor and Cochran, 1989).

                         Confidence intervals can be used whenever calculating birth defect prevalence,
                         although their use is controversial. This issue is discussed extensively in a recent
                         commentary (Costa and Kirby, 2003) and a theory and methods paper (Correa-
                         Villasenor et al., 2003) on the use of confidence intervals and on errors and
                         undercounting in birth defects surveillance data. The interested reader is referred
                         to these publications.

**Why to Use**           From a theoretical viewpoint, prevalence (e.g., X cases per 10,000 among
                         deliveries in 1999) can also be considered to be just one sample in time, and
                         confidence intervals give an idea of the range of values within which the true
                         value is likely to be found. From a practical viewpoint, confidence intervals are
                         particularly useful when dealing with small numbers of cases or where the birth
                         defect prevalence for one group will be compared with that of other groups. This
                         is because confidence intervals can help minimize reader concern about
                         prevalence values that appear high or different when this is most likely due to
                         random fluctuation. Some states have found this to be particularly helpful, for
                         example, when looking at prevalence for counties, areas smaller than counties, or
                         racial/ethnic groups. While the best way to compare prevalence values between
                         different areas is always to use a statistical test, it is not practical for a
                         surveillance system to anticipate all the comparisons readers will want to make.
                         Confidence intervals thus provide a quick way for readers to get a rough idea of
                         the impact of chance on the data.

**Why Not to Use**       Some surveillance programs ascertain all cases of birth defects, so that the
                         prevalence reported is not just a sample but is considered to reflect the
                         underlying true prevalence. Therefore, the use of confidence intervals is
                         considered by many to be irrelevant. Calculating confidence intervals also
                         increases statistical work for program staff. Finally, some data users, for example
                         community groups or the media, may find confidence intervals confusing.

**How to Calculate**     Upper and lower 95% confidence limits are shown in the table below; they are
                         the end points of the corresponding confidence intervals. Note that calculation of
                         confidence intervals for prevalence is merely the calculation of confidence
                         intervals for the number of cases – the denominator portion of prevalence does
                         not change.

                         *For a prevalence based on a small number of cases.* For small numbers of cases
                         (arbitrarily defined here as fewer than 30), use the Poisson distribution since birth
                         defects are considered to be rare events.

                         The easiest way to use the Poisson distribution is to refer to a table that provides
                         the upper and lower 95% confidence limits for an observed number of *cases*
                         (reproduced below for up to 29 cases). Then follow Steps 1 through 3.

**Table of 95% Confidence Limits for the Number of Cases, for 1-29 Cases,
Based on the Poisson Distribution**

| Number of Cases | 95% Confidence Limits | |
|---|---|---|
| | Lower | Upper |
| 0 | 0.0000 | 3.6889 |
| 1 | 0.0253 | 5.5716 |
| 2 | 0.2422 | 7.2247 |
| 3 | 0.6187 | 8.7673 |
| 4 | 1.0899 | 10.2416 |
| 5 | 1.6235 | 11.6683 |
| 6 | 2.2019 | 13.0595 |
| 7 | 2.8144 | 14.4227 |
| 8 | 3.4538 | 15.7632 |
| 9 | 4.1154 | 17.0848 |
| 10 | 4.7954 | 18.3904 |
| 11 | 5.4912 | 19.6820 |
| 12 | 6.2006 | 20.9616 |
| 13 | 6.9220 | 22.2304 |
| 14 | 7.6539 | 23.4896 |
| 15 | 8.3954 | 24.7402 |
| 16 | 9.1454 | 25.9830 |
| 17 | 9.9031 | 27.2186 |
| 18 | 10.6679 | 28.4478 |
| 19 | 11.4392 | 29.6709 |
| 20 | 12.2165 | 30.8884 |
| 21 | 12.9993 | 32.1007 |
| 22 | 13.7873 | 33.3083 |
| 23 | 14.5800 | 34.5113 |
| 24 | 15.3773 | 35.7101 |
| 25 | 16.1787 | 36.9049 |
| 26 | 16.9841 | 38.0960 |
| 27 | 17.7932 | 39.2836 |
| 28 | 18.6058 | 40.4678 |
| 29 | 19.4218 | 41.6488 |

Source:  Diem and Lentner, 1970.

**Step1.** Calculate prevalence.

the number of cases with birth defect A in an area and time period  x          10,000
the number of live births in that area and time period

**Step 2.** Look up the lower 95% confidence limit for the number of cases with birth defect A. Using this new number in the numerator, calculate the lower 95% confidence limit for prevalence:

Lower 95% CL for prevalence =     <u>lower 95% CL for cases  x     10,000</u>
                                                            number of live births

**Step 3.** Look up the upper 95% confidence limit for the number of cases with birth defect A. Using this new number in the numerator calculate the upper 95% confidence limit for prevalence.

Upper 95% CL for prevalence =     <u>upper 95% CL for cases  x    10,000</u>
                                                            number of live births

EXAMPLES using data from the California Birth Defect Monitoring Program 1983-1986.

| Birth Defect | Cases | | Number of Live Births[3] | Prevalence[6] | |
|---|---|---|---|---|---|
| | Number[1] | 95% CI[2] | | Value[4] | 95% CI[5] |
| Anophthalmia | 18 | 10.67 - 28.45 | 452,287 | 0.40 | 0.24 - 0.63 |
| Glaucoma | 27 | 17.79 - 39.29 | 452,287 | 0.60 | 0.39 - 0.87 |

Notes                                                                   Source:  Croen et al., 1990.
(1)  Number of cases ascertained from surveillance
(2)  95% confidence interval for that number of cases
(3)  Number of live births derived from vital records
(4)  Prevalence = [(1) / (3)] X 10,000
(5)  95% confidence interval for the prevalence
(6)  Prevalence expressed as cases per 10,000 live births

*For a prevalence based on a large number of cases.* For a large number of cases (arbitrarily defined here as 30 cases or more), use the normal distribution. Why? As the number of cases grows larger, the Poisson distribution approximates (i.e., looks more and more like) the normal distribution. The formulae below are approximations for calculating confidence intervals using the normal distribution (Rothman and Boice, 1982, p. 29, formula 19).

Shorthand: let   $c$ = number of cases

$b$ = number of live births

1. Calculate the lower confidence limit using the following:

$$\text{Lower 95\% CL for prevalence} = c \times \left(1 - \frac{1}{9c} - \frac{1.96}{3}\sqrt{\frac{1}{c}}\right)^3 / b \times 10000$$

2. Calculate the upper confidence limit using the following:

Upper 95% CL for

$$\text{prevalence} = (c+1) \times \left(1 - \frac{1}{9(c+1)} + \frac{1.96}{3}\sqrt{\frac{1}{(c+1)}}\right)^3 / b \times 10000$$

3. To determine the 90% confidence limits, replace 1.96 with 1.645. To determine 99% limits, replace 1.96 with 2.575.

4. To obtain confidence limits for the number of cases instead of the prevalence, apply the formulae but do not divide by births (*b*) or multiply by 10,000.

EXAMPLES using data from the California Birth Defect Monitoring Program, 1983-1986.

| Birth Defect | Cases | | Number of Live Births[3] | Prevalnce[6] | |
|---|---|---|---|---|---|
| | Number[1] | 95% CI[2] | | Value[4] | 95% CI[5] |
| Aortic stenosis | 73 | 57.22 - 91.79 | 452,287 | 1.61 | 1.27 - 2.03 |
| Cleft palate | 320 | 285.89 - 357.05 | 452,287 | 7.08 | 6.32 - 7.89 |

Notes                                                                    Source: Croen et al., 1990.
(1) Number of cases ascertained from surveillance
(2) 95% confidence interval for that number of cases
(3) Number of live births derived from vital records
(4) Prevalence = [(1) / (3)] X 10,000
(5) 95% confidence interval for the prevalence
(6) Prevalence expressed as cases per 10,000 live births

*Software support.* For a few prevalence values, confidence limits (and hence the resulting intervals) can be calculated by hand or using statistical software such as PEPI. One program in PEPI (POISSON) gives the table values for any number of cases; for a large number of cases it gives a normal approximation. PEPI software and documentation are available at:

http://sagebrushpress.com//pepibook.html

For many prevalence values, it is useful to write programs, for example in SAS, to calculate the confidence limits along with the prevalence.

## 8.6.2 Comparing Prevalence Values Using Confidence Intervals

The best way to compare prevalence values (e.g., for different maternal ages) is to do a statistical test; one type of statistical test is described in the second section below ("When both prevalence values are based on 30 or more events"). However, in the first section below we present a quick method recommended by the National Center for Health Statistics (NCHS) that works better when one of the prevalence values is based on fewer than 30 cases. Note that the NCHS-recommended method is conservative (i.e., there will be fewer statistically significant differences than would be found by actually performing a statistical test).

**When one of the prevalence values is based on fewer than 30 cases.** First compute the 95% confidence intervals for both prevalence values. Check to see if those intervals overlap. If they *do* overlap, the difference is not statistically significant at the 95% level. If they *do not* overlap, the difference is indeed 'statistically significant' or unlikely to be explained by chance alone. (Method recommended by NCHS in Ventura et al. [2000].)

EXAMPLE

The prevalence of holoprosencephaly among African-American women in Texas (2.39 cases per 10,000 live births) is over three times higher than among White women in Texas (0.78 cases per 10,000 live births). Is the difference statistically significant?  First compute the 95% confidence intervals.

**Holoprosencephaly Among Two Race/Ethnic Groups in Texas, 1996/97**

| Group | Cases | Live Births | Prevalence* | 95% CI for Prevalence |
|---|---|---|---|---|
| African American women | 7 | 29,254 | 2.39 | 0.96 - 4.93 |
| White women | 8 | 102,193 | 0.78 | 0.34 - 1.54 |

\* cases per 10,000 live births                                          Source:  Ethen and Case, 2000.

These two confidence intervals overlap. Thus, based on this approach, the difference between prevalence of holoprosencephaly in African-American women compared to White women is not statistically significant.

**When both prevalence values are based on 30 or more events.** This approach is based on calculating the confidence interval for the difference between the two prevalence values. If this interval includes 0.00, then the difference in the values is not considered to be statistically significant. Since this approach uses information from both prevalence values at the same time, it is more statistically powerful than the NCHS-recommended method. That is, if a difference truly exists, this approach will identify that more often than will the NCHS-recommended method. This approach uses the standard error for the difference between the two prevalence values (Rothman, 1986, p. 170, formulae 11-15).

Statistical software like the RATES2 program within the PEPI package can also be used to calculate this confidence interval.

Shorthand: let   $RD$ = higher prevalence - lower prevalence

   $c_1$ = number of cases used to calculate the first prevalence

   $c_2$ = number of cases used to calculate the second prevalence

   $b_1$ = number of live births used to calculate the first prevalence

   $b_2$ = number of live births used to calculate the second prevalence

1.  Calculate the lower confidence limit using the following:

$$\text{Lower 95\% CL for prevalence difference} = RD - \left(1.96\sqrt{\frac{c_1}{b_1^2} + \frac{c_2}{b_2^2}} \times 10000\right)$$

2.  Calculate the upper confidence limit using the following:

$$\text{Upper 95\% CL for prevalence difference} = RD + \left(1.96\sqrt{\frac{c_1}{b_1^2} + \frac{c_2}{b_2^2}} \times 10000\right)$$

3. To obtain 90% confidence limits, replace 1.96 with 1.645. To obtain 99% limits, replace 1.96 with 2.575.

4. If the confidence interval *does not include 0.00*, then the difference would occur by chance less than 5 times out of 100 (for 95% confidence intervals); i.e., the two prevalence values are significantly different.

EXAMPLE

Is there a statistically significant difference between African-American and Hispanic women in the prevalence of births with atrial septal defects?

**Atrial Septal Defect Among Two Race/Ethnic Groups in Texas, 1996/97**

| Group | Cases | Live Births | Prevalence * |
|---|---|---|---|
| African-American women | 133 | 29,254 | 45.46 |
| Hispanic women | 835 | 160,094 | 52.16 |

\* cases per 10,000 live births                                           Source: Ethen and Case, 2000.

1. The lower 95% confidence limit for the prevalence difference = -1.80.

2. The upper 95% confidence limit for the prevalence difference = 15.20.

3. The 95% confidence interval for the prevalence difference thus = -1.80 to 15.20.

4. The interval includes 0.00. Therefore, the difference between the two prevalence values is not statistically significant at the 95% confidence level; i.e., there is no statistically significant difference.

# 8.7  Issues to Consider If Data Reveal Unusual Patterns

It is very important to rule out relatively straightforward explanations for a change in the birth prevalence of a birth defect. Among the more common reasons are:

➢ Changes in medical diagnoses and technologies

➢ Changes in reporting that lead to changes in case ascertainment

➢ Changes in the population at risk (focus on age, period, and cohort effects)

➢ Random variation

Please refer to Kallen (1998, pp. 83-87) for a more extended discussion of the issues identified above.

The analytical capabilities of the surveillance system should support evaluation of the likelihood of these factors being responsible for observed changes in prevalence. Although the remainder of this section addresses "changes" in birth defect prevalence over time, it can also be applied to "differences" in birth defect prevalence between areas.

## 8.7.1  Changes in Medical Diagnoses and Technologies

To detect changes in medical diagnoses, it is important to compare isolated and multiple birth defects cases. Minor changes in the way a condition is diagnosed or reported can affect the coding and classification of specific birth defects. For example, the prevalence of neural tube defects among live-born infants may have declined during the 1980s from levels reported in the 1970s, due to the development and widespread availability of prenatal diagnostic tests, such as maternal serum alpha-fetoprotein screening and ultrasonography. The severity of spina bifida cases may be less today than in the 1970s due to the selective therapeutic termination of the more severe cases, which are more likely to be identified prenatally.

The birth prevalence of some disorders may increase due to new technologies. For example, fragile X syndrome, a chromosomal breakage disorder, is diagnosed much more often today than 10 years ago and was unknown 20 years ago.

## 8.7.2  Changes in Reporting and Case Ascertainment

Re-verification that the frequency is an un-duplicated count is also appropriate. Many suspected "clusters" reported by the media or concerned citizens involve multiple counting of the same cases. With a birth defects surveillance program that ascertains cases from multiple sources, it is important to ensure that each case is counted only once, even if reports are received from several health care providers or delivery settings. The same is true of changes in reporting. As hospitals shift to computerized diagnostic indices, reporting artifacts could decrease the numbers of cases of specific birth defects while increasing others. This is because a limited number of ICD-9-CM codes are retained in the index, and conditions that appear to be minor in the eyes of the medical records clerk may be omitted. If surveillance staff rely exclusively on the diagnostic indices to identify charts to abstract, some conditions may be missed. See also Chapter 5 on Classification and Coding and Chapter 6 on Case Ascertainment Methods.

## 8.7.3  Changes in the Population at Risk

Population characteristics can be controlled for by using stratum-specific prevalence, age-adjustment, and similar methods. However, as most statistical surveillance methods are based on the frequency of events rather than on proportions, rates, or prevalence, separate analyses will need to be conducted to rule out changes in the population at risk. Analysts who routinely examine birth defects surveillance data will have access to detailed, current population estimates and should examine the demographic and reproductive health characteristics of all women giving birth to identify changes in the population at risk.

## 8.7.4  Random Variation

It is also possible, and in fact very likely, that an observed difference in the frequency of a specific birth defect is due to random variability. With relatively low birth prevalence, cases of a particular condition will be quite rare, and the coincidence of two or more cases in space and/or in time may be just that: a coincidence. Confidence intervals are one way to address random variation.

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# *Chapter 9*

# *Data Management and Security*

# Table of Contents

# 9.1  Introduction

This chapter is designed to provide basic guidance on the mechanical and administrative aspects of establishing and operating a birth defects surveillance program, covering a range of topics related to the development of an efficient, effective, and secure program. This chapter is intended to serve as a guide to planning the development of a new surveillance program, as well as to the review of practices and procedures in place within existing programs.

Issues covered in this chapter include computer hardware and software, data capture procedures, transmission of data, data file management, personnel management, physical aspects of the surveillance program office and, finally, data confidentiality and security considerations. We discuss the functionality of a data processing system in Section 9.2, followed by more detailed discussions of hardware and software in Sections 9.3 and 9.4, respectively. Data management is introduced in Section 9.5 on process standards, while the specific topics of data entry (Section 9.6), record linkage (Section 9.7), and record consolidation (Section 9.8) are described in further detail in subsequent sections. The importance of ongoing communication with data sources to identify and correct emerging problems is discussed in Section 9.9, and in Section 9.10 we address physical security and confidentiality issues.

# 9.2 Functional Data Processing System Features

The functionality of the system for processing and managing surveillance data must be able to support all the necessary processes and activities required by a program. This chapter is intended to develop the key considerations and capabilities that are applicable to surveillance operations. There is a wide variation in case volume, approach to data collection, budgets, and goals of various birth defects surveillance programs. Overall mission, size, and scope will determine the best combination of procedures and features for a given program.

The basic operations of a birth defects surveillance system can be accomplished using minimal computer hardware, software, and systems. The characteristics outlined below provide a broad scope of useful features and capabilities.

**Computerized data collection.** As case reports on birth defects cases are received, data should be captured within an electronic database designed to maximize a program's ability to manage the surveillance system and utilize the resulting data.

A program's capacity to receive computerized data from reporting facilities and other sources can ease the burden of case reporting and reduce or eliminate the need for recapturing already automated data. Improving the efficiency of data collection can minimize effort in the reporting facility and at the surveillance program, while reducing errors often due to "re-automation." Increased efficiency can also improve relations with reporting facilities and support compliance with reporting requirements. Below we present critical considerations related to accepting and processing electronic reports from reporting facilities.

➢ *Reporting case data within electronic files, rather than paper reports,* requires the exchange of detailed information on data submission requirements and on the characteristics of the files provided. Design issues, such as file formats and structures, and coding schemes must be understood to ensure accurate data exchange.

➢ *Any limitations in the reporting facility's computer database must be identified* to ensure that submitted data can meet programmatic needs. Any shortcomings or incompatibilities between the facility's system and reporting requirements must be recognized and addressed. Examples of such concerns include facility data systems that are missing standard (required) data items or that code a given item using coding rules that are not entirely compatible with the program's coding schemes.

➢ *As the data systems used to generate the data files are revised, any effects that such changes may have on the submitted data should be identified.* Information of this type must be communicated by the reporting facility to the surveillance system.

➢ *Submitted data must be reviewed for quality control.* This review should compare the data submitted with source documents or files to validate that the data are being represented faithfully within the surveillance database. The review should identify any data distortions caused by differences in processing systems, coding structures or rules, and conversion routines used to build the extract file or to import the data into the surveillance program.

**Transmission of electronic data and data telecommunication.** Below we present considerations related to transmission of electronic data.

➤ If case reports are accepted as electronic files, a standard format, file structure, code structure and medium for submission, i.e., tape or disk, must be developed and documented.

➤ Programs receiving passive case reports may elect to accept data in formats and code structures that follow the reporting facility's database structure and rules. In this case the facility – whether a hospital, a diagnostic laboratory, or other facility – must be expected to refer to the standard for submissions and provide the data in a format and code structure that is compatible with and convertible into the standard format for reporting cases to the surveillance system.

➤ Whether converted or nonstandard data files are supplied, each facility must identify any compatibility/consistency problems apparent between the source system and the standard.

➤ Secure methods for delivery of forms and data files need to be recommended by the surveillance program and followed by each facility.

**Various modes for data entry.** Below we present considerations related to data entry modes.

➤ To automate the information received in the form of paper case abstracts, a data capture mechanism is required. Approaches to accomplish this task include:

- Classic data entry by keying data into a fixed format file

- Optical scanning

- Data capture through the use of custom screens

➤ Principles associated with the data entry process, which will ultimately enhance efficiency and data quality, include:

- Standardizing data review, query, and preparation procedures

- Verifying the keyed data

- Editing at the point of entry

- Editing of the completed input file

➤ In addition to capturing case data internally, consideration should be given to developing software that enable facilities to report case data electronically. Providing such software to the reporting facilities enables them to use standard file formats and coding and editing procedures for the data they submit. Editing the data at the point of entry, in particular, can reduce the need for later follow-back.

**Receipt and integration of new case data while retaining data on reporting activities.** Below we present considerations related to the integration of new case data.

➤ To the degree practical, the program needs to be able to receive submitted data for processing in the form of electronic files, paper submissions, and, potentially, through web-based or other direct data entry across secure connections.

➤ Integration of data into the system should be done such that integrity of the individual reports received is maintained. This allows the surveillance system to document the data source properly and to monitor reporting quality.

**Ability to link surveillance data to new reports and files from other data sources.** Below we present considerations related to linkage of surveillance data to new reports and files from other data sources.

> ➢ The program must have appropriate software and system computing capacity to screen incoming data against existing surveillance data in order to identify accurately duplications in reporting.

> ➢ The program must have the ability to implement a variety of strategies to link surveillance data with data from other sources in a manner that allows cases within other data sets to be identified as important. Through this means the program can augment the surveillance data base with information on cases identifiable through other data systems, as well as acquiring new case reports. This capability is essential if the surveillance program is to facilitate research studies.

**Ability to modify system easily and inexpensively**. The system used must provide flexibility with respect to systems modifications, edit specifications, and other data handling processes, as well as permitting modifications of code structure and data set variables within the database. When possible, programs should avoid developing the database using software systems that require considerable time and expertise to modify. It is best if modifications are controlled and can be made by birth defects surveillance staff.

**Ability to handle updates.** The system must provide an easy way to update registry data as new information on cases is received. Updates may include information on additional hospitalizations, further diagnostic work, or corrections to earlier reports.

**Editing data.** The system must support data editing at various steps throughout the data collection process. Data editing should be carried out as data are collected, processed, and incorporated into the program's database. Conducting edit checks as early as practical in the data collection process is an efficient way of improving data quality. Key stages for data editing include:

> ➢ At the point of data abstraction

> ➢ During data entry

> ➢ As files of new report data are prepared

> ➢ As case files are updated with new cases

> ➢ As additional data on known cases are added

Common edit procedures include field code range checks, table look-up of diagnostic and other codes, inter-field consistency checks, and editing across records for individual cases.

**Report preparation.** Staff must have the computer capability and training to conduct statistical analyses; to interpret the resulting statistical information; and to prepare text, tables, and graphics in the form of reports. Examples of statistical analyses include establishing basic case counts and rates, developing summary data on treatment information, reviewing prevalence trends, adjusting rates, calculating variance components and standard errors, and developing measures of the observed and expected prevalence of specific conditions.

**On-line case queries.** The database should be readily accessible to staff using different types of information to identify specific cases.

**Easy maintenance of reference tables/files.** The various reference files used to process and edit incoming reports and to develop statistical data on those reports must be easy to maintain and update. Such reference files may include tables of diagnostic and procedures codes, code groupings, geographic code dictionaries, hospital and laboratory code dictionaries, among others.

**Extracting files/subsets.** The capacity should exist to generate readily subset files of the surveillance data. This capacity should allow inclusion of data on cases selected using a variety of criteria and inclusion of specific variables for selected cases. File subsets often are needed for statistical analyses, quality control work, field site visits, and other uses.

**Quality control information on data sources, amounts, and quality.** To monitor case reporting timeliness and quality, the system will need to store sufficient information to support calculations of reporting timeliness and other data reporting quality measures. The system must allow for assessment of reporting quality overall and by reporting source.

**Systems security, administration, and backup.** The system must include features to protect data and programs from loss due to systems failure or user error and to maintain the confidentiality of patient and provider data. The computer system must provide a secure environment with security features designed and enabled to protect data from inappropriate access. Such measures must include a system of user name and passwords, along with a system to control the access of users to the computer server and drive locations where data are stored. These must be updated promptly with staff changes.

Program staff must be able to control or oversee these system administration activities. The system must also provide redundant back-up procedures to protect against system failure. This should include back-up and recovery procedures with regular and reliable copying of existing surveillance data and systems to tape or disk.

**Archiving of data and systems.** The surveillance data and the systems used to develop and maintain the data must be archived according to a predetermined schedule to protect against catastrophic loss. Archiving procedures must ensure appropriate preservation of submitted case abstracts and the routines used to process abstracts and to analyze case information. Archiving should also encompass statistical analyses, special studies, and the procedures used in those studies.

**Cost effectiveness.** The computer system and hardware used must be selected to fit both the needs and the budget of the surveillance program. The initial cost of the system and the cost to maintain and support both the operating computer system and specific programming requirements are critical considerations in selecting an appropriate system.

**Adequate performance.** The system selected must be responsive and provide adequate computing speed, disk storage, and working memory space to address the needs of the surveillance program.

# 9.3 Hardware

**Computer hardware.** Individual work stations and overall processing platforms should be selected to handle the work of the surveillance program and allow simultaneous on-line use of the data by multiple users and the various software packages used by the staff. Systems speed, number of concurrent users, active memory, disk capacity, robustness, and compatibility are all important considerations.

**Systems back-up hardware matched to size of system.** Systems back-up strategies must be complemented by hardware of sufficient size and speed to generate systems back-up on a prescribed schedule without eroding systems performance.

**Printers, printing capacity, and quality/variety.** Hardware that permits printing in the volume required by the program and that will produce high-quality printed tables, charts, and reports is important. Printing capabilities may be required for high-volume printing of envelopes or other specialized printing. If a surveillance program has a large-scale follow-back, the ability to print in-house materials and mailings that carry names and addresses significantly enhances security of the information at relatively low cost.

**Graphics and slide production capabilities.** Hardware that can be used to develop Microsoft PowerPoint presentations or slides is important. Slide makers and LCD projectors should be available to surveillance program staff.

**Communications hardware and links.** The staff of the birth defects surveillance program must be able to send and receive e-mail and to access the Internet. Data collection through hospitals and use of data by staff must also be supported by appropriate computer communication systems.

**Strategy for planned obsolescence.** The hardware used by the surveillance program must be able to operate software and systems that are actively supported by the software or systems suppliers. Planning for replacement of existing hardware should be an ongoing process. This should ensure keeping pace with changing software and systems requirements, enabling staff to manage the surveillance database effectively.

# 9.4 Software

The basic software selected to run the surveillance database must provide the features required to meet programmatic needs. It must have the capacity and robustness to conduct required procedures, be compatible with other similar data management systems, and be supportable.

**Data analysis software.** Standard statistical software should be available to analyze surveillance data. Statistical packages must have a full range of capabilities for developing standard statistical tables (including counts and rates) and conducting more complex analyses (such as standard error calculations or observed-to-expected ratio estimates). In addition, the software must support the design of tables, as well as presentation features such as titles, footnotes, graphics and, potentially, mapping. (See also Chapter 8 on Statistical Methods).

**Record locking and file locking.** The data management system must provide for data security and confidentiality as well. Systems should be considered with confidentiality and security features that enhance the proper protection of data. Depending upon the types of direct database access various system users are permitted, the data management software may need to control data access at the level of the file, the record, and the individual variables. This may require various levels of file access, which could be handled by using data management software with these capabilities.

**File security software.** Software that can regulate access to file servers and to specific computer drives or computer files, and maintain various levels of file access rights, is essential for storage of data on a Local Area Network (LAN)-based or mainframe system. Staff should either manage the administrative features within this software, or these activities should be under their direct supervision.

**Multi-user capability.** Software used to access and manipulate the data may need to have multi-user capabilities, allowing access by multiple users during most, if not all, file management routines. The need for this capability will depend on staff size and the scope of the surveillance program.

**Integrated and stand-alone utility programs.** The database must be accessible to program staff. Software and skills needed to develop ad hoc and specialty software routines must be available, as necessary, to manage and maintain files or to conduct specialized analyses. Such custom routines may be required on an ad hoc or a routine basis.

**Record linkage software.** Software is needed that supports data linkage. This capability is essential to de-duplicate new report data and to link cases to corollary files, such as birth or death files. Record linkage capabilities are also essential to the conduct of cohort studies that can link cases to files of study subjects. The ability to link data, with a high degree of accuracy, is critical to data quality, to conducting basic surveillance functions, and to research. Some states have developed their own custom – designed programs to meet record linkage requirements (e.g., Colorado).

Linkage capability can take several forms, ranging from on-line case-by-case queries to electronic comparisons of large databases. Data can be linked electronically through either pre-programmed routines or ad hoc routines and can be based on deterministic or probabilistic linkage procedures. The specific strategy and approach used by a surveillance program will depend on its size, overall mission, and resources available.

➤ ***Deterministic record linkage*** procedures, which involves the literal comparison of fields or columns within fields for exact matches, can be developed relatively easily and can be supported by most database management software. If this approach is used, it is essential to audit and refine the procedure painstakingly to ensure a high degree of matching accuracy.

➤ ***Probabilistic linkage*** bases record linkage decisions on determined probabilities that two records are likely matches. This technique generally is accepted as quite reliable when applied appropriately. However, it is dependent upon costly proprietary software packages that may not interface well with other data systems used by the surveillance program.

Regardless of the approach used, the results obtained through record linkage must be reviewed periodically for quality. The presence of unidentified duplicates within the case data and combining report data for different children into a single record are two obvious hazards of improper linkage. These false positive and false negative rates must be minimized by reviewing linkage quality regularly.

# 9.5  Process Standards

In this section we discuss the following aspects of process standards: inputs into the surveillance system (Section 9.5.1), instructions for reporting facilities on proper submission of data (9.5.2), procedures for initial review and query of submitted data (Section 9.5.3), procedures for receipt and logging of shipments (Section 9.5.4), and forms and batch control procedures (Section 9.5.5).

## 9.5.1  Inputs

Proper management of data within the surveillance program needs to begin through careful coordination with those providing the data and through following appropriate internal practices and procedures. These must be designed to promote accurate reporting and complete processing, ensuring a trackable system where processed data can be re-traced back through to the data originally submitted.

The procedures developed need to accommodate the various forms in which data are reported and the sources from which the data are derived. Data coming in to a surveillance program can vary widely with respect to the way they are transmitted and their content. This is true across states and within a state. The data can be provided in the following ways:

- Paper/electronic abstracts for reportable conditions
- Hospital discharge data
- Medicaid data
- Early intervention program data
- Data on services to children with special needs
- Birth and death record data
- Medical examiners reports

## 9.5.2  Instructions on Proper Submission of Data

Clear and concise instructions must be developed and distributed to all those involved in reporting cases to the surveillance program. Necessary components of these instructions include:

- Precise definitions of what constitutes a reportable condition/case
- Item-by-item explanation of information to be reported
- Timelines for reporting
- Acceptable reporting methods – paper, electronic, File Transfer Protocol (FTP)
- How and where to ship reports
- Procedures recommended to ensure secure shipment
- Procedures for handling corrections and updates to previous reports
- Sample of abstract
- Detail on electronic submissions, if appropriate
- Definition of terms, as appropriate

> ➢ Name of a contact person in case of questions

These instructions should be readily available to those with a 'need to know', should be prepared to minimize any anticipated potential misunderstandings, and should be updated routinely. Instructions need to be customized and targeted to specific data sources – such as laboratories, hospitals, physicians, or medical examiners – to reflect differences in what is expected from each.

Complete documentation of the receipt and preparation of case data from internal sources is also required. Examples include information from data systems for programs that provide specific information on children with reportable conditions. The nature of the data system and the schedule for providing or obtaining data, the format and technical specifications of the data all need to be documented. This is necessary to ensure coordination between the birth defects surveillance program and other data systems and/or sources.

## 9.5.3  Initial Review and Query

As reports are received, and during the intake process, it is important to establish procedures to screen incoming reports and data. These screens should be designed to avoid unnecessary work and to identify and resolve quickly any gross problems with the submission. These screening activities might result in a submission being returned and not processed. Other types of screening may occur at various points within intake processing of reports.

For paper reports, potential screens could include very basic things, such as examining the mailing for physical damage, proper addressing or, perhaps, tampering. Paper reports could be screened, as they are inventoried, to be sure they are completed adequately and that the case is truly reportable. If the paper report is primarily a case-finding tool, it might first be screened against cases in the database to determine whether it is new.

Pre-screening of data submissions should include checking for possible computer viruses, determining whether the file is readable and in an appropriate format and file structure, and establishing whether the count of records within the file is correct.

## 9.5.4  Receipt and Logging of Shipments

As data are received in the form of paper reports or automated files, forms- and data-control procedures need to be followed. Procedures should be designed to ensure that all data submissions are processed. They must also provide a mechanism for rechecking the status of the surveillance program database to validate that all information has been processed properly and appropriately. This can be accomplished by developing a log to record receipt and processing of reports by facility. Such a log could contain basic information about each submission. This might include: date received, reporting facility, number of reports, date span for the reports, format of the reports, date prepared, report numbers assigned to the batch, file or batch name assigned to the data, and the date data processing was completed. Maintenance of a log serves as a control point for the data. It can also be designed to permit monitoring of the reporting status of individual facilities.

Depending upon the type of report being processed, other approaches may be appropriate to ensure completeness. For example, data that serve primarily for case finding may need to be screened first against the surveillance program's database to determine whether the report is for a new case, thereby permitting previously reported cases to be quickly dropped from the case-finding data.

## 9.5.5  Forms and Batch Control Procedures

For data control and tracking, it is important to use a systematic procedure to inventory and to identify unique reports. Classically this is accomplished for paper reports through use of a series of sequential numbers, with each form assigned a unique number. Forms are then organized sequentially into batches of manageable size. A similar procedure can be used for automated data submissions to assign each electronic record a unique identifier and to maintain a record of the file name assigned to each batch of reports submitted.

These procedures allow staff to locate a specific report easily and provide a mechanism for data inventory. Missing report numbers can be listed and resolved as report file completeness is evaluated. Data edits for each report can reference these numbers to identify and resolve any concerns with the data in that report.

# 9.6 Data Entry

The most basic aspect of developing a surveillance database is preparing an electronic file of reported information. While the proportion of paper case reports received varies widely across surveillance programs, each program must have reliable mechanisms for entering data from manual reports into an electronic file. In addition, many programs can provide reporting sources with software that can be used for submitting cases. Applying some simple concepts to the automation of information can help provide data files of consistent quality.

**Interactive edits.** Developing a process for capturing data destined to reach the surveillance provides an opportunity to build functional editing of entries into the operating procedures. This is especially effective when edits are used to question incoming data at the point where the patient's chart is available for review. Editing data at the point of origin is the most efficient method to ensure high quality.

Interactive edits can be very simple checks, such as ensuring that only numbers are entered into a numeric field, ensuring a date entered is a valid date, or preventing a required field from being left blank. More complex edits might involve providing links to a database of valid codes for diagnoses or procedures, editing for consistency across fields, or screening each case to determine if the child was reported previously.

In designing and developing interactive editing procedures, it is important that the objectives be kept in focus. A process is needed for producing high-quality electronic data efficiently and effectively. Interactive editing needs to be functional. It should be designed to screen for impossible or improbable entries. It must also be efficient, providing the operator with a clear explanation of the perceived problem and a ready mechanism for resolving the issue.

**Verification.** Verification procedures are another tool for controlling the quality of incoming data. The key data processing steps of information coding and the actual process of data entry are candidates for verification. Verifying data is an old and time-tested method of monitoring and controlling errors introduced into data through data processing procedures. These practices do not improve the quality of the reported data, but they do minimize degradation in data quality during data processing. The purest example of data entry verification is blindly re-keying previously entered data using software that compares the newly keyed data, key stroke by key stroke, to that entered earlier. Any discrepancies are identified and resolved by the verifier.

To verify data is to double check the data to ensure it is captured accurately. Verification procedures can take two basic forms, namely, independent and dependent verification. There are also two basic strategies relative to the scope of verification: verifying each incoming case or verifying a sample of cases. In *independent verification*, the verifier is not provided with the previous work and must essentially redo the work. The two versions are then compared and any discrepancies resolved. For *dependent verification*, the verifier has access to the original work and reviews the entered data, comparing it to the source document; in the case of data entry, essentially proof reading the work.

Focusing on verification as a tool for efficiently developing data files of consistent quality, verification can be developed incorporating the quality and skill of the processing staff with efficient methods for screening and resolving processing errors. As an example, dependent verification of all the diagnostic coding done by new staff might be done by experienced staff and continued until a "qualified" level of accuracy is consistently demonstrated. Once the new staff member has qualified, only sample independent verification might be done.

Information obtained through verification can provide important insights into staff training needs. These results can also ensure a consistency of understanding and interpretation across staff involved in data preparation, highlighting any inconsistencies.

**Forms/record and batch controls.** Since data arrive in a variety of forms and from numerous sources throughout the year, effective methods to inventory all incoming data are important. As a corollary to logging the receipt of data shipments, control of individual records is very important.

As reports – both paper and electronic – are received and early on in their processing, a report number needs to be assigned to each report to serve as its unique identifier. This identifier provides a ready mechanism to inventory the incoming reports and, later, the consolidated files of processed reports. This report identifier also enhances coordination of the work during later stages of file editing and processing.

There are a variety of schemes for assigning a report identifier. The most basic is a sequential number that begins with the year the report was received followed by a simple sequential number. By including record type/source codes within the prefix for the sequential number, the type of report or information source can be incorporated into this identifier. Such information is often useful in developing management information regarding database status.

A system for numbering data entry work files needs to be developed and employed to properly control and inventory work batches. Each work batch needs to be assigned a unique batch identifier. A log should be established to record the report identifier numbers within each batch. The log should include the date completed, the individual completing the batch, the individual verifying the batch, and the date the batch was processed into the surveillance program. This information will aid in assuring all reports are processed and in tracking down any discrepancies. Information in the log will help assess processing issues, such as timeliness and staff accountability.

**Procedures appropriate for a variety of data inputs.** It is important to map out the proper handling and intermeshing of data from each data source carefully to ensure data quality. As mentioned earlier, sources of information can vary widely, both in type and quality of data. In designing the data entry process, the form in which the incoming data are presented can create a need for customized procedures.

Tailoring the procedures to match the data source and data format can add to efficiency and enhance final data quality. These adjustments might take many forms, including facilitating data entry through customized data entry screens for certain report types. Specialized editing to match the data source and, perhaps, to screen for code conversion errors may be required. Some data sources might be considered primarily as sources of case ascertainment. The first processing step might be screening cases against the program's database to determine if the case has been reported previously.

**Training/certification and instruction for data preparation.** Program staff members involved in data collection and processing must have the skills required to accomplish their work accurately. The skills required vary across key functional activities, namely abstracting case data, coding the information, and entering the data. Data management and editing routines will not correct data quality problems that occur if staff members are not properly trained.

Surveillance programs need to have a strategy for training new staff that allows them to learn the new job; measures their understanding of the work; provides feedback on problems and progress; and determines, in some objective way, that the new staff member's work has reached an acceptable level of quality. Staff skills and the rigor with which work is reviewed will vary among surveillance programs. Whether a surveillance program utilizes active or passive case ascertainment influences the skills needed. Hiring staff with training and experience in health information management may prove important. By the very

nature of birth defects surveillance, there will always be a need to train new staff in a number of areas that are unique to the program and where it is not possible to hire experienced staff. There must be a strategy to ensure that staff assigned to a task have the skills the task requires.

As a component of continuous training, detailed manuals are needed that document procedures and serve as a reference source for staff. Staff should be encouraged to refer to these manuals and to identify errors, inconsistencies, and misinterpreted sections. Updating these guides periodically ensures that the manuals/instructions remain functional and current and able to serve as training guides for new staff.

Future editions of *The Surveillance Guidelines* will address training issues for surveillance programs in greater depth.

**Input file processing functions.** The management of data quality within electronic birth defects data files is important as well. The procedures and processes for handling the quality of processed electronic data are similar to those used for paper reports. The tools available to a birth defects surveillance program are basic data processing and management practices that are not unique to these types of data. Electronic data files readily lend themselves to editing and clean-up. Standard computer routines can be used to screen files for obvious errors or inconsistencies, to spot problems with the data efficiently, to summarize findings, and to organize results in ways that allow the efficient correction of any errors.

Key components of input batch processing are outlined to provide an inventory of the tools available for functional data quality control. The combination of practices employed by a given surveillance program needs to match the methods and procedures used for data file development.

**Editing.** Development of data editing procedures is a standard activity in any database development effort. As with interactive editing during data entry, computer routines can be developed to identify a variety of data problems. Standard edits often include:

> ➢ Field range checks
>
> ➢ Report number range checks to identify missing records
>
> ➢ Inter-item consistency checks
>
> ➢ Field validity checks
>
> ➢ Code validation through table look-up, i.e., diagnostic or procedures code tables
>
> ➢ Consistency across multiple reports for the same case
>
> ➢ Hard versus soft edits and use of edit flags

The organization of the results from edits requires the same care in design as do the edit criteria. The results of an edit run need to be organized to make error resolution and file correction as efficient as practical.

**Tracking information.** As potential problems with data are identified, it may be necessary to ask the reporting source for clarification or for additional information. A basic procedure to monitor outstanding requests for clarification or correction should be used.

**Printed case abstract.** In conjunction with efforts to correspond with staff at the data source about reports, a ready mechanism to print an abstract of a report can improve the effectiveness of communications and may enable correction of other errors in a report that cannot be identified by the edit routines. The capability to print a case summary easily can prove useful for multiple purposes.

**Error correction.** It is important to have effective and efficient procedures for error correction. Reports that identify edit exceptions can be linked to the edit results to pull up rapidly, or to *queue*, the records needing attention. A well-designed process can minimize the potential for introduction of errors in the course of record correction.

**Case-by-case and multi-record correction.** Mechanisms to correct records one at a time are important. The capability to update multiple records simultaneously can also be useful. When used judiciously, multi-record correction can save time and reduce the potential for error.

**Add/delete.** The capability to delete spurious or redundant records can prove to be very useful.

**Linkage and assignment of case identifier.** As input files are processed and screened for duplication, a system for uniquely identifying each case is necessary. While a program may choose to number and retain all reports received, it is critical that a specific child's reports all have the same case identifier. This is necessary for record and file linkage. In a program where data are consolidated immediately, an identifier for each child is still a critical component of the system. A mechanism for assigning identifiers to newly reported cases is necessary. In the case of electronic submissions, this process should be automated.

**Facility reports.** Summaries of data quality relative to screening and editing of incoming reports is important for maintaining an accurate picture of the quality of submitted data. Summary reports that permit the tracking of report quality over time and across facilities can be designed. Such information is very useful in identifying facilities that are candidates for data quality reviews and/or in-service training. These reports can complement efforts to work with facility staff to correct any persistent problems.

# 9.7  Record Linkage

The proper operation of any birth defects surveillance program depends on developing and following procedures for efficient and effective record linkage. These procedures should be developed carefully. The accuracy of the procedures used to link individual case reports needs to be measured and monitored. Instances of the same child being in the database as different children and different children being presumed to be the same child must be estimated. Developing and monitoring linkage procedures carefully is as crucial for programs that manually search for potential matches as it is for those that use electronic linkage.

Not only is it important to link incoming reports accurately to the historic file to locate previously reported cases, but the ability to link to other databases is also essential. Procedures need to be tailored and evaluated specifically for each type of linkage. Goals for linkage completeness that reflect these expectations need to be established.

Linking birth defects case data with files from other sources may be done to meet a number of objectives. These include:

> ➢ Deleting duplicate data
>
> ➢ Case-finding
>
> ➢ Augmenting the information available for a case
>
> ➢ Conducting special studies or program evaluations

The level of precision and efficiency that can be expected from a matching process are functions of several factors. Key among these are:

> ➢ Quality of data within the files to be linked
>
> ➢ Number of fields common to both databases
>
> ➢ Logic employed to compare the files
>
> ➢ Time available to review and assess each link

The matching strategy developed should maximize the results and minimize the resources employed to obtain those results. Estimating the level of precision for any linkage procedure can be used to assess the advisability of revision. These estimates are also important for evaluating the suitability of using the linked data for specific purposes.

With respect to assembling the required data on each case, linkages to birth certificate files and death certificate files are extremely important. These sources can provide the surveillance program with valuable information on each case. For example, linkage to the birth certificate file has the added benefit of identifying reports for a single child that may not have been linked properly during the processing of incoming data. Linkage to birth and death records can also provide the ability to track changes to a child's name over time. This can assist in collating data on a single child that might otherwise be treated as distinct cases. In some jurisdictions, access to this kind of information will depend on legally prescribed restrictions.

# 9.8  Record Consolidation

When multiple reports are received on the same case, differences can be expected in some of the information across reports. By developing a summary of the information on each case, consolidating the information across reports into a single summary, the information about the case can be enhanced. A number of issues must be addressed in any information consolidation effort. The categories of information that could be consolidated or summarized, and the key issues relative to summarization, include (1) demographics and identifiers and (2) diagnostics.

**Demographics and identifiers.** Most demographic items are constants and do not change with the age of the child. These include date of birth, race or ancestry, mother's age. Updating missing data fields using data from subsequent reports is generally appropriate. Conflicts across reports for these fields can be difficult to resolve, but may be predicated on the source of the data, prioritizing data from specific files or facilities. Changes can be expected to occur over time in identifier fields such as name, parents' names, and address. Selection of the appropriate data to be included in a summary needs to be based on the purpose of the summary. For example, data for referral or outreach efforts need to be current, while data for auditing birthing hospital records should represent information at birth.

**Diagnostics.** As multiple reports for a child are received, collecting diagnostic information across all reports can result in significant redundancy. When the same diagnoses are reported repeatedly, this redundancy is simple to manage. As diagnostic data change across reports, there are three possible causes. Each of these raises specific issues relative to proper management:

➢ *New conditions being diagnosed.* Newly diagnosed conditions clearly must be included in any summary for the child.

➢ *Previously diagnosed conditions reported with greater or lesser specificity.* Redundancies in diagnostic codes caused by differences in specificity can be problematic. In the absence of accepted guidelines for doing so, to eliminate redundancies (increasingly specific diagnostic codes) using intuitive logic can be problematic. The logic must be thought through clearly, with the intended use of the resulting summary in mind.

➢ *Actual changes to a previous diagnosis.* Changing a diagnosis can reflect a revision based on better information or an alternative diagnosis, for example, a difference of opinion. A key problem with such changes is the need to differentiate a changed diagnosis from a condition that has been newly diagnosed. Having an effective mechanism in place for facilities to report corrections to diagnostic information can help reduce confusion in interpreting subsequent reports. Making changes to diagnostic data should be done carefully and through close coordination with facility staff. Some programs may prefer not to change original information, but rather flag it as inaccurate or no longer valid. In this way, the integrity of the database remains intact: inaccurate information is not "counted", data quality evaluations can be conducted, and new or confirmed information is accepted. This is especially useful when comparing reported information with the results of medical records review by surveillance staff.

**Procedures data.** Redundancy can be expected in data reported for procedures, since it is not uncommon for a child to have some treatment or corrective procedures performed multiple times. Therefore, such data can be consolidated reliably only if the date each procedure was performed is reported, or available through medical record review, to identify specific procedures by date received.

# 9.9  Feedback to Data Sources/Abstractors

When data problems are identified during report processing, it is important to communicate those problems to staff at the data source. Facilities and/or individuals providing data should be interested in learning of problems the surveillance system encounters with respect to the completeness and accuracy of their data. With passive reporting systems, communicating errors, resolving inconsistencies, and reviewing apparent discrepancies represent effective feedback mechanisms. An efficient mechanism is needed to provide feedback on problems, although the necessary corrections to the information may seem obvious to surveillance program staff. Staff of the reporting facility will benefit from the feedback and may need to correct the information within their own records.

# 9.10  Security

Many of the data assembled by a birth defects surveillance program are extremely sensitive. For this reason, a program must initiate and maintain a comprehensive strategy for data security that ensures data are protected from improper access or inappropriate use. Developing a security plan that establishes and demonstrates a commitment to data protection is essential in reaching the program's long-term goals and objectives.

## 9.10.1  Personnel Issues

Four aspects of security management fall under the category of personnel issues.

- ➤ *Hiring practices.* Attention needs to be paid in selecting new staff members to screening candidates to ensure they can be relied upon to handle confidential data appropriately. A work history that includes responsibly handling confidential data is an example of desirable experience. It is important to request and check references for all prospective employees. If possible, security background checks should be conducted prior to making a hiring decision.

- ➤ *Written procedures on security and access.* New employees need to be informed clearly of the procedures regarding appropriate access to and use of data, particularly any files that include personal identifiers. Written materials that describe the nature of the data and the rules and policies relative to data handling must be reviewed with employees. These materials must cover all aspects of employees' actions for which they are accountable. These materials need to be discussed with each employee to ensure that the employee has every opportunity to ask questions so that they understand the policies explicitly. A written policy on the release of identifiable or potentially identifiable data must be included. It is essential that all aspects of data release be identified within the policy, along with who has the authority to authorize a release. Such a policy must include the "business activities" of returning data diskettes and corresponding on data editing problems with data providers, as well as release of identifiable data for research use or in conjunction with child-find referral activities.

- ➤ *Security and confidentiality agreement/oath.* Each employee who has access to the program's data must sign a confidentiality pledge. The pledge should be in the form of a comprehensive statement that outlines the confidentiality policy in broad terms. In addition, this document should include a statement that the employee understands the confidentiality policies and the potential consequences for violating these policies. Finally, the document must include an oath on the part of the employee that they will abide by these policies. As significant changes to the confidentiality policies are made, each employee must sign a new pledge that reflects the new policies.

- ➤ *Disciplinary policy.* Whenever there is an allegation of mishandling confidential data or where unauthorized access is suspected, the incident must be investigated. Such an investigation must be conducted carefully and in a manner consistent with existing employment laws and personnel practices. Appropriate disciplinary action must be taken if it is established that an employee has violated the confidentiality policy. Any deliberate violation of policy that results in the inappropriate release of confidential data should be grounds for dismissal and for potential criminal action, depending upon the law governing these data.

# 9.10.2 Transportation and Information Handling

Basic security concepts that should be considered relative to shipping and handling information are listed in this section. Standard office practices and procedures for handling materials that include confidential data need to be developed and followed. These are necessary to avoid problems due to inappropriate or inadvertent access to these data.

The privacy regulations developed as part of the Health Insurance Portability and Accountability Act, or HIPAA, place significant responsibilities on hospitals, physicians, and others to properly safeguard confidential data on their patients. These regulations place strict procedural standards on health care facilities, heightening concerns about patient privacy held by health care facilities and providers. It is important to adopt information exchange practices with data sources that do not create a potential liability under the provisions of HIPAA (see Chapter 2 on Legislation). For example, common and efficient methods for exchanging information, such as fax or e-mail, need to be avoided or used with great attention to appropriate security. This is because faxed images can be intercepted and printed, can be inadvertently sent to the wrong fax or to a fax that is unattended or otherwise not secure. E-mail shares all these problems in addition to the fact that e-mailed materials will become part of the e-mail back-up systems and copies of sensitive materials will become interspersed with other documents that may well be public information. The existence of these back-up files means a loss of control over the data and access to the data. The problem of potentially intercepting e-mail only compounds this problem.

With these thoughts in mind, key considerations relative to good data handling and transporting practices are provided below.

> ➤ *Instructions to data sources for addressing and shipping of incoming reports and information.* All facilities and individuals who ship abstracts or data to the surveillance program must be provided with current and precise address information. Data sources should be encouraged to ship data in a secure manner where chain of custody signatures are required, such as certified mail or FedEx. If a shipment is received that was misaddressed, the data source reporting the cases should be contacted promptly by telephone, with a follow-up letter, and be advised of the correct addressing of shipments. In addition, standard practice should include prompt acknowledgement of shipment receipt. As staff at the data source learn to expect an acknowledgement of each data shipment, failure to receive an acknowledgement will alert them to the possibility that a shipment has been lost or delayed.

> ➤ *Use of fax or e-mail for forwarding or receiving sensitive data is not advisable.* These methods of transmission should not be considered secure unless the sensitive information is encrypted and password protected. Fax machines that both send and receive such materials need to be attended during transmission.

> ➤ *Managing the work station.* Employees need to be trained to manage their desktops. Confidential materials should not be on a desk if they are not being used actively and should not be left unattended during breaks or lunch periods. Similarly, passwords should be required for staff access to any personal computer that holds confidential data or that allows access to confidential data through a network or other computer connection. Such equipment should not be left unattended with connections in place that would permit unauthorized access. Care must be used in displaying confidential data on the computer monitor in order to ensure that persons who do not have authorized access cannot read them. All materials must be filed properly and locked when not in use. Following these common sense practices reduces the possibility of inappropriate access and should be applied conscientiously to the desk, the personal computer, and the files assigned to each staff member.

➤ *Physical access to abstracts, other documents.* Limiting direct physical access to files and other materials that include confidential data is a basic step in reducing the likelihood of someone seeing information to which they are not privy. The design and layout of offices can be done in a way that enhances staff members' ability to carry out their responsibilities without exposing confidential material to others. Careful planning in this regard, combined with good desktop management practice, will minimize inadvertent access.

➤ *Procedures and furnishings to lock up documents and diskettes.* Employees must be provided with the office furnishings needed to adequately secure documents. Locking desks and locking file cabinets are essential, with thought given to assignment and management of keys for these locks and the organized storage of extra keys.

➤ *Procedures for shipping reports and information from the program.* Program staff need to follow secure practices, as when they send any confidential materials to data sources. It is essential that such shipments are addressed properly, and the address should be confirmed if there is any doubt about its correctness. Materials should be shipped using a method/carrier that obtains a signature to verify receipt. Confidential data should not be shipped through e-mail or FTP unless the security of the connection is ensured or an adequate encryption technique is used to disguise the data.

➤ *Shredding and destruction.* Considerable care must be taken to avoid any potential for disclosure of data when confidential material is discarded. Employees must be conscious of the need for care when discarding any program-related materials that include identifiers or that would be considered confidential. Computer listings, correspondence, and other materials need to be screened to be sure that confidential data are handled appropriately. Staff should be provided with access to a shredder, and paper abstracts or printouts with confidential data should be shredded promptly. Any large volumes of confidential materials that need to be disposed of must be destroyed in a secure way.

Similar standard precautions must be established for computer storage devices. Diskettes should be reformatted, rather than simply deleting files. As hard drives on personal computers are replaced, the old drive must be reformatted or any data remnants otherwise destroyed, for example, by storing them between strong magnets for a period of time.

➤ *Transportation of data.* When staff members are in the field, all confidential data must be carefully safeguarded. Documents should be transported in locked brief cases or otherwise protected. The security of portable computers must be ensured. Confidential materials must be kept locked in the vehicle trunk while traveling. During overnight stays these materials should be removed from the vehicle and placed in a hotel room rather than left in a vehicle overnight.

## 9.10.3  Physical Security

Physical features of the worksite can enhance information security significantly. There are two specific ways the facility housing a surveillance program can maximize security:

➤ *Restrict physical access to the work area.* To the degree possible, access to the surveillance program's work area should be controlled. Ideally, it should be isolated with a card entry access system. Reducing or eliminating travel into and through the work area translates directly into reducing or eliminating opportunity for inappropriate data access.

➤ *After-hours security.* At a minimum, the office area must be locked securely after working hours. Ideally, the office area should be protected against unauthorized access through use of an alarm system that includes motion detectors and that is monitored centrally and continuously. If possible, no janitorial services should be carried on after hours.

Periodic maintenance work should generally not be conducted after hours unless surveillance staff are alerted and have an opportunity to take any and all extra precautions to ensure appropriate security of the data.

## 9.10.4  Computer Security

Proper data security requires a comprehensive approach to computer security. A number of key aspects to any plan designed to protect electronic data files are listed below.

➢ *User ID and password.* A system for unique user IDs and passwords is a cornerstone of computer network security. Staff should not be allowed to share ID and password information. Departing employees must be deleted from the system promptly. Periodic outdating and changing of passwords should be standard. Employees need to understand the importance of these activities and know that their personal login is critical to protect. This is because activities on the system will be traceable to the user's ID and password.

➢ *Virus scan – current.* In receiving electronic data, it is essential that diskettes and other electronic files be scanned for viruses prior to loading onto the personal computer or the network. A comprehensive and continuously updated virus scanning package should be used for this purpose.

➢ *Control of user access to data.* Careful management of user rights to the network or other computer system can significantly enhance data security. It is important to minimize to the extent possible unnecessary access to files. Steps must be taken to decrease or eliminate both potential misuse of data and inadvertent damage or destruction of data that are accessed inappropriately. Planning the architecture for data storage can complement limiting access to the various data files and greatly enhance security in the process. Much like user IDs and passwords, this level of security must be continuously maintained, with access modified as staff work assignments change over time.

➢ *Discarding of old personal computers, hard drives.* There are a number of special considerations regarding the security of electronic files. Simply deleting a file from a hard drive or diskette does not actually erase the data. This problem is not always properly addressed as old computer equipment is swapped out or discarded. There must be procedures developed for disposal of computer storage devices that ensure none of the data are recoverable.

## 9.10.5  Policy on Release of Data

Written procedures must be established that describe the proper mechanisms for release of information from the surveillance program. Written procedures are necessary to provide surveillance staff with a clear understanding of proper data handling and release. The process for obtaining approval for access to the data, and authorization for release of the information, must be described in detail. There must be no confusion among the staff on this critical topic.

Confidential data release procedures should include the specific practices required for proper preparation of tabular statistical data, as well as de-identified micro data files. These micro data files must be designed to guard against inadvertent disclosure of confidential data. The procedures must delineate clearly the approval process that governs and regulates release of identifiable or potentially identifiable data. Issues related to sending identifiable information to data sources and to other sources of information about cases of birth defects must be covered. Providing access to confidential information for research purposes must be discussed, describing the types of research projects that may gain access to these data and the system's process for reviewing and approving such projects. Finally, the conditions under which the information can be used for administrative purposes must be covered. This should include using the data to ensure that children and families are referred appropriately for needed services, if this is part of the surveillance system's objectives.

# Chapter 10

# Data Collaboration and Dissemination through the NBDPN

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# 10.1 Data Collaboration and Dissemination through the NBDPN

With the support and collaboration of the Centers for Disease Control and Prevention, the National Birth Defects Prevention Network (NBDPN) collects, analyzes, and disseminates state- and population-based birth defects surveillance data. In this way, the NBDPN plays an active role in turning data from throughout the US into useful information and encourages the use of birth defect data for decisions regarding health services planning, such as secondary disabilities prevention and referral to services.

The NBDPN State Data Committee coordinates the procedures and processes required in this effort. One of this committee's collaborative activities is the publication of an annual report of birth defects programs. The report includes detailed descriptions of the individual state birth defects surveillance programs, tables of data submitted by the participating states, and selected data analyses. The report includes diagnoses of interest, information regarding the format for submitting data for the annual report, and the criteria for the state directory that describes each birth defects surveillance program.

The NBDPN State Data Committee also discusses issues related to data suppression, confidence intervals, statistical analysis, and presentation of data, which are often of concern to the states. Because of the committee members' technical expertise in working with state data, the committee also has a role in evaluating the feasibility and merits of NBDPN participation in other data projects. Any use of data aggregated under the auspices of the NBDPN, including data projects and ad hoc studies, must be approved by the NBDPN Executive Committee.

All decisions regarding privacy, security, and confidentiality issues related to releasing or submitting surveillance data from individual programs to the NBDPN are handled at the state level.

Interested parties are invited to refer to the NBDPN website (at http://www.nbdpn.org) for contact information relating to the State Data Committee and for instructions on how to submit data to the NBDPN for the annual report or for ongoing special projects.

# Guidelines for Conducting Birth Defects Surveillance

# Chapter 11
# Data Presentation

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Copies of *Guidelines for Conducting Birth Defects Surveillance* can be viewed or downloaded from the NBDPN website at http://www.nbdpn.org/bdsurveillance.html.

Comments and suggestions on this document are welcome. Submit comments to the Surveillance Guidelines and Standards Committee via e-mail at nbdpn@cdc.gov.

You may also contact a member of the NBDPN Executive Committee by accessing http://www.nbdpn.org and then selecting Network Officers and Committees.

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∗International Committee of Medical Journal Editors. Uniform requirements for manuscripts submitted to biomedical journals. *Ann Intern Med* 1988;108:258-265.

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# Figures and Tables

# Appendices

# 11.1  Using Data for Decision-Making

*This chapter focuses on the fundamentals of data presentation for a birth defects surveillance program. A birth defects research program will have needs that go beyond what is addressed in this chapter. Readers are referred to the references and technical appendices in this chapter for additional information. The reader may also wish to refer to Chapter 8 (Statistical Methods) of* The Surveillance Guidelines *for more in-depth treatment of some of the topics touched upon in this chapter. Finally, the Members Only section of the National Birth Defects Prevention Network (NBDPN) website will be posting materials on more advanced aspects of data presentation as they become available.*

Collecting data for data's sake wastes precious resources. There is no good reason to collect data unless we intend to use them, generally to *inform* someone in a position to *do* something about the story our data tell.

Surveillance data in particular are intended for use in accomplishing the purposes and objectives of the surveillance program. In Chapter 1 of *The Surveillance Guidelines* we discussed the five major purposes of birth defects surveillance and their related objectives, as presented below.

➢ *Epidemiologic.* Epidemiologic objectives include developing timely baseline birth defects rates, monitoring trends and relationships to environmental factors, performing cluster investigations, and providing a basis for ecologic and etiologic studies

➢ *Planning and prevention.* Planning and prevention objectives include providing data for services planning, providing a basis for prevention strategies, and evaluating the efficacy of preventive services and programs.

➢ *Educational and social.* Educational and social objectives include informing the public about public health importance, informing parents about resources and care facilities, providing data for studies of economic impact, and providing data for follow-up studies of long-term effects.

➢ *Healthcare and human services.* Healthcare and human services objectives include referring children to services and resources and evaluating services utilization.

➢ *Clinical.* A clinical objective is providing the basis for clinical research.

Of course, not all surveillance programs pursue all of these purposes and objectives, but every program pursues some combination of them, and all collect data as a means to achieve them.

In order to fulfill the objectives of a birth defect surveillance program in all of these core areas, data must be collected in a complete, accurate, and timely manner. They must also be processed and interpreted in a way that ensures the availability of useful information to those with the responsibility to carry out specific activities that meet the program's objectives. Under some circumstances, this is relatively straightforward. For example, if a programmatic objective is to connect babies with specific birth defects and their families with appropriate medical and social services, then data collected on diagnosis and parent contact information immediately provide the information needed to initiate an appropriate referral. Frequently, however, there is a need to aggregate, analyze, and interpret data and subsequently present the resulting information to a variety of partners capable of taking necessary action. It is this latter more complex process that is the focus of this chapter.

## 11.1.1  The Data-to-Action Continuum

Yet data are, after all, only data. How is it that the data so carefully collected by surveillance program staff are transformed into the many different kinds of actions necessary to achieve their programmatic objectives?

There are two points to consider in answering this question. First, surveillance staff clearly cannot accomplish all of these important objectives without the help of their partners. Second, the transformation of data into action is not a discrete one-time occurrence—such as standing up with your slide presentation in front of a live audience—but rather a complex process involving extended collaboration between surveillance staff and their partners over time. To be sure, it is through presenting data in a clear manner in response to expressed interests of a particular "audience" and in support of an actionable message that this transformation begins to occur. Yet we need to bear in mind that, while the data presentation theories and skills discussed in this chapter can be mobilized in aid of this transformation, they are in fact only one aspect of the larger collaborative process that transforms data into action.

We can conceptualize this transformation as having four stages (see Figure 11.1, the Data-to-Action Matrix), with surveillance program staff and their partners closely involved in each one. Sources are abstracted to obtain *data*. Data are analyzed and interpreted to obtain *information*. Information is communicated to develop *knowledge*. And knowledge is used to inform *action.* Data presentation, then, is one of several skills that support this process, as we convey information to our program partners in order to generate the knowledge needed to embark on actions that meet our shared objectives.

## 11.1.2  Products of the Data-to-Action Transformation

Figure 11.1 suggests that each stage of the data-to-action transformation results in a distinct "product": data (Stage 1), information (Stage 2), knowledge (Stage 3), and action (Stage 4).

Let's take a moment to clarify these terms. While this chapter is entitled "Data Presentation," we are not really talking about presenting *data*, but rather about presenting the *information* generated from data in the expectation of building *knowledge* for ourselves and our partners. Although the terms 'data' and 'information' and even 'knowledge' are often used somewhat interchangeably, there are important distinctions between them.

Simply put, the purpose of data is to record "something" and the purpose of information is to build knowledge. Data (from the Latin meaning "something given") consist of raw facts or unedited stimuli. They are based on the symbolic recording of something, such as numbers, facts, and figures. Data provide a foundation for and can be developed into information, but they must be combined and integrated with other data before they become useful.

While information includes data, data do not necessarily include information. Information is data with semantic association and is the result of processing, manipulating, and organizing data in a way that adds to the knowledge of the receiver. When augmented by meaning or interpretation, data become information. It is the information developed from data that provides answers to our questions and those of our partners about birth defects, thereby increasing our knowledge.

**Figure 11.1 Data-to-Action Matrix**

| | ACTORS | NATURE OF PRODUCT | PRODUCT TYPES | PRESENTATION MODE |
|---|---|---|---|---|
| **DATA** | Data Providers | • Timely<br>• Complete<br>• Accurate | • Counts<br>• Ratios<br>• Proportions<br>• Rates | • Surveillance databases<br>• Data tables |
| **INFORMATION** | Data Interpreters | • Relevant<br>• Useful<br>• Compares like with like | • Within region<br>• Across regions<br>• Comparisons by population, time, geographic areas, benchmarks | • Histogram<br>• Frequency<br>• Bar chart<br>• Graph<br>• Spot map |
| **KNOWLEDGE** | Knowledge Makers | Generated through iterative, multi-directional communication | • Tailored presentations | • Discussion<br>• Written feedback on presentations |
| **ACTION** | Action Takers | • Appropriate<br>• Evidence-based<br>• Maximally effective and cost-effective | • Estimating frequencies<br>• Referrals to services<br>• Planning services<br>• Planning interventions<br>• Conducting research<br>• Cluster investigations | • Surveillance reports<br>• Websites<br>• Scientific publications<br>• Policy papers<br>• Guidelines<br>• Intervention protocols<br>• Risk communications<br>• Press releases, media articles and shows |

From the perspective of this chapter, our goal is to obtain data from a data provider (Stage 1–Data Provision), analyze and interpret it so that it becomes information (Stage 2–Data to Information), and then present it to and discuss it with one or more potential action takers so that it becomes knowledge that can be used to meet programmatic objectives (Stage 3–Information to Knowledge). It is then the action takers' responsibility to see that the new knowledge is used to meet the specific objectives of the program for which it has been collected and for which it is relevant (Stage 4–Knowledge to Action).

This chapter discusses each of the stages in the model in turn. We spend less time on the first and fourth stages, as both are thoroughly discussed elsewhere in *The Surveillance Guidelines*. Stage 4 (Knowledge to Action) is discussed further in Section 1.4 of *The Surveillance Guidelines* (Uses of Surveillance-based Birth Defects Data), and most of the rest of *The Surveillance Guidelines* address Stage 1 (Data Provision).

# 11.2  Stage 1 – Data Provision

| ACTORS | NATURE OF PRODUCT | PRODUCT TYPES | PRESENTATION MODE |
|---|---|---|---|
| **Data Providers:**<br><br>• **Data sources (active and passive)**<br>• **Abstractors**<br>• **Data collectors** | **Data that are:**<br><br>• **Timely**<br>• **Complete**<br>• **Accurate** | **Data Types**<br><br>• **Counts**<br>• **Ratios**<br>• **Proportions**<br>• **Rates**<br>• **Maps/GIS**<br>• **Multivariate analysis** | • **Surveillance databases**<br>• **Data tables** |
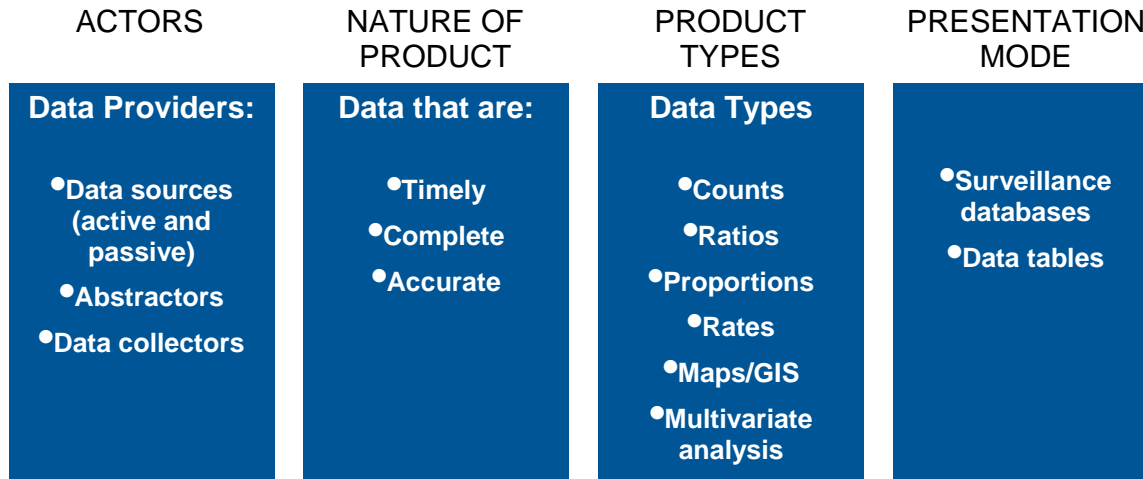
With respect to the ability of surveillance data to provide useful information, the old axiom from statistics "garbage in, garbage out" holds true. Before a surveillance staff member can begin to think about how to present data, knowing what data to present and feeling confident that the data are accurate and reliable is paramount.

In fact, the value provided by the information developed using birth defects surveillance data depends heavily on the quality of those data and the completeness and accuracy with which they are collected. The majority of the technical content of *The Surveillance Guidelines* is directed toward helping to ensure completeness and accuracy of the data that are collected and their resulting validity.

From an epidemiologic perspective, when we refer to ***data validity*** we are concerned with whether the data regarding cases in a study or surveillance program accurately reflect the numbers and characteristics of the cases that occur and that are eligible for inclusion in the data set. When we are attempting to determine or measure the occurrence of birth defects in a population, it is essential that we include all of the cases that meet the established case definition (completeness). For cross-sectional or case-control studies, while completeness is important, in the absence of including all cases validity is driven by whether the cases that are included accurately reflect all the cases that occurred in the study population (population at risk) with respect to epidemiologic variables related to characteristics of person, place, and time.

In the next section we discuss some of the analytical and interpretative issues involved in turning surveillance data into information.

## 11.3  Stage 2 – From Data to Information

| ACTORS | NATURE OF PRODUCT | PRODUCT TYPES | PRESENTATION MODE |
|---|---|---|---|
| **Data Interpreters:**<br><br>•**Data managers**<br>•**Epidemiologists**<br>•**Statisticians**<br>•**Public health staff** | **Information that is:**<br><br>•**Relevant**<br>•**Useful**<br>•**Compares like with like** | **Information Types**<br><br>•**Within region**<br>•**Across regions**<br>•**Comparisons by population, time, geographic areas, benchmarks** | •**Histogram**<br>•**Frequency**<br>•**Bar Chart**<br>•**Graph**<br>•**Spot Map**<br>•**Area Map** |

In *The Surveillance Guidelines*, we follow the Centers for Disease Control and Prevention (CDC) definition of *surveillance* as established in Chapter 1. The key themes of the CDC definition of surveillance are the integration of data collection, analysis, interpretation, dissemination, and application. It is in moving from analysis to interpretation that data are converted to information.

Some aspects of birth defects lead to potential confusion or ambiguity in reporting information about them and their distribution. In this section we discuss a number of analytical and interpretive issues that should be considered when developing and presenting birth defects data. These include:

➢ Providing contextual information (person, place, and time)

➢ Missing or unknown data

➢ Importance of comparison

➢ Approaches to measuring occurrence (prevalence versus incidence)

➢ Level of focus (which in part arises from the complex etiology and comorbid nature of many birth defects)

➢ Risk factors and the importance of timing with respect to exposures

➢ Privacy and data suppression (see also Appendix 11.1)

➢ Using Geographic Information Systems (see also Appendix 11.2)

### 11.3.1  Providing Contextual Information—Person, Place, and Time

When presenting data, it is useful to consider the key epidemiologic constructs of *person*, *place*, and *time*. What population is reflected in the data? From what location were data collected? And on what time period are the data based? One must be able to accurately and precisely answer these questions for the findings to be relevant. For example, a presentation may report very interesting results based on a sample that was collected in a very disorganized and biased manner, making it impossible to define exactly what

population is reflected in the data set. Unfortunately, these results would be of limited value because it is impossible to define to whom the findings are relevant.

Similarly, whenever variation by person, place, or time occurs, analyses should examine possible differences or trends. If a sample includes multiple ethnic groups, are there differences between these groups? Or if data were collected over a decade, were trends seen over time? A presentation should acknowledge that such trends were examined and differences reported if observed.

In the actual presentation, it is often useful to present data grouped on the basis of person, place, and time. When doing so, it is important to be mindful of widely accepted groupings inside or outside your organization. For example, *person* characteristics such as age, race, and ethnicity can be grouped based on Office of Management and Budget classifications. Audience members will be familiar with such groupings and, more importantly, they will be better able to relate the findings to their own data based upon these common groupings than they would if the presenter organized the data in some idiosyncratic manner. Similarly, *place* can be presented in a variety of ways, including aggregating based on town, county, zip code, or census tract.

Information collected over *time* can lead to more complex issues, such as the decision to report raw curves or a moving average. The complexity of time-varying data requires that one be clear on both the time period and method used in presenting such information.

## 11.3.2  Missing or Unknown Data

An aspect of data presentation often overlooked is the importance of providing information about the extent of missing/unknown data for study variables. Information that is missing or unknown can be just as important to understanding results as is the available information. This is especially true when the amount of missing information is more than minimal. Missing or unknown information can be reported in such data displays as tables, histograms, and pie charts by including a category labeled 'unknown' (e.g., maternal age $\leq$ 34, maternal age 35+, and maternal age unknown). If the way the information is being presented does not allow for a row/column/line/bar/slice to be designated as 'unknown', a footnote should be added to the data display informing the audience about the extent of the unknown data. Maps based on geocoded data, for example, could add a footnote with the "percentage of data that was not geocoded" to the geographic resolution presented.

## 11.3.3  The Importance of Comparison

Epidemiologic data tend to be numeric and presented either as counts, ratios, proportions, or rates. In addition they are usually presented as information specific to the epidemiologic parameters of person, place, and time. Information presented in this manner provides a way of making meaningful comparisons between different populations and different periods of time. Note: the points made below are of particular importance when one will be comparing data collected at different levels (local, state, regional, national) or by different programs.

Fundamental to epidemiology are the principles of comparisons:

➢ Between areas/populations
➢ Within an area/population over time

These comparisons often involve consideration of epidemiologic variables such as sex, plurality, race/ethnicity, pregnancy outcome, maternal age, etc. Comparisons are also usually of some measure of

occurrence, in the case of birth defects *prevalence* of a specific malformation or groups of malformations. Increasingly, there is interest in making comparisons between some health status indicator at the local or state level and a benchmark, such as a Healthy People 2010 objective or an agency-developed objective.

For comparisons to convey useful information, it is essential that **like be compared with like**. When comparative data are presented, the audience must know if this holds. In terms of birth defects data, there are at least four points that need to be clearly established if meaningful comparisons are to be made:

> ➤ What is being counted? Are the outcomes—case definitions—comparable? (see Chapter 3)
> ➤ How are cases ascertained? Were similar methods of case ascertainment used? (see Chapter 6)
> ➤ Are the pregnancy outcomes from which the cases were ascertained comparable?
> ➤ Are comparable measures used to summarize data?

Each of these is worth considering with respect to the information that can be provided based on surveillance data.

## What Is Being Counted?

Comparability of outcomes revolves around disease coding, classification, and the aggregation of cases. At the most general level, if we refer to "the occurrence of birth defects," we need to be clear about what is included in that term. In the past, birth defects were usually considered to be synonymous with congenital malformations and referred to diagnoses with ICD-8 and ICD-9 codes 740.0 to 759.9. Some surveillance programs, however, may follow the much more general March of Dimes definition of birth defects that includes metabolic and functional abnormalities as well. When comparing data between programs that use different definitions of the term 'birth defects', there are likely to be sufficient differences between what programs are counting as to make comparisons difficult, if not meaningless.

Even programs that use the same definition for the term 'birth defects' may vary in terms of what they include (and count) under a specific group of birth defects. One example of this relates to the reporting of studies of neural tube defects. In the past it was common to see reference to the occurrence of "central nervous system (CNS) malformations." Anencephaly and spina bifida might make up the majority of the cases, but cases of hydrocephaly and microcephaly would often be included as well. Clearly, comparing the results of a study that reported on the occurrence of all CNS malformations with one that consisted only of cases of anencephaly and spina bifida would be inappropriate.

Programs may also differ in the ways they define a specific birth defect. For example, most surveillance programs do not include preterm babies with atrial septal defects as cases. The Metropolitan Atlanta Congenital Defects Program (MACDP), for one, does not include infants of less than 36 weeks gestation at delivery among their reported cases of this defect (Correa et al., 2007). Therefore, if a program does not establish a gestational age criterion for atrial septal defect as part of the case definition, then comparison of their prevalence data with those of MACDP would be misleading.

## How Are Cases Ascertained?

The second key aspect to data comparability relates to how the surveillance program ascertains cases. For example, some have expressed concern that surveillance programs relying on the reporting of cases by hospitals (passive case ascertainment) may identify a smaller percentage of the true cases that occur than will programs that send abstractors from their staff out to hospitals to actively search records for potential cases (active case ascertainment). Such differences may be more perceived than real, depending on the individual surveillance programs involved.

Perhaps a better example of potential differences in completeness of ascertainment based on methods of case identification would be an attempt to compare data from a program that identifies cases only from vital records (birth and fetal death certificates) with data from a program that identifies cases based on medical record review. Several studies have identified serious problems with under-reporting of malformations on vital records (Watkins et al., 1996).

### Are the Pregnancy Outcomes from Which Cases Were Ascertained Comparable?

Another issue with respect to comparisons relates to the populations from which cases are identified. While some surveillance programs are able to identify prenatally diagnosed cases that result in pregnancy termination and include them in their numerator, many are not. This difference is particularly important for defects such as anencephaly and spina bifida, which are being diagnosed prenatally with increasing frequency. In one of the first studies conducted by the NBDPN, prevalence data over time were presented separately for programs that did (9 states) and did not (13 states) ascertain prenatally diagnosed and electively terminated pregnancies where a fetus with anencephaly or spina bifida was identified (Williams et al., 2002b). Figures included in this paper clearly show the potential effects of inappropriately comparing prevalence from programs that do and do not include cases from terminated pregnancies in their data.

### Are Comparable Measures Used to Summarize Data?

Once it is decided what to count and how to collect the data on what is being counted, it is important to ensure that the measures used to present the resulting information are the same. If the presenter is calculating the measures from base data, the same measure (e.g., birth prevalence expressed as cases per 10,000 live births) should be used for each of the different population groups, areas, or time periods. However, if the presenter is compiling or comparing already calculated measures, it is prudent to understand how these were calculated. For example, several surveillance programs within the NBDPN have presented birth prevalence as cases per 1,000 live births, while others have used cases per 10,000 live births. This difference should be quite evident in most instances. Less evident is the fact that surveillance programs in the NBDPN tend to use only live births in the denominator (see Chapter 8 Statistical Methods), whereas reports from other groups, such as the International Clearinghouse and EUROCAT, may include spontaneous fetal deaths and/or pregnancy terminations in the denominator. While the inclusion of these outcomes in the denominator will not have the same impact as if they are included in the numerator, it will result in slightly lower prevalence values (Sever, 2006).

When comparing groups within a population it is also good to ensure that specific birth prevalence is being calculated, i.e., that both the numerator and denominator are restricted to the same population. Occasionally, we find prevalence figures where the denominator is based on the whole population and the numerator comes from a subgroup. The above issues can be checked by carefully reviewing the Methods section of the reports from which data are being drawn.

## 11.3.4 Approaches to Measuring Occurrence—Prevalence Versus Incidence

Birth defects arise developmentally within the first few weeks after conception. As a result, many affected embryos (i.e., cases) will spontaneously abort before a woman is aware she is pregnant. Consequently, in epidemiologic terms, it is impossible for one to reliably assess the *population at risk*, as the number of pregnancies that reach the critical gestational phase where a given birth defect can arise is unknown. In addition, it is unknown how many of these affected pregnancies result in spontaneous abortions. As discussed elsewhere, it is not possible to accurately estimate the *incidence* of a birth defect—the number of new cases of a defect occurring in a population at risk during a specific time period—because one

cannot establish the number of new cases of the birth defect nor the population of conceptuses that were viable (and thus "at risk") at the relevant point of development (Mason et al., 2005). Most epidemiologists in the field suggest that data be presented and discussed in terms of prevalence, often reported as *prevalence at birth* or *birth prevalence*.

As noted, in reporting the occurrence of birth defects, prevalence estimates are often calculated so that the numerator includes cases that do not appear in the denominator. For example, while the denominator commonly consists of the number of live births, if data are available, it is generally preferable to include birth defects observed among fetal deaths and induced terminations in the numerator. The resulting prevalence is a ratio, which generally includes a multiplier—typically 10,000—so that the reported prevalence of most defects will have at least one unit to the left of the decimal point. Numerically 1.6/10,000 is the equivalent of 0.16/1,000. For further detail see Chapter 8 "Statistical Methods."

Birth prevalence provides a method of expressing the occurrence in a population in a way that supports comparisons. When the number of live births is used as the denominator, to be meaningful it should represent the same geographic and temporal "population at risk" that the birth defects cases come from. For example, in Missouri in 1989–1995 there were 193 cases of tetralogy of Fallot delivered statewide. This provides the numerator for the calculation of prevalence. The 532,592 live births delivered statewide in 1989–1995 are the denominator. The number of cases (193) divided by the number of live births (532,592) times the multiplier of 10,000 yields a prevalence of 3.62 case per 10,000 live births.

## 11.3.5 Level of Focus

Different types of birth defects can have different causes and arise through several different biologic pathways. Moreover, an individual child can have defects in multiple organ systems. This creates another fundamental issue, which can be thought of as the choice of level of focus: whether the focus is on individual (specific) birth defects or on individuals with birth defects. When the focus is on *individual birth defects*, the occurrence of specific birth defects is of interest. In contrast, when the focus is on *individuals with birth defects*, one is interested in the issue of how many people have birth defects.

How one chooses between these approaches depends on the question being asked or how the data will be used. If one is interested in identifying possible teratological effects of environmental contaminants, for example, the focus may shift from a single birth defect to the occurrence of any potentially related birth defect. This may involve examining the potential association between various chemicals and the occurrence of all types of birth defects.

Many scientists argue that analyzing all birth defects together rather than examining specific defects is of limited value. Importantly, how different types of defects can be aggregated in a biologically meaningful way is an issue of interest. Just as reports on infectious disease do not look at infectious diseases as a group, but present information on specific diseases (measles, shigella, AIDS, syphilis, toxoplasmosis, malaria, etc.), birth defects should be examined in the same way. For example, the epidemiology and causes of outcomes such as neural tube defects, gastroschisis, and Down syndrome are different so the logic of lumping them together may be questionable. Nevertheless, approaches for grouping defects in biologically and etiologically meaningful ways are being pursued.

In addition, it is important to recognize that many times a child will have more than one type of birth defect. For example, 58% of children in the Texas registry have more than one birth defect. Therefore, reporting the numbers of cases of individual types of birth defects, without informing the audience of the extent of multiple diagnoses, may unintentionally lead to an overestimation of the number of individuals in the population with birth defects. Furthermore, many audiences may be specifically interested in the number of persons with birth defects, since this information can be relevant for advocacy and health planning purposes.

## 11.3.6  Risk Factors and the Importance of Timing with Respect to Exposures

Surveillance programs often collect limited data on risk factors for birth defects, as well as on cases. However, there are important distinctions between those types of data routinely collected and those obtained as part of special studies (such as cluster investigations) or in conducting epidemiologic research. This discussion focuses on risk factor data that are often collected routinely and their presentation.

Exposures known to be risk factors for birth defects are quite limited, one of the issues that makes additional epidemiologic studies so important. Examples include *maternal metabolic imbalances* (such as diabetes) and *viral infections* (such as rubella), as well as a small number of *drugs* and *occupational/ environmental chemicals*.

Three sociodemographic variables for which data are routinely available can potentially be considered risk factors for some birth defects. These are maternal age (date of birth), maternal race and ethnicity, and maternal education. As discussed in Chapter 4 (Data Variables), the first two of these are considered as core variables, while the last is a recommended variable.

In presenting data on these variables, *maternal age* is usually grouped either into quinquennia ($\leq$19, 20–24, 25–29, 30–34 and $\geq$ 35) or into two age groups (<35 and $\geq$35). The latter grouping is used as it is particularly relevant to Down syndrome risk and prevalence. As discussed in Chapter 4, *race and ethnicity* should be presented in categories that are comparable with the federal standards in current use. If data on *maternal level of education* are collected, then they should be presented in the same categories used on the birth certificate. Following these recommendations in presenting data on the above sociodemographic variables allows information on cases to be compared with that from the certificates of live births for the at-risk population.

A final type of risk factor information to be considered here is *maternal place of residence* (address). This, too, is considered a core variable and is basic to the use of geographical information systems, a topic discussed later in this chapter. In terms of presenting data on residence, cases are usually aggregated into some geopolitical unit (such as counties) or into administrative units (such as health regions), for which information on live births is available. How these aggregated data are presented to the public or to data users other than surveillance program staff is considered below in the discussion of data suppression.

While perhaps not pertinent to the way data are presented in a general sense, the issue of maternal residence as a risk factor raises an important point about presenting information in epidemiologic studies. Particularly with the increasing utilization of GIS, the location of the mother's place of residence is sometimes used as a surrogate for exposures in studies of risk factors associated with the ambient environment (Sever, 1997). In considering residence as a surrogate for exposure in studying birth defect risk factors, it is important to know the location of the mother's residence at the time in gestation when relevant developmental events are occurring. Periods of sensitivity are well known for many organs and structures and, for the most part, these are during the embryonic period, early in pregnancy (Mortensen et al., 1991).

Unfortunately, most surveillance programs collect only information on the mother's address at the time of delivery, when it is residence during embryogenesis that is biologically relevant. This is important in assessing possible risks related to the ambient environment because several studies have shown that a

large percentage of women move between conception and delivery (Canfield et al., 2006). Residence at delivery, therefore, is not only limited in its usefulness as a surrogate for exposure, but in many cases it does not reflect biologically relevant exposure, since it does not represent where the woman lived when crucial events in embryogenesis were taking place. This limitation should be noted when data on maternal residence are presented as part of epidemiologic studies of environmental reproductive hazards.

## 11.3.7  Privacy and Data Suppression

Specific birth defects are often rare events (sometimes extremely rare) leading to yet another set of issues that must be considered when presenting birth defects data. The public health professional must balance the potentially conflicting goals of information dissemination with protection of the privacy of persons in the community. When the number of cases in a diagnostic category within a group or stratum (such as race or sex) is small or the population from which the cases are determined is small, the risk of allowing a specific individual to be identified may be deemed too large to be acceptable. In such cases, steps must be taken to protect an individual's privacy. In addition to protecting privacy, prevalence information is often suppressed when concerns exist regarding possible statistical unreliability of estimates that are based on small numbers.

The most common method of preventing the identification of specific individuals in tabular data is through *cell suppression*. This means not providing counts in individual cells where doing so would potentially allow identification of a specific person. Cell suppression can also be done by combining cells from different small groups to create larger groupings that reduce the risk of identifying individuals. While there are also more sophisticated data perturbation methods that use statistical noise to mask sensitive information, these are generally more suitable for use with economic or financial data than with public health data.

In general, the more restrictive a suppression rule, the less information a given table or report will provide. The weaker a suppression rule, the greater the potential threat of revealing confidential health information. It is a question of balancing the threat to individual privacy with the public health value of presenting the data.

Overall, deciding when and how to suppress birth defects information is more a social, political, and legal issue than a technical one. The technical aspects are quite straightforward, but the contextual and procedural/policy issues are likely not to be. These all need to be considered and balanced in the local context before informed decisions can be made to suppress or not to suppress data in program reports or other documents.

Surveillance program administrators and technical staff should be aware that standards used to suppress data may already be set in state laws or in departmental or institutional rules and regulations. It is the responsibility of surveillance staff and administrators to be aware of these standards and practice within their limits. If standards are not established, it behooves a surveillance program to establish rules that will be followed consistently. This is best accomplished with the assistance of an advisory committee, an institutional review or privacy board, or a similar body.

Appendix 11.1 reviews the basic methods, issues, strengths, and vulnerabilities of cell suppression.

# 11.3.8  Geographic Information Systems (GIS)

The application of Geographic Information Systems (GIS) methods has become an integral component of aggregating, analyzing, evaluating, and displaying health data. The current practical applications of GIS in epidemiologic studies range from descriptive statistics (i.e., plotting data on a map) to evaluation of spatial relations between environmental exposures and health outcomes.

Several definitions exist for geographic information systems. One of the most recent, as found in *Healthy People 2010,* defines GIS as "powerful tools combining geography, data and computer mapping" (U.S. Department of Health and Human Services, 2000). Software packages available today, such as ArcMap and MapInfo, integrate many GIS functions. These include (1) database management, (2) data manipulation and analysis, and (3) data presentation (i.e., displaying data on a map). To be included in GIS, the data should have some kind of geographical or spatial component that can be translated into digit maps. Appendix 11.2 contains a brief introduction to GIS mapping along with a list of suggested references.

# 11.4  Stage 3 – From Information to Knowledge

| ACTORS | NATURE OF PRODUCT | PRODUCT TYPES | PRESENTATION MODE |
|---|---|---|---|
| **Knowledge Makers:**<br><br>•**Data providers**<br>•**Data interpreters**<br>•**Action takers** | **Knowledge that is:**<br><br>**Generated through iterative, multi-directional communication between collectors, interpreters, and users of data** | **Knowledge Types**<br><br>•**Presentations tailored to information needs and backgrounds of various audience types** | •**Discussion**<br>•**Written feedback on presentations** |

As mentioned previously, the key themes of the CDC definition of *surveillance* are the integration of data collection, analysis, interpretation, dissemination, and application. In the previous section we spoke of moving from analysis to interpretation, whereby data are converted to information. In this section we are more concerned with dissemination of information with an eye toward application, whereby information is converted to knowledge capable of informing action. We now turn to some of the more technical, as opposed to the more philosophical or theoretical aspects of data presentation. In the broadest sense, we are here concerned with the clarity of the information presented and a lack of ambiguity in the message to be communicated.

We can conceive of the process of communication as having five major components—the sender (presenter), the medium, the message, the objective(s), and the receiver (audience). It is important in the development of a data presentation to keep all of these components in mind. It is also important to realize that communication is not simply a linear process of conveying the message from the sender to the receiver, but rather often involves a loop from the sender to the receiver, back to the sender, and back to the receiver.

In general, we suggest working backward through the communication sequence when designing your presentation. That is, instead of beginning with yourself (the sender) and what you want to tell the audience, begin by thinking about the audience (the receiver) and its information needs. Beginning with the audience will help you determine the objectives of the presentation, formulate the message, and select the best medium to use in conveying that message. Below we walk you through the process of developing a data presentation by (a) accurately characterizing the audience and understanding its needs, (b) establishing the purpose or objectives for a given presentation, (c) developing the content of and ensuring the clarity of the message, (d) selecting the most appropriate medium for the message, and (e) being aware of biases you as the presenter may have. We do not mean to suggest that consideration of elements a-e must be undertaken sequentially. However, all need to be considered carefully in the context of the presentation as a whole, even if some are apparent "givens". For example, if you are told you must prepare a report for the Governor on x topic, then you know the audience and the medium as well as the overall objective of the report, namely "to provide information on x." Still you would do well to learn

more about why the Governor is interested in x, what specific information is being sought, and how the information will be used before developing the report.

## 11.4.1  The Receiver—Understanding the Audience and Its Information Needs

Know your audience! One of the central tenets of any presentation is identifying the audience being addressed and recognizing the information needs of its members. This includes taking into consideration the audience members' backgrounds, interests, and bases of knowledge. For example, a presentation to epidemiologists may include detailed information on complex analyses, yet these should be presented only as a summary to an audience of policy makers. The former may expect—and insist on—a presentation including numerical estimates of standard errors, confidence intervals, etc., while the latter will respond better to straightforward graphical displays that illustrate the key points. Even in an apparently homogeneous audience there can be significant heterogeneity. For example, a presentation to a parent group may include both highly informed individuals who have extensively researched a particular birth defect, as well as new parents who may be wholly unfamiliar with the field.

If the nature and level of expertise of your audience is not clear to you, do not hesitate to talk to someone in a position to know more about the audience and why the presentation has been requested or arranged.

## 11.4.2  The Objective(s)—Determining the Purpose of the Presentation

The type of information an audience is interested in and the questions posed can vary considerably, which in turn will influence your objectives in developing the presentation. An audience consisting of policy makers may be hoping to learn about population trends and attributable risk. Researchers may be interested in the prevalence of cases based on various demographic variables, while service providers may be most interested in the geographic distribution of cases and services. These differences lead to different types of questions that will require different analytic approaches and may lend themselves to different formats of data presentation. In Appendix 11.3 (the Data Users Matrix) we characterize a number of possible audiences for a birth defects surveillance presentation in terms of their likely information needs and presentation approaches that might meet those needs.

In sum, one must be prepared to use different approaches to audiences that differ in current levels of knowledge regarding the topic, as well as in having different interests, objectives, and information needs. The questions of interest to a particular audience will drive both the analytic approaches and the medium or format selected for presentation.

## 11.4.3  The Message—Developing Content and Ensuring Clarity

Having meticulously collected, cleaned, and analyzed a surveillance program's birth defects data, the proud owner of neatly tabulated findings may well wonder, why it is necessary to also express these findings in graph or chart form. Shouldn't the numbers speak for themselves?

The answer is yes, of course, the researcher should be able to verbally convey the most important results and to summarize succinctly characteristics of the data. In addition, it is certainly helpful to make complete tabular data available to the consumer of epidemiologic results (i.e., the audience). However, while individual learning styles differ, most people are primarily oriented to interpreting visual information as opposed to tabular data (Spence, 1990) and can more easily make judgments about that information based on a limited number of simple cues: smaller/larger, brighter/darker, increasing/diminishing. Therefore, a graphical display increases the efficiency with which your audience processes your information (Legge et al., 1989). Remember, too, that data presentation is aimed at

meeting a specific purpose; whether stated or not, you have an objective and a message to convey, and your audience needs to understand it.

In this section we discuss graphical representations (such as graphs and charts), concluding with tips for you to keep in mind as you develop a data presentation. We then offer guidance on how to choose the appropriate format for displaying a given type of data, with further detail provided in Appendix 11.4. We conclude this section with a discussion of the characteristics of a clear, informative table.

## Graphs and Charts

The discussion below will enable you to create graphical representations of your data that meet the following requirements:

➢ Convey results accurately
➢ Allow for efficient interpretation
➢ Engage the interest of the audience

**Conveying results accurately.** Essentially, all of the information conveyed through graphs and charts allows for comparison and answers a single question: which is larger? This is a question of proportionality. Therefore, it is important that visual elements reflect the same proportions as the data they represent. For example, Sample Figures 1A and 1B demonstrate cases per 10,000 live births for a specific birth defect, but the figures use a different range of values on the y-axis. This practice distorts the actual differences in proportion making it appear as if the rates of these two defects are quite similar, when in fact dislocation of the hip is about twice as common in this population (Muscatello et al., 2006).
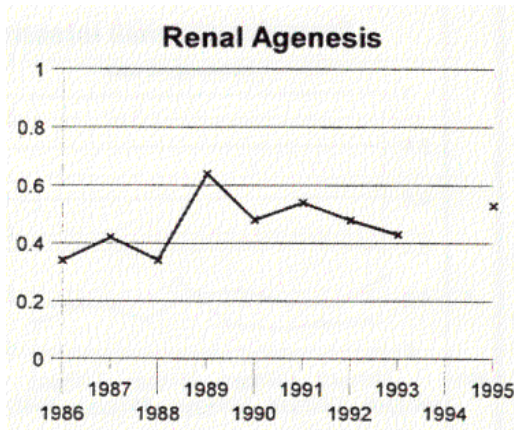


**Sample Figure 1A. Cases per 10,000 live births 1986–1995**
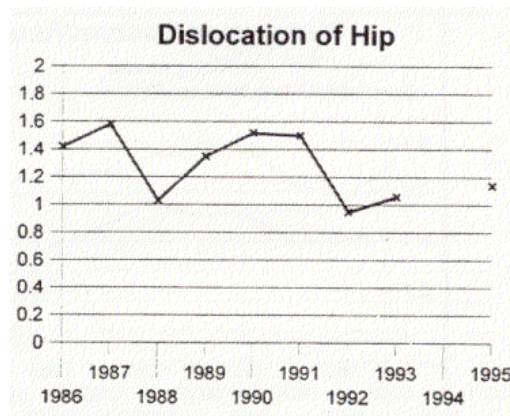


**Sample Figure 1B. Cases per 10,000 live births 1986–1995**

However, it is not always desirable to use the same scale for all charts. Sample Figure 1C demonstrates cases per 10,000 live births using the same scale as Sample Figure 1A, but since absence of limbs is so much rarer than renal agenesis, it is difficult to detect any difference among years for Limb Absence. Therefore, it is important to weigh the essential information you want to convey before deciding on scale (as well as other features); in this case, which is of primary concern: between-defect comparisons or illustrating a trend for one particular defect?
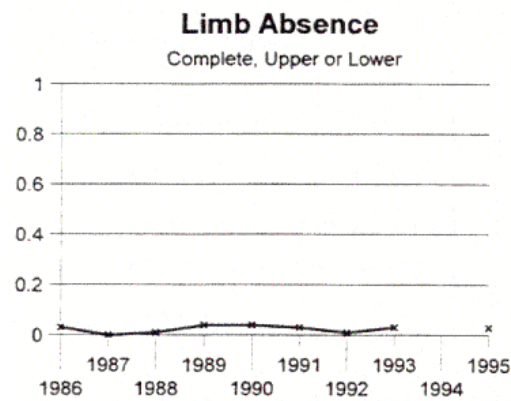
**Sample Figure 1C. Cases per 10,000 live births 1986–1995**

Chart design characteristics that can distort proportions when changing scales across multiple graphs include:

➢ Two different graphs examining the same outcome, but based on different time periods or different lengths of time.

➢ A bar graph of several time-based groups, where the groups correspond to different lengths of time.

➢ Graphs of statistical functions, such as regression lines, that extend beyond the range of values observed in the data.

➢ Use of three-dimensional graphical elements.

**Allowing for efficient interpretation.** To support efficient interpretation of data an important principle to follow is the *ink-to-data ratio*. Simply put, try to minimize the proportion of "ink" (or what would be ink on a printed page) that is employed in actually representing data. This means eliminating extraneous graphical elements that do not convey additional meaning, such as slide backgrounds, clip art, animations, and other elements of what is often referred to as "chart junk."

Chart junk can appear in two varieties. The first is extraneous material unrelated to the actual data. This type of junk is relatively easy to eliminate as it tends to be under the control of the person using the graphing software. So resist the temptation! In cases where the junk is generated by the graphing software, do not hesitate to edit it out wherever possible.

The second form of chart junk involves certain graphic styles that require a large amount of space to convey a small amount of data. In this regard, the key is to focus on the data themselves, rather than the data "containers." Data containers are shapes used to reflect data, such as bars and line markers, and minimizing their size can be particularly helpful if one is presenting a large volume of data.

For example, consider which of the figures below is easier to understand, Sample Figure 2A or 2B. *Hint:* See how many instances of chart junk you can identify in Sample Figure 2A.[1]
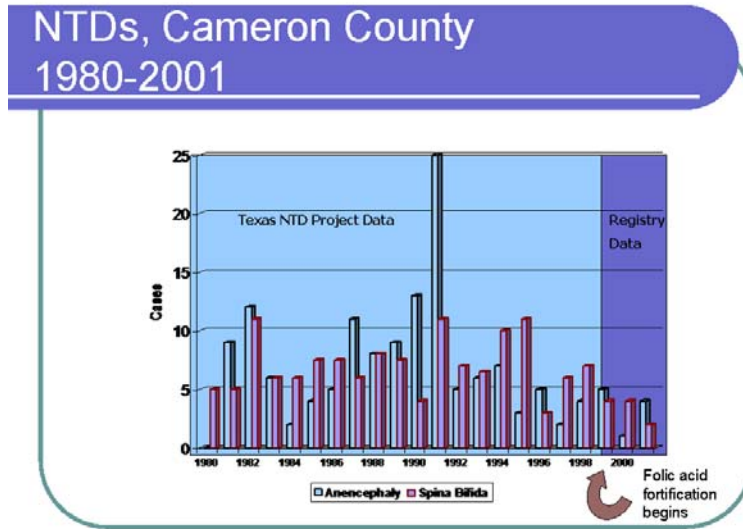


**Sample Figure 2A. Example of excessive "ink:data" ratio**
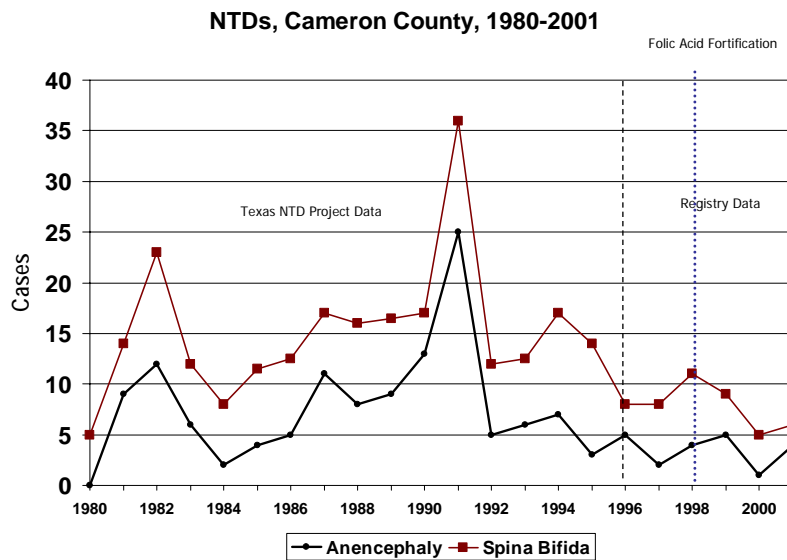


**Sample Figure 2B. All "ink" conveys essential information**

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[1]  In Sample Figure 2A, the differing color backgrounds, the slide design elements, and the stylized arrow, combined with 3D bars, employ a great deal of "ink" to convey the same information as conveyed in Sample Figure 2B.

Sample Figures 2C through 2F present further techniques to reduce the amount of "ink" in a chart or graph. In 2C, adding data labels to the bars allows you to eliminate additional "ink" in the form of gridlines, while allowing the viewer to accurately assess the value of each bar. Horizontal orientation allows category labels to be spelled out rather than abbreviated. Sample Figure 2D contains no legend; rather each data series is labeled directly, with color coding used to ensure correct pairing of label with series. Sample Figure 2F (versus 2E) also uses direct labeling instead of a legend, and changes X axis scaling to every other year, which is sufficient for these data.
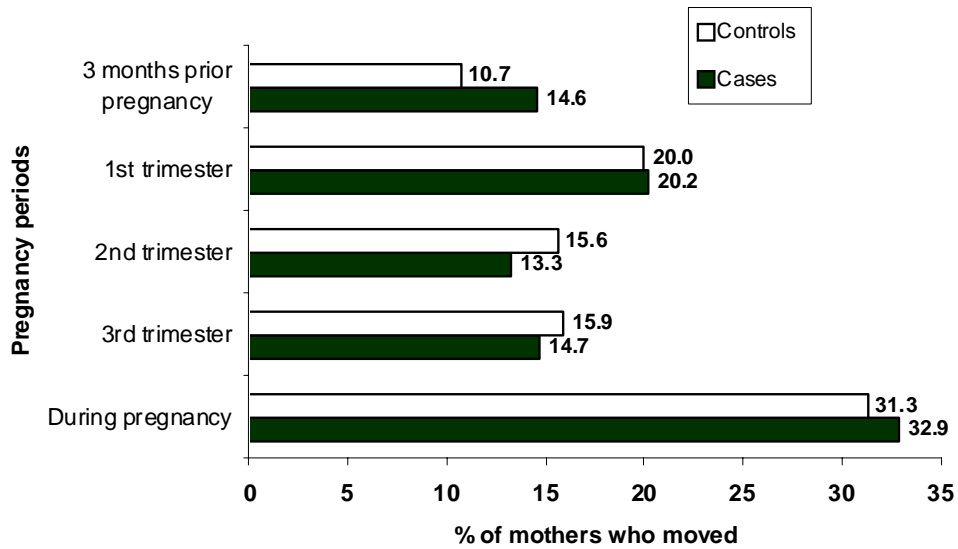


**Sample Figure 2C. Use of data labels to eliminate additional "ink" in the form of gridlines. Horizontal orientation allows category labels to be spelled out.**

### NTDs by County, 1998-2001
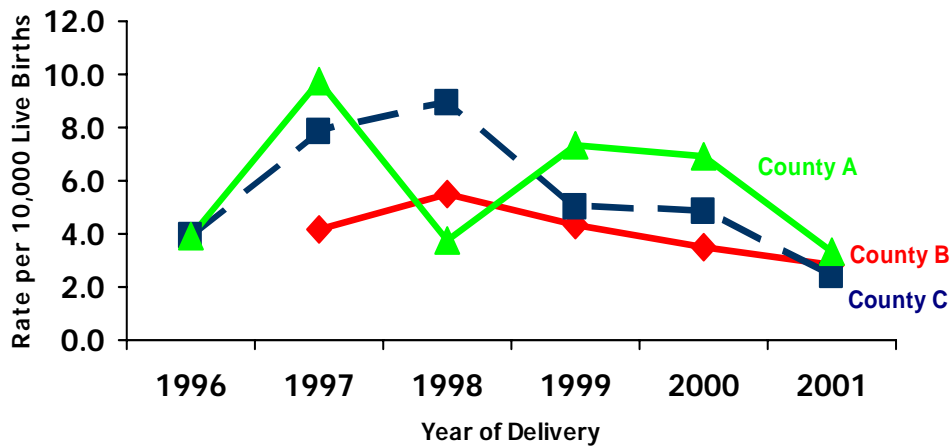


**Sample Figure 2D. Direct labeling of data series rather than legend. Use of color coding to ensure correct pairing of label with series.**

**Sample Figures 2E and 2F. Note that Figure 2F uses direct labeling rather than a legend, and changes X-axis scaling to every other year.**

As a general rule, an intelligent reader/observer should be able to clearly interpret a chart or graph without referring to supplemental text or materials. If a figure will be used in a live presentation, the information presented visually can be minimized to the extent that it will be supplemented orally. However, copies of an oral presentation or figures used in formats without benefit of augmentation by a presenter should contain sufficient information to stand alone yet still be understood.

**Engaging audience interest.** While tabular data lend themselves to accurate interpretation, especially by those accustomed to working with numbers, they nevertheless require more time to process (Spence, 1990), are tedious to follow in a slide presentation, and are less accessible to non-technical audiences. A compromise suggested by Tufte (2003) is to use handouts, including the actual data tables, in lieu of the standard 2x3 printed version of slides.

Cautions about chart-junk notwithstanding, certain visual elements can improve audience engagement. For example, color can be an effective means of increasing visual interest and adding clarity to a figure (compare the differing impact of Sample Figures 3A and 3B). Color can also be used to portray increasing data density (the amount of information conveyed relative to the size of a figure) or to add an additional level of information to a figure. For example, the size of a dot may indicate the number of babies born at a hospital, while the color of the dot indicates the percentage of births who spend more than 24 hours in a neonatal intensive care unit. However, avoid too much color, as well as combinations of colors that may distract, confuse, or mislead readers.
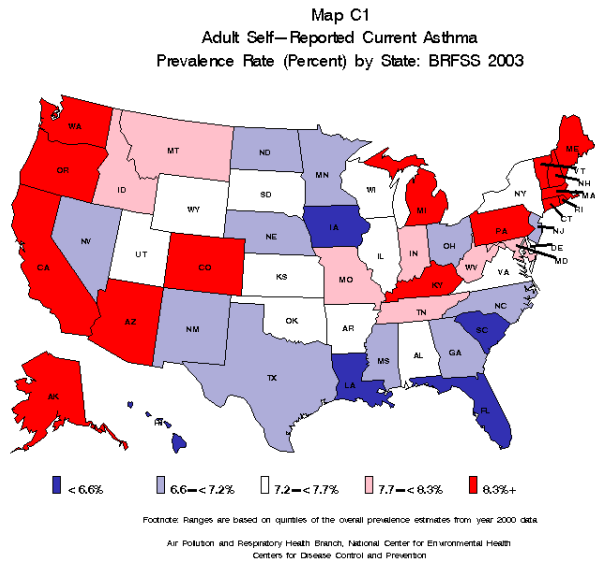


**Sample Figure 3A. Example of a map using color codes**
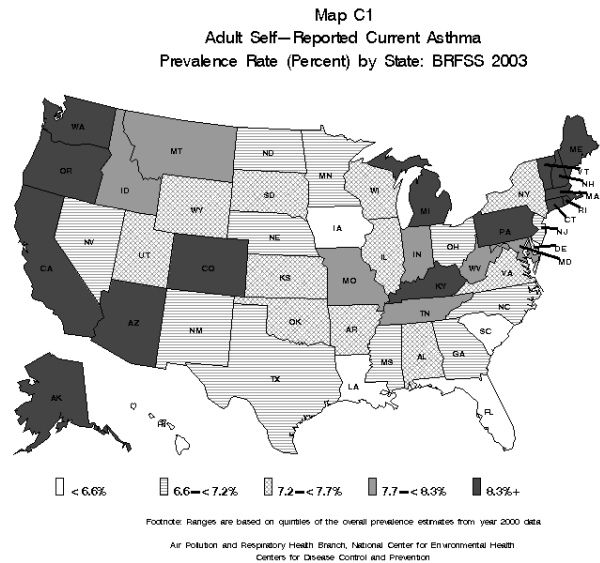
**Sample Figure 3B. Example of same map in gray scale**

Understated, subtle backgrounds, textures, and other graphical elements can be eye-catching but can also easily be over-used. Furthermore, no amount of visually stimulating material on a chart can take the place of a presenter whose tone of voice, bearing, and engagement with the audience bespeak a clear understanding of and excitement about the information being presented. Table 11.1 below contains some summary tips for graphical data presentation.

**Table 11.1 Summary Tips for Graphical Data Presentation**

| General Tips |
| --- |

- Remember that the default graphing settings on your software package (e.g., PowerPoint) are rarely the best for creating an effective graph. If you do not have the time or interest to customize your own slides, consult an expert in your organization.
- Use a clear and simple font (e.g., a sans serif font such as Arial).
- Use footnotes to explain acronyms and methods (Muscatello et al., 2006).
- Restrict the use of abbreviations to those that will be known to everyone in a potential audience or readership, or provide a list of the less well-known abbreviations used, keeping them few in number and usage.
- Indicate the units that are being used (e.g., age in days, weight in grams).

| Analytical Tips |
| --- |

- Emphasize differences between groups—identical patterns across groups can be stated and/or expressed in a bullet point and do not need to be portrayed in a figure.
- Avoid comparisons across multiple figures.

| Visual Tips |
| --- |

- Avoid the use of background pictures, or additional pictures, lines, or shapes that are added solely to "beautify" a figure.
- Avoid the use of unnecessary or heavy gridlines. Use white spaces with a bar instead of a grid line.
- Eliminate 3-D bar graphs, which add lines and shading while providing no additional information. Furthermore, two-dimensional charts are generally interpreted more quickly and accurately than those in 3-D (Hughes 2001).
- Eliminate unnecessary legends. Legends—if absolutely needed—can be placed inside the plot area for a graph. This increases the maximum size of the graph. Rather than a legend, use direct labeling if possible.
- Simplify labeling (Muscatello et al., 2006). For example, a time series on the X axis need not always have every year listed—it is implied that 1995 is the point between 1994 and 1996.

| Staying on Message |
| --- |

- Remember your core message and do not present irrelevant data (e.g., detailed methodological information if not a methodological study).
- For certain audiences (e.g., lay persons or policy makers), consider wording the title as a plainly stated question that guides interpretation of the graph (Muscatello et al., 2006). For example, "Is gastroschisis more common among babies born to younger mothers?" rather than "Patterns of prevalence of gastroschisis by age of mother"
- Show your charts and tables to someone unfamiliar with the data and ask them how they interpret the "bottom line" message from each. Revise to improve clarity.

## What Type of Graph or Chart Should I Use?

Appendix 11.4 contains information on some of the more common types of graphs and charts along with suggestions on how to choose a type appropriate to the data you are planning to display.

Before making your final decision, however, you should also ask yourself two questions that relate less to the nature of your data and more to your own personal preferences and the needs/interests of your audience:

➢ *Am I comfortable explaining this graph or chart?* If the answer is no, find an alternative format with which you are more comfortable.

➢ *Given my audience, should I sacrifice detail for clarity, or clarity for detail?* For example, an audience of foster parents would probably benefit from clarity with less detail, whereas an audience of epidemiologists will readily comprehend your meaning and will rather be looking for additional detail about methods or sample characteristics.

## Tables

Despite the usefulness of graphical data presentation formats such as those just described, there will be times when a table is still the ideal choice. Tables display data in a systematic way and help readers locate specific information readily. Simple tables can stand alone in a slide presentation or be used as a supplemental handout when presenting summary data in graphical format.

Good tables have (see Sample Table 1):

➢ A table number

➢ A table title that clearly identifies the data displayed

➢ Column and row headings

➢ At least 3 horizontal lines (below the title, column headings, and data fields)

➢ Decimal alignment

➢ Expanded forms of abbreviations used in the tables, generally as footnotes

➢ Additional explanatory footnotes as needed

**Sample Table 1. Counts of selected birth defects cases and maternal country of birth, 2004**

|  | Maternal country of birth | | | | | | | |
|---|---|---|---|---|---|---|---|---|
|  | U.S.-born* | | Mexico/CA** | | Others*** | | Missing | |
|  | count | % | count | % | count | % | count | % |
| Controls | 539 | 48.4 | 498 | 44.7 | 68 | 6.2 | 8 | 0.7 |
| Heterotaxia | 63 | 36.6 | 97 | 56.4 | 11 | 6.4 | 1 | 0.6 |
| Omphalocele | 42 | 48.3 | 44 | 50.6 | 0 | 0.0 | 1 | 1.1 |
| Gastroschisis | 58 | 43.3 | 63 | 47.0 | 12 | 9.0 | 1 | 0.7 |
| Oral clefts | 49 | 52.1 | 38 | 40.5 | 7 | 7.4 | 0 | 0 |

CA=Central America
\*    50 U.S. States, Puerto Rico, Virgin Islands (U.S.)
\*\*   Mexico, Costa Rica, El Salvador, Guatemala, Honduras, Nicaragua, Panama
\*\*\* Algeria, Argentina, Bolivia, Brazil, Others

# 11.4.4  The Medium—Ensuring Its Appropriateness

Now that you have considered your audience and its needs, established the purpose for communicating your data, and developed the content and clarity of the message, it is necessary to select the most appropriate medium for the message so that it reaches the right people in a way that will help them to understand, interpret, and use the information. The selection of an appropriate medium, or communication channel, varies depending on the format of your message and the audience's access to the medium.

Communication channels can be active or passive. *Active channels* require the audience to engage with the information; *passive channels* require less interaction. Interpersonal communication, print readership, and Internet communication are examples of active channels, while passive channels include television and radio. A study comparing media type and source of information with the personal context of health-oriented attitudes and behaviors (Dutta, 2007) has demonstrated that health-oriented individuals sought active channels as primary sources of information. Non health-oriented individuals were more likely to obtain information, such as prevention messages, through passive entertainment-education channels.

Information might reach your intended audience directly, via publications, or more indirectly, such as through interpersonal communication by a social service professional relaying information to a family affected by birth defects. As you communicate information through one channel, consider how the data will be interpreted as they flow through other channels (Valente et al., 1996). Below we briefly discuss some of the more common communication channels used for the presentation of birth defects data.

- ➢ Reports and publications
- ➢ Professional presentations
- ➢ Mass media
- ➢ Websites
- ➢ Community outreach

## Reports and Publications

Birth defects data are commonly presented in reports, including internal documents, working papers, and scientific publications. Use guidelines from journals for content and format. The level of detail should be based on the audience and its needs. Follow the principle of *tell 'em:* "Tell 'em what you're going to tell 'em, tell 'em, and tell 'em what you told 'em" (Collins, 2004a). Summarize the key points of the report in an abstract or executive summary, highlight your message clearly, and conclude with a summary. A well-written abstract should be able to stand alone without reference to the article or report being summarized and should concisely outline all relevant topics while excluding unnecessary detail, generally in 200 words or less.

Within the report, pay careful attention to describing explanatory table headings and figure legends. A review of graphical presentations published in *Journal of American Medicine* and *Annals of Emergency Medicine* (Cooper et al., 2002) identified few indicators of poor quality graphs: lack of definition of symbols, internal errors, contradictions with the text, numeric distortion, lack of visual clarity, nonstandard graphic conventions, or extraneous decoration. However, 31% of graphs were not self-explanatory, meaning the reviewers could not unambiguously interpret the graph despite reading the study design and legend of the graph. Additionally, 48% of graphs did not illustrate the underlying distribution and 48% did not depict important covariates.

**Professional Presentations**

At professional meetings, data are generally presented as poster presentations or platform presentations.

**Poster presentations.** A poster presentation is a visual display that summarizes your research or programmatic project. The display is mounted on a poster board provided at the meeting. The display includes visual aids such as data tables, charts and photos, along with a limited amount of text presenting the highlights of your topic. Conference participants should be able to quickly understand the work you are presenting including, as appropriate, your central research question or hypothesis, your research approach, and your results. After reviewing your poster, many participants will ask you questions and share their observations. Poster presentations can be an ideal way to:

➢ Provide a limited amount of information to a diverse audience
➢ Start productive conversations with new colleagues
➢ Summarize work you have recently completed
➢ Obtain useful feedback in developing the study further or in developing a manuscript
➢ Advertise your work to colleagues or potential employers

Poster presentations provide key opportunities for scientists to network and discuss shared interests with colleagues.

Successful posters tell an interesting story and are visually appealing, logically organized, and easy to read. Visually appealing posters are simple, uncluttered displays that use a variety of tools to convey information (e.g., data tables, figures, photographs). Color adds interest, but be conservative about the number of colors you use. Bright colors can be disconcerting. Judicious use of underlines, boldface type, and bullets can succinctly highlight important information. "White space" is critical to creating an uncluttered look. A poster printed on a single large (8' x 4' or 4' x 4') sheet of paper is the easiest to view and mount on the poster board.

When constructing figures, charts, or tables, focus the viewer's attention on the data by reducing or eliminating chart "junk" such as non-essential lines or redundant percent symbols (%). Limit the number of decimal points presented. When you can, label data directly rather than referring the reader to a legend. If possible, convert tabular material to figures that are easy to understand. (See Section 11.4.3 for further discussion of charts and graphs.)

Logically organized posters start with a banner title across the top with the authors listed below, followed by their institutional affiliations. Poster content—text and visuals—should be organized so that they begin in the upper left corner of the poster and end in the bottom right corner. Readers will look at the poster from the top down and from left to right. The layout should follow the format of your conference abstract: generally covering the topics introduction or background, methods, findings, and conclusions. Many posters include the abstract as the initial block of text. Each section should have a brief heading, and sections should be separated by a little "white space." The text should be condensed to key points and grouped into blocks of no more than 50–75 words. Avoid abbreviations or acronyms that may be unfamiliar to your viewers.

Posters that are easy to read use fonts that are legible from a distance of 3–5 feet. For the poster title, use a very large font (84 point or larger). Author name and affiliation information can be displayed in 72 point.

For other elements of the poster, consider these guidelines:

➢ Headings and subheadings – at least 32 point
➢ Text, figure legends, and tables – at least 18 point

Keep the font style for similar content consistent throughout. Be sure that format headings and text of the same level of importance use the same font size. Avoid upper-case or "ALL CAP" fonts. Dark letters on a light background are easiest to read.

Some people may ask that you "walk" them through your poster. Avoid reading it! Instead, summarize the big picture of what you did and why. Use the poster's graphics to illustrate your major findings and support your conclusions. Presenters often provide a condensed version of their poster for interested viewers (e.g., a PowerPoint handout). You might also consider handing out additional information, such as supplemental data tables. *Always include your contact information.*

**Platform presentations.** Platform presentations are delivered through a structured talk or lecture, commonly using presentation visual aids, such as MS PowerPoint. Effective PowerPoint presentations *support,* rather than replace, the delivery of your presentation. Do not be tempted to read directly from your slides. The quality of the presentation depends on the quality of the presenter's communication of the information and not entirely on the quality of the visual aids (Collins, 2004a).

As with any public speaking activity, speaking softly, unclearly, or in a monotone voice; using excessive hand gestures; and speeding through slides without giving the audience a chance to digest the information will not communicate your message well. Pay attention to the pace and timing of your talk, allowing pauses but also following time limits. Prepare your presentation for compatibility with any computer, bring back-up copies of your presentation and, most importantly, rehearse. Rehearsing, especially in front of a representative audience, will help you become comfortable with your presentation, provide an opportunity to clarify any points that are potentially confusing, and enable you to assess the presentation's natural and logical flow (Collins, 2004b). It will also give you another chance to proofread for potentially embarrassing errors.

When preparing your visual aids, follow principles of clarity, readability, and simplicity. For clarity, design your slides with only a few key points per slide. A standard recommendation is the "rule of six": 6 lines per slide and 6 words per line (Collins, 2004b). Use contrasting background and text colors so your words are readable, but avoid hard-to-read color combinations such as red/green, brown/green, blue/black. Font sizes should be at least 24 pt for text and 36–40 pt for titles, but also consider the size of the room you are presenting in to ensure the people furthest from the screen can read the slide. Setting the entire text in bold can also increase readability.

In terms of simplicity, emphasize the most critical point on each slide. Include pictures and graphs for visual interest when they are relevant, but choose them wisely to minimize distraction from the main point. Tables can be difficult for audiences to read and interpret; look for other ways such as graphs or text to communicate the same information more clearly. If you do choose to use a table, be sure to make use of white space so that the audience can easily see the most salient points without sifting through clutter (Ryder, 1995).

Finally, remember that it is not the topic or data alone that creates a meaningful presentation. Strategic communication of understandable information is the key to successful delivery of data through the professional presentation medium (Thompson et al., 1987).

**Mass Media**

Dissemination of birth defects data to the general public occurs through many channels: printed news material, television, radio, and websites are just a few examples. Since these media have a broader reach than presentations at professional conferences, the audiences will be more heterogeneous. It is important to integrate the target audience's cultural values into the strategy when selecting the appropriate communication channel, but the ethical challenges of communicating information accurately through mass media are difficult to avoid (Guttman, 1996). A review of 10 years of health content in the media concludes that "popular media is not likely to facilitate understandings helpful to individuals coping with health challenges" (Kline, 2006). The topic of birth defects tends to be misrepresented in the media, generating unnecessary public anxiety (Marks, 1993). If mass media is chosen as a communication medium, think about how the public understands and interprets risk, so that it is not interpreted inaccurately (McComas, 2006).

While there is no method that will match all needs for knowledge, understanding the needs of potential users will help determine if mass media channels are appropriate as well as the best way to tailor the message through the medium (Williamson, 2005). Communication strategies should consider the audience's access to information channels, motivation for information, literacy and numeracy, likelihood of interpreting complex data, and cultural context.

**Websites**

Using websites to convey information about birth defects to the public is becoming increasingly common as health-oriented individuals actively seek knowledge, but these individuals' trust in the information source is paramount. Analysis of data from the Health Information National Trends Survey (Rains, 2007) shows that "trust in information-oriented media, entertainment-oriented media, and one's health care provider all predicted Web behavior and perceptions." Users of the Internet as a source of information are most likely to be women who have high knowledge about resources, regardless of format, and are likely to discuss the information they find with health care providers (Warner and Procaccino, 2007). These women typically have a higher level of education and socioeconomic status (Pandey et al., 2003).

Websites are also useful for disseminating data to research, surveillance, program, and policy users. For all audiences, the website should be clearly laid out, interactive, tailored to the audience, and regularly maintained and updated for current information.

**Community Outreach**

Another way to communicate birth defects data to the public is through community outreach. Think about creative ways of disseminating information in addition to more traditional routes; look beyond PowerPoint, posters, and reports. Your audience could be someone affected by a birth defect who may or may not attend conferences, read journals, or look at websites. As mentioned earlier, non-health oriented individuals may not actively seek information, especially if they have low literacy or numeracy skills, and consequently low health literacy skills. The attributes of health literacy are "reading and numeracy skills, comprehension, the capacity to use information in health care decision-making, and successful functioning as a healthcare consumer" (Speros, 2005). Over 50% of Americans have limited literacy and numeracy abilities according to a 1992 National Literacy Survey so health materials should be written in simple terms to increase understandability. The health literacy approach is not "dumbing down" data, but simplifying it into reader-friendly plain language so the message is communicated clearly (Stableford and Mettger, 2007).

Some examples of community outreach strategies include:

> ➢ Strategically disseminating materials (brochures, posters and pamphlets) in public locations,
> ➢ Delivering information at community events or health fairs,
> ➢ Connecting with key community gatekeepers such as health promoters who share birth defects information through interpersonal communication.

Understanding the local context is imperative for developing appropriate communication strategies for community outreach.

Remember: "A word of caution that can't be repeated often enough: The medium does NOT replace the message, be it Morse code or interactive video-on-demand. The principal objective remains to choose the right message, for the right people, at the right time and to ensure that it gets through in the most efficient and effective manner" (Chamberlain, 1996).

## 11.4.5  The Sender—Being Aware of Biases

Finally, as a presenter, one rarely faces an audience without having one's own personal interests and objectives. These may range from seeking funding to promoting a particular theory or model and may or may not align with the objectives and interests of the audience. We should nevertheless strive to present information in as impartial and balanced a manner as possible. This includes not omitting or minimizing contrary information, or choosing or manipulating figures or statistics in order to support a given objective.

## 11.4.6  Pulling It All Together

What are the factors that drive data presentation at the stage when you are transforming information into knowledge? As stated previously, when planning a data presentation, it is important that you as presenter, and catalyst in the transformation, pay attention to all the other elements of the communication process. That is, that you (a) understand the audience and its needs, (b) establish the objective(s) for the presentation, (c) determine—based on earlier analysis and interpretation—what the message is and how most clearly to present it, and (d) decide upon the communication medium. That is, the elements listed below must all be suitably "matched" in a data presentation:

> ➢ Audience and its needs
> ➢ Objective(s)
> ➢ Message (information being shared)
> ➢ Communication medium

The three case studies presented below demonstrate how these elements of a presentation must be coordinated and addressed.

| **Informing the Public about Birth Defect Prevalence** | |
|---|---|
| **Audience** | The public |
| **Objective** | To inform the public about the frequency of a birth defect in an area, e.g., a state or public health region |
| **Message** | The observed prevalence of birth defects during a specified time or trends over time |
| **Communication Medium** | Tables or graphs that are clearly labeled, with the terms and categories defined so that they are intelligible to the intended audience. The medium could be a published report; a press release, with supporting technical documentation; or a document on the surveillance program's website. |

| **Informing Policymakers about Birth Defects Issues** | |
|---|---|
| **Audience** | Legislators or policy makers |
| **Objective** | To support efforts to increase health services or justify continuation of funding for the surveillance program itself. |
| **Message** | The magnitude of a problem or the resources needed to maintain a surveillance program. |
| **Communication Medium** | Clear, succinct bulleted text with supporting graphs and tables. |

| **Responding to Community Members about Birth Defects Clusters** | |
|---|---|
| **Audience** | Community members |
| **Objective** | To respond to concerns about birth defects clusters |
| **Message** | Relationship (if any) between birth defects clusters and environmental hazards |
| **Communication Medium** | Established state protocols for dealing with this issue and including description of how information regarding the cluster and its investigation is communicated to concerned stakeholders. Important to communicate information to the community, both during the investigation and at its conclusion, using clear and simple messages (Williams et al., 2002a). |

# 11.5  Stage 4 – From Knowledge to Action

| ACTORS | NATURE OF PRODUCT | PRODUCT TYPES | PRESENTATION MODE |
|---|---|---|---|
| **Action Takers** | **Actions that are:** | **Action Types** | |
| • Data reporting staff | • Appropriate | • Estimating frequencies | • Surveillance reports |
| • Surveillance staff | • Evidence-based | • Referrals to services | • Websites |
| • Policymakers | • Maximally effective and cost-effective | • Planning services | • Scientific publications |
| • Decisionmakers | | • Planning interventions | • Policy papers |
| • Intervention designers/ implementers | | • Conducting research | • Guidelines |
| • Health care providers | | • Cluster investigations | • Intervention protocols |
| • Media | | | • Risk communication |
| • Families | | | • Press releases, media articles and shows |
| • Community members | | | |
| • Fellow scientists/ researchers | | | |

As mentioned earlier, the key themes of the CDC definition of *surveillance* are the integration of data collection, analysis, interpretation, dissemination, and application. In the previous section we spoke of dissemination of information with an eye toward application, whereby knowledge capable of informing action is developed. In this section, we speak of application or the undertaking of action(s) in order to achieve programmatic objectives. To illustrate this stage, we present a vignette of a surveillance program as it moves through different developmental phases (nascent, developmental, mature) and how the data produced at each phase of a program's development can be mobilized to inform action.

The stage of development of a registry or surveillance program has important implications for data presentation. The following vignette describes the experiences of one program director in this regard. The text is in the first person to reflect the program director's appraisal of the events surrounding the presentation of data to different audiences at different developmental stages of the program and with different types of action likely to result.

> *In my experience, the quality of our data increased dramatically from our initial data set to the second and has increased incrementally thereafter. We are continuously evaluating our methods and data, with the goal of being more complete, more accurate and reducing bias. Nonetheless, I believe all of our data have had some value and were worth presenting to selected audiences.*

> *I received our first data set the day I was asked to take responsibility for the State of Contentment's birth defects surveillance program. I was handed a flexible folder that in essence*

*was the registry. In it were a couple floppy disks, several sheets of paper with diagnoses listed on them, and a couple of envelopes containing various parts of copied discharge sheets. Not an ideal data set, but it was the result of a pilot project where hospitals in one region of the state reported their birth defects cases from one year to the department of health. The regional perinatal center had prepared a formatted Excel spreadsheet for the project data, but they were the only hospital to use it. While not standardized and not complete, these were the best data we had at the time.*

*We compiled the data into a table based on the tables of birth defects in the NBDPN annual report and presented them at a meeting organized by the local chapter of the March of Dimes. The meeting coincided with the March of Dimes' annual legislative lobbying day. It was a relatively informal meeting, and we provided handouts of the data to a mixed audience made up primarily of March of Dimes volunteers; a number of neonatal intensive care unit (NICU) nurses, geneticists, and neonatologists also attended the meeting. The March of Dimes was particularly interested in the data, as they had lobbied the legislature to establish a birth defects surveillance program, legislation which included authorization of the pilot project. The presentation was informal, accompanied by a warning that the data were very messy and likely to be incomplete. Nonetheless, the audience was enthusiastic. The volunteers asked a lot of questions, as did the professionals who also offered a good deal of advice. Among other things, I recall learning the importance of using standardized case definitions; the number of cases of patent ductus arteriosus was likely inflated because there was no control for low-birth-weight infants. The presentation was followed by a reception for the legislators whom the March of Dimes had lobbied earlier that day.*

*Following the meeting, I developed a plan to use data from our Hospital Discharge Data System linked with the Birth Certificate Data System to identify birth defect cases. This provided a state-wide population-based assessment. We did the extractions and linkages for a one-year birth cohort, the same year's data that were used in the pilot study. At the next March of Dimes annual meeting we presented the overall state data, along with a comparison of the regional pilot study data and the linked data. Once again there was a lot of give and take, and it was readily apparent that the linked data were more complete and accurate. With the birth certificate linkages, we also had considerable data on the characteristics and conditions of the birth population, the denominator for the calculation of strata-specific prevalence estimates. Once again the presentation was followed by a reception with the legislators. A year later a number of the legislators who attended the reception voted to provide funding for our plan to establish a state-wide birth defects surveillance program. The data were not perfect, but they clearly had value.*

*In the meantime, the single-year data were also submitted for the NBDPN annual report and presented at the opening of a state American College of Obstetricians and Gynecologists (ACOG) meeting. The ACOG meeting was formal with a PowerPoint presentation and the audience, primarily physicians and nurses, was very interested and inquisitive. The data showed specific birth defects rates that appeared high relative to national rates and differences among regions of the state. Much of the discussion following the presentation was on the possible reasons for the observed differences. Some of the hypotheses involved potential artifacts in the data, whereas others involved regional differences in behaviors and populations. Once again the interaction was informative for the presenter as well as the audience.*

*Subsequently we have given presentations at two American Public Health Association annual meetings; one presentation focused on a plan to evaluate the hospital discharge data, using active case/control reviews, and the other on risk factor analyses using the linked birth certificate and hospital discharge data. To date the program has compiled six years of population-based statewide surveillance data using the linked birth-hospital discharge data and two years of active*

*case/control reviews. A linkage of the two data sets and their evaluation should be completed soon and will likely provide greater depth and information than any of the previous presentations. The key point is that each of the above-mentioned data sets had both informative and intrinsic value when presented to the appropriate audience, along with clear warnings regarding the data's potential limitations.*

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# *Appendix 11.1*

# *Data Suppression*

# Appendix 11.1 Data Suppression

Specific birth defects are often rare events (sometimes extremely rare) leading to a set of issues that must be considered when presenting birth defects data. The public health professional must balance the potentially conflicting goals of information dissemination with protection of the privacy of persons in the community. When the number of cases in a diagnostic category within a group or stratum (such as race or sex) is small or the population from which the cases are determined is small, the risk of allowing a specific individual to be identified may be deemed too large to be acceptable. In such cases, steps must be taken to protect an individual's privacy.

The most common method of preventing the identification of specific individuals in tabular data is through cell suppression. This means not providing counts in individual cells where doing so would potentially allow identification of a specific person. Cell suppression can also be done by combining cells from different small groups to create larger groupings that reduce the risk of identifying individuals. While there are also more sophisticated data perturbation methods that use statistical noise to mask sensitive information, these are generally more suitable for use with economic or financial data than with public health data. This appendix reviews the basic methods, issues, strengths, and vulnerabilities of cell suppression. In addition to protecting privacy, prevalence information is often suppressed when concerns exist regarding possible statistical unreliability of estimates that are based on small numbers.

## Suppression Criteria

The first question is whether or not to suppress. Surveillance program administrators and technical staff should be aware that standards used to suppress data may already be set in state laws or in departmental or institutional rules and regulations. It is the responsibility of surveillance staff and administrators to be aware of these standards and practice within their limits. If standards are not established, it behooves a surveillance program to establish rules that will be followed consistently. This is best accomplished with the assistance of an advisory committee, an institutional review or privacy board, or a similar body.

Suppression rules are typically based on a predetermined criterion for the number of diagnosed cases and/or the number of births in the population or subpopulation from which the cases were identified. These numbers may also be thought of as the numerator and the denominator, respectively, of a prevalence estimate. Generally, suppression rules focus on the size of either the numerator or the denominator, the ratio of the numerator to the denominator, or the difference between the numerator and denominator. However the values that trigger suppression vary greatly from one institution or place to another, and there are no set standards. In practice, the rules used vary from relatively liberal to very conservative. Suppression rules for some of the population-based data systems used to assess progress toward the Healthy People 2010 objectives are presented in Table A11.1-1.

**Table A11.1-1 Data suppression rules for population-based data systems in the HP2010**

| Data System | Suppression Criteria |
|---|---|
| HIV/AIDS Surveillance System | < 4 cases |
| National Notifiable Diseases Surveillance System | Race and Hispanic origin if < 4 cases |
| STD Surveillance System | County: < 4 cases; State: < 6 Cases; National: None |

Source: Klein et al., 2002

Each of these suppression criteria is based on simple case counts, but they vary in terms of whether the suppression is of the overall counts or by substrata such as race/ethnicity or geography. In contrast, some surveillance programs will not report data on a birth defect if the case count is less than 5, regardless of the population size, whereas many regularly report single cases. When evaluating the prevalence of birth defects or investigating potential birth defects clusters, it is often necessary to consider birth populations that may consist of small numbers of births. In this situation information on individual cases may be essential to fulfill some of the program's public health functions but should not be included in formal reports.

While reporting small numbers of cases may threaten privacy, the threat may be greatest when reporting from small populations or when the difference between the number of cases and the population count is small. This has led to suppression rules that assess the difference between the prevalence numerator and the denominator or the case count and the population size (e.g., Land, 2001). For example, given the suppression criteria requiring a minimum difference of 15 and a single case of anencephalus in a birth population of 16, the denominator minus the numerator rule would allow the data to be shown. However, in a birth population of 15 the same data would not be shown. Given the nature of anencephalus, an alternative relevant event-specific denominator may be infant deaths in the population. In that case with a birth population of 16, a single anencephalus case would not be shown unless all the infants had died. Thus, even the seemingly simple question of the relevant population to be considered may not be straightforward and should be considered carefully in deciding when to suppress.

## Extent of Suppression

Having made the decision to suppress, the question becomes what and how to suppress. The solution that provides the greatest protection of privacy is to suppress an entire table whenever a single cell presents a threat, whereas the solution that provides the least protection is to suppress a single offending cell or only those cells deemed sensitive. Suppressing only sensitive cells is called *primary suppression*. However, when a single cell is suppressed, if column and row totals are provided, they can be used to compute the value of the suppressed cell. Similarly, suppressing multiple cells may allow the values of many or all of the suppressed cells to be revealed through a series of simple arithmetic solutions. This leads some agencies to practice *complementary suppression*, also referred to as *secondary suppression*, in which nonsensitive cells are suppressed in order to support the suppression of sensitive cells. If not properly done, however, the values or approximate ranges of cells in tables created with complementary suppression can also be obtained through the application of simultaneous equations (Geissing, 2001). Complex computer algorithms can be used to determine what cells must be suppressed in order to protect sensitive information. However, these algorithms are not always effective and become excessively complex in large tables (Duncan et al., 2001). One also confronts the issue of increasing data loss when large numbers of cells are used in complementary suppression.

## Threat of External Data

A final issue to be considered in deciding when and how to suppress sensitive information is the potential availability of data in multiple tables. It is not enough to simply evaluate the present table with its columns and rows; one must also consider the possible availability of complementary tables. This is especially true in the era of web-based interactive information systems that generate tables for custom queries on demand. Consider a hypothetical case where, in the process of creating a table for an annual report, it was determined that cells showing pyloric stenosis counts for the black population were potentially sensitive and the decision was made to provide only the total number of cases. Subsequently it is determined that effectively suppressing the black population's case counts would require

complementary suppression of the white population's case counts. Given that the white population's data were not sensitive, they may be subsequently published in a separate table. If so, the resulting data could be combined with the original table in order to reveal the black population's data. A similar situation would arise if, to protect privacy and present all of the data, the population strata were collapsed and subsequently data for one of the strata were published.

## Summary on Suppression

The more restrictive a suppression rule, the less information a given table or report will provide. The weaker a suppression rule, the greater the potential threat of revealing confidential health information. It is a question of balancing the threat to individual privacy with the public health value of presenting the data. Overall, deciding when and how to suppress birth defects information is more a social, political, and legal issue than a technical one. The technical aspects are quite straightforward, but the contextual and procedural/policy issues are likely not to be. These all need to be considered and balanced in the local context before informed decisions can be made to suppress or not to suppress data in program reports or other documents.

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# *Appendix 11.2*

# *Use of Geographic Information Systems (GIS) to Map Data*

# Appendix 11.2 Use of Geographic Information Systems (GIS) to Map Data

The application of Geographic Information Systems (GIS) methods has become an integral component of aggregating, analyzing, and evaluating health data. The current practical applications of GIS in epidemiologic studies range from descriptive statistics (i.e., plotting data on a map) to evaluation of spatial relations between environmental exposures and health outcomes.

Several definitions exist for geographic information systems. One of the most recent, as found in *Healthy People 2010,* defines GIS as "powerful tools combining geography, data and computer mapping" (U.S. Department of Health and Human Services, 2000). Software packages available today, such as ArcMap and MapInfo, integrate many GIS functions. These include (1) database management, (2) data manipulation and analysis, and (3) data presentation (i.e., displaying data on a map). To be included in GIS, the data should have some kind of geographical or spatial component that can be translated into digit maps.

## Digital Map Formats

GIS applications use either a vector or a raster map format, or a combination of the two. In *vector* maps (Figure A11.2-1) geographic features are represented by points (e.g., location of infants with birth defects), lines (e.g., streets), and polygons (e.g., census tracts) (Rogers, 1999). These features are based on latitude and longitude coordinates of the different objects. The vector format is the most commonly used in public health. In *raster* maps the data are stored as digital images (e.g., orthophotos, scanned maps) (Vine et al., 1997). Usually a grid cell is used to represent a feature, and these cells can be connected. As such, smaller cells provide a more detailed resolution. Obtaining quality maps for a given geographical area for the time period of interest is crucial as maps are static while environments change.

## Bringing Health Data into GIS

Ultimately, the application of GIS to birth defects data requires the transformation, as accurately as possible, of health records containing addresses or location information into geographic objects. This process is called *geocoding*, also known as address matching. During geocoding, latitude and longitude coordinates are assigned by the GIS software to each address by matching against an *address-range* (i.e., street segment) in a street reference map such as the Census Topologically Integrated Geographic Encoding and Referencing (TIGER) files (Croner et al., 1996). Interpolation is used to estimate the actual address location within the given range (Rushton, 1999). The address match rate depends on several factors, including the completeness of addresses in health records and the accuracy of reference maps (McElroy et al., 2003). In case of incorrect or missing house numbers and/or street names, coordinates are usually assigned to a centroid of a larger geographical entity, such as a census tract or a ZIP code. If available, other reference files such as tax parcel databases can also be used for geocoding purposes. Alternatively, in areas where latitude and longitude coordinates have not been predetermined (e.g., rural communities), a global positioning system (GPS) device can be used, although this may prove time and resource demanding. Figure A11.2-2 shows an example of how a point is placed within an address range.

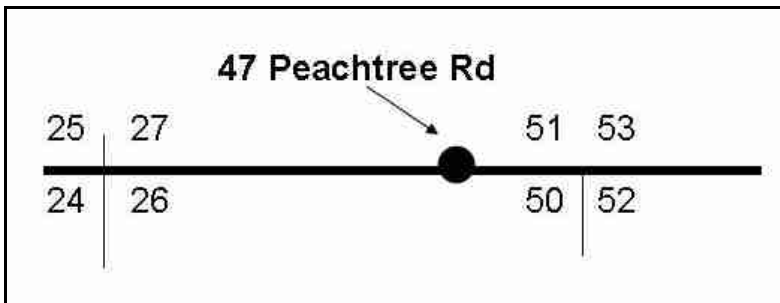**Figure A11.2-1 An Example of GIS Data Layers**
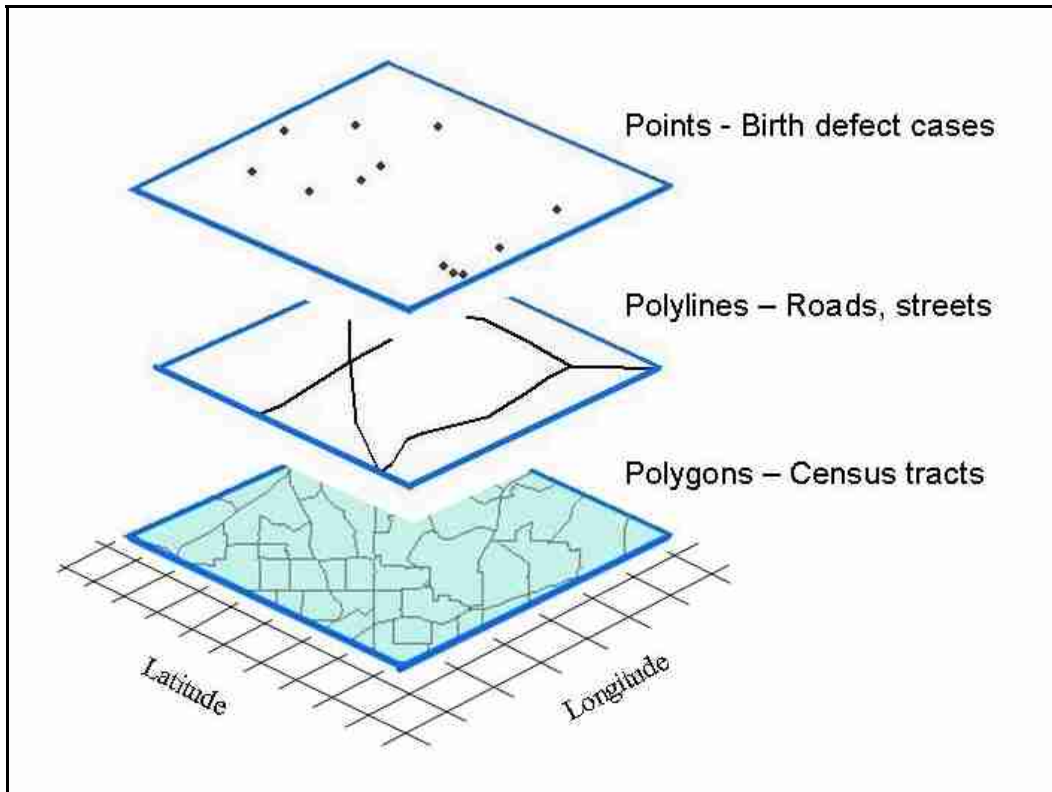


**Figure A11.2-2 A Specific Location Within an Address Range**

## Mapping Data

Once health data are brought into a GIS database, users need to be aware of several important issues in data mapping. For example, different spatial databases must have the same scale and projection (McLafferty and Cromley, 1999). Otherwise data will be distorted or cannot be mapped together. *Map scale* shows the relationship between a unit of length on a map and the corresponding length on the ground. It is also an expression of how much the area represented has been reduced on the map. The smaller the scale, the larger the area displayed on a map. *Map projections* are attempts to portray/transform the surface of the three-dimensional earth or a portion of the earth on a flat map using a mathematical model. Some distortions of conformality, distance, direction, scale, and area always result from this process. Maps that focus on maintaining one feature (e.g., preserving distance) must distort other features (e.g., area, shape). Maps that accurately reflect area are called *equal-area maps*, while maps that correctly show the distance between points are called *equidistant maps*.

Two types of maps frequently used in public health research are dot-density and choropleth maps (Rogers, 1999).

**Dot-density mapping.** *Dot-density maps* are the simplest way to display events. These maps use dots or other symbols to represent the number of occurrences of a given data characteristic (Thrall, 1999). Each dot or symbol used on the map may represent a single entity (one dot = one case) or a group (one dot = 1,000 people). Dot-density maps are useful for area comparisons. However, dot-density maps need to be interpreted with caution regarding the "symbol to data characteristic" ratio. It is also important to keep in mind that dots do not always indicate the exact location of the data. An example of a dot-density map for metropolitan Atlanta is presented in Figure A11.2-3 (Source: Siffel et al., 2006, Figure 1, p. 828).

**Figure A11.2-3 A Dot-Density Map of Metropolitan Atlanta**
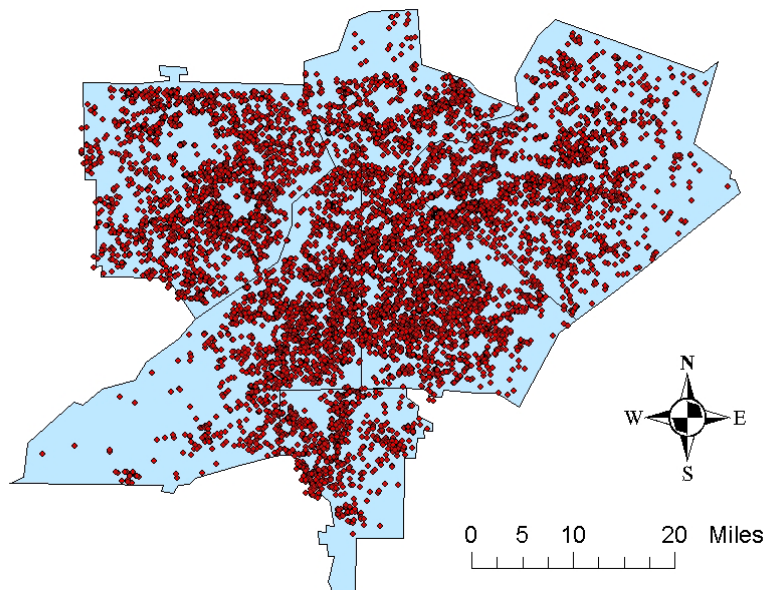
**Choropleth mapping.** *Choropleth maps* are area maps in which polygons (e.g., census tracts, counties) are shaded, colored, or patterned according to the extent to which a given attribute (such as population size or disease rate) is associated with each polygon. Choropleth maps are also called *thematic maps* or *shaded maps*. An example of a choropleth map for metropolitan Atlanta is presented in Figure A11.2-4 (Source: Siffel et al., 2006, Figure 2, p. 828).

It is important to choose the right characteristics for map presentations as the choice of color, pattern, size, polygon shape, and class intervals can impact how one interprets the information presented in a map. Single-color maps with varying color intensity (shades) are often an effective means of presenting data, but the use of differing patterns can help a black-and-white or grey-scale map. Similar-size polygons are recommended to the extent possible, as a few large polygons can dominate a map, leading to misinterpretation of information. Proportions or rates can be displayed by different class interval schemes, such as equal intervals (equal ranges of values) or quintiles (equal number of polygons falling into each class defined by dividing the range of values). The latter method is particularly useful for presenting skewed data. These methods are standard in GIS software.

**Figure A11.2-4 Choropleth Map of Infants per Census Tract in Metropolitan Atlanta, 1990**
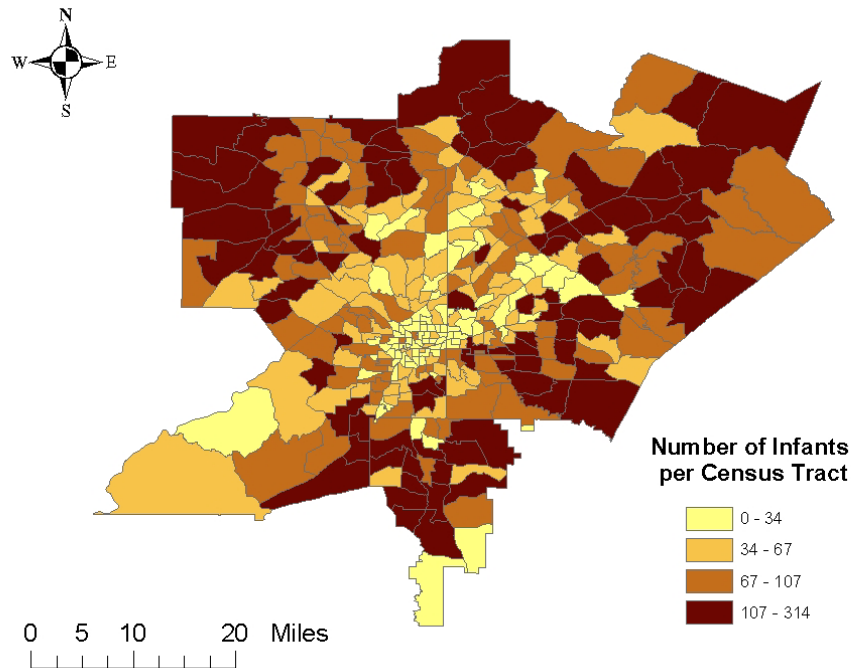
## Additional Technical Details

Maps showing point locations or even aggregate data in a small geographic area have the potential to reveal the identity of individuals (Cox, 1996). Therefore, as noted elsewhere in this chapter (see Appendix 11.1 on Data Suppression), one must generally limit the presentation of disaggregated birth defects information. While GIS methods and techniques exist for protecting privacy and limiting disclosure of information by geographically masking individual records (Armstrong et al., 1999), the use of masked data in small-area analysis can limit one's ability to detect clusters of cases (Kamel Boulos et al., 2005). As such, careful choice of geographical units and data aggregation are vital.

Below we present several practical suggestions for preparing and presenting maps above and beyond those already mentioned.

- ➢ The use of the same scale, colors, class intervals, and legends when presenting a series of maps.

- ➢ The inclusion of a scale bar and a "North" arrow.

- ➢ The use of patterns when printing in black and white. Color maps produced on a black-and-white printer usually do not provide as good results as grey scale.

- ➢ Avoid the use of red and green on the same map.

- ➢ Be wary of font-related problems. If symbols, which are special GIS fonts, are used on a map, do not export the map as an MS Windows meta file (.wmf). This type of file requires access to the GIS fonts. Similarly, do not include such files in presentations being made on an unfamiliar computer. If the GIS fonts are not available, other fonts will be substituted for symbols in the image. Instead, export your maps as JPEG files.

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# *Appendix 11.3*

# *Data Users Matrix*

# Appendix 11.3 Data Users Matrix

| Sample Questions Asked | Information Needs/Data Presentation Suggestions |
|---|---|
| **Surveillance Program Staff** | |
| *This group is likely to require process indicators useful for management. Members of this group may notice possible clusters. This audience also needs additional information about denominator issues and data quality.* | |
| ▪ How many abstracts were completed per field staff person?<br><br>▪ We have noticed more cases of birth defect x in this hospital; is that unusual? | ▪ Data on labor hours and abstracting rates.<br><br>▪ Data on birth prevalence, usually in comparison to some standard, such as the entire state.<br><br>▪ Internal exhibits in terms of surveillance parameters: Completeness/Ascertainment, Case Processing Times. |
| **Researcher** | |
| *This audience is likely to be interested in:*<br>   o *Descriptive epidemiology (describing occurrence of birth defects by person, place, and time).*<br>   o *Analytic epidemiology (finding causes of birth defects).*<br>   o *Obtaining birth defect cases for related studies.*<br>   o *Methodological issues.*<br>   o *Availability of data on individual types of birth defects and on cases, both identified and de-identified, for ecologic studies and etiologic research.* | |
| ▪ What are the patterns of birth defect occurrence by person, place, and time?<br><br>▪ What is associated with risk of birth defect x?<br><br>▪ How many cases of specific birth defects are represented in the database of the surveillance program?<br><br>▪ How do I get access to records for persons included in the surveillance program for research studies? | ▪ Printed and web reports and charts with routinely tabulated results allow for quick response to many inquiries.<br><br>▪ Specially tabulated results<br><br>▪ Line item data with and without confidential information.<br><br>▪ Interactive web-based tool for custom queries allows for easy access and reduces staff time in responding to simple routine and non-routine aggregate data requests. See, for example, http://soupfin.tdh.state.tx.us/bdefdoc.htm |

| Sample Questions Asked | Information Needs/Data Presentation Suggestions |
|---|---|
| **Physician and Other Health Care Providers** | |
| *This audience is often interested in outcomes (pregnancy, how does the child do, survival/treatment), as well as birth defect prevalence. Members of this group may notice possible clusters. This audience also needs additional information about denominator issues and data quality. Statistical output for this audience may well run more detailed in terms of clinical detail. Allow for the presentation of small cases series. Members of this group often have the skill to interpret tabular data, but do not assume they will necessarily have the statistical sophistication that may be lacking in other audiences.* | |
| <ul><li>Which defects are most common?</li><li>What are the trends in birth defects over time?</li><li>Are there unexpectedly high rates in my area or facility?</li></ul> | <ul><li>Printed reports.</li><li>Web-based data.</li></ul> |
| **Social/Education Service Provider** | |
| *Needs from this audience are usually geographic in nature. Birth defects programs can promote use of data among this group by providing maps and other data that respond to these information needs.* | |
| <ul><li>Describe the population we serve in terms of location, income, other variables.</li><li>How many babies with complex congenital heart defects do you estimate will be born during the next five years in the area for which our children's hospital provides clinical care?</li></ul> | <ul><li>Mapping location of clinics compared to appropriate birth defect cases.</li><li>Time series analyses with projections (e.g., how many children with x syndrome will be born in x county for the next five years in order to project needs for special education teachers, etc.?).</li></ul> |
| **Local Health Department** | |
| *This audience tends to be interested in epidemiological data. Members of this group may also want to link your data with data they have on environmental concerns (e.g., factories, toxic waste sites). There is a risk of misuse of data if users do not understand the unique aspects of birth defects data.* | |
| <ul><li>What are our rates?</li><li>How do we compare with the rest of the state?</li><li>How do we compare with the nation?</li><li>Are there links between birth defect clusters and local environmental concerns?</li><li>What clusters (in our area) are you dealing with?</li><li>What are the trends in birth defects over time?</li></ul> | <ul><li>Epidemiologic data provided by zip code, city, county, region.</li><li>Case characteristic summaries as tables, as well as rate tables and graphics, including trends. Consider also the presentation of rate ratios.</li><li>Reports of cluster investigations; maps of clusters investigated.</li><li>Have a document written in language accessible to the lay person explaining some of the finer points of interpreting birth defects data.</li></ul> |

| Sample Questions Asked | Information Needs/Data Presentation Suggestions |
|---|---|
| **Maternal and Child Health Programs** | |
| *Members of this group might be professionals in Women Infants, and Children (WIC) or Title V programs. They tend to be interested in trends over time, rates, outcomes, and surveillance operations.* | |
| ▪ Do you have any information on folic acid education programs that can be targeted to our Hispanic clients? | ▪ Items here will likely address racial disparities and the demographics that underlie differences in race, including cultural health practices and SES. <br> ▪ GIS/spatial analysis may be relevant here. |
| **Family of Child with Birth Defect(s)** | |
| *This audience tends to like information about known causes and risk factors for particular birth defects. Members of this group are interested in seeing birth defects data linked with potential teratogens or environmental concerns and the outcomes. They may desire information on educational, social, and clinical services available for children with specific types of birth defects.* | |
| ▪ I have a child with a cleft lip. What information do you have on the causes of this condition? <br> ▪ I just had a baby with spina bifida. What information can you give me about this condition and where can I get specialized care for her? <br> ▪ Do you have any information on support groups in my community for parents of children with Down syndrome? <br> ▪ What caused my child's birth defect? <br> ▪ Have there been clusters investigated in my area? | ▪ Literature summaries. <br> ▪ Maps of cluster investigations. <br> ▪ Charts comparing local/state rates to other areas. <br> ▪ Be sure to explain the difference between individual- and population-level information |
| **Students (public health, medical, nursing, allied health, or other college/university)** | |
| *Needs among this audience might include:* <br>   o *General education (e.g., lectures to a class)* <br>   o *Specific education (e.g., practicum placements)* <br>   o *Research (e.g., data and guidance on papers, theses, dissertations)* <br><br> *You might present to these types of students during Grand Rounds or at local seminars or conferences. They may also submit specific requests as a result of papers or projects they are working on.* | |
| ▪ Can you provide me with information about changes in the occurrence of neural tube defects in ___ following the fortification of cereal grains with folic acid? | ▪ Printed reports. <br> ▪ Copies of manuscripts. <br> ▪ Interactive web-based tool for custom queries allows for easy access and reduces staff time in responding to simple routine and non-routine aggregate data requests. <br> ▪ Raw data. |

| Sample Questions Asked | Information Needs/Data Presentation Suggestions |
|---|---|
| **News/Media Person** | |
| *Media personnel are generally interested in comparative rates region/state/nation. Also, they will likely need information about causes and risk factors. Their questions may or may not relate to a specific community concern or cluster. They tend to request large amounts of data and use very little of it.* | |
| ▪ What is the cause of x birth defect?<br><br>▪ Are the cases of x birth defect linked with the toxic dump, military base, factory, vaccine, etc? | ▪ Printed and web reports and charts with routinely tabulated results allow for quick response to many inquiries.<br><br>▪ Have a document written in language accessible to the lay person explaining some of the finer points of working with and interpreting birth defects data. |
| **Legislator/Policy Maker** | |
| *This type of information request usually comes from higher up in the agency or from advocacy groups. Top information needs include cost of program, cost of birth defects to the state, number of people served by the program, and staffing data. This audience would be interested in surveillance data connected to other information such as regional variation or costs. Members of this group may also request administrative data—improvements in program efficiency, budget information, increases in caseload (live births, hospitals).* | |
| ▪ How many staff (FTEs) does the surveillance program employ?<br><br>▪ What is the cost of the program?<br><br>▪ How many babies with birth defects are born each year in my legislative district?<br><br>▪ What are the estimated lifetime costs of caring for a child with spina bifida?<br><br>▪ What is the impact of this policy? (e.g., decrease in rates of NTDs after fortification of food supply with folic acid) | ▪ Narrative reports.<br><br>▪ Graphs of birth prevalence over time (for before/after comparisons) or comparing communities with different policies.<br><br>▪ Statistical exhibits intended for this audience should address not only descriptive aspects, but also the quantitative burden of disease. |
| **Advocacy Group** | |
| *The needs of this group will vary depending on what they are advocating for. This audience will need exhibits at two levels—exhibits for the lay public and exhibits for policy makers—and the distinction needs to be clear.* | |
| ▪ What are the rates in z location for birth defect x?<br><br>▪ Why are you not collecting data on birth defect x?<br><br>▪ What is the cost to y unit of government for treating birth defect x? | ▪ Cost, magnitude, impact.<br><br>▪ Birth defects data linked with environmental data. |

| Sample Questions Asked | Information Needs/Data Presentation Suggestions |
|---|---|
| **Other Community Members** ||
| *Members of this group usually contact the registry to report concern of a possible cluster. They are frequently interested in environmental exposures and birth defects.* ||
| ▪ What are the rates of birth defects in my community?<br>▪ Are birth defects higher here than elsewhere?<br>▪ If higher, can the excess birth defects be linked to environmental concerns? | ▪ Data on occurrence, usually in comparison to some standard, such as the entire state. |

# *Appendix 11.4*

# *What Type of Chart or Graph Should I Use?*

# Appendix 11.4 What Type of Chart or Graph Should I Use?

Research illuminating human perceptions of graphical representations offers us clues as to how to select the best representation for a given type of data. Below we present examples of the most common types of graphs and charts, along with suggestions on when they might be used.

*Pie charts* can be effective for communicating simple proportions (see Figure A11.4-1). When comparing several proportions, convention dictates that none of the radii should be at the "12 o'clock" position (Hollands, 2003). Pie charts do not need legends, instead the series name and percentage should be positioned next to the appropriate slice.
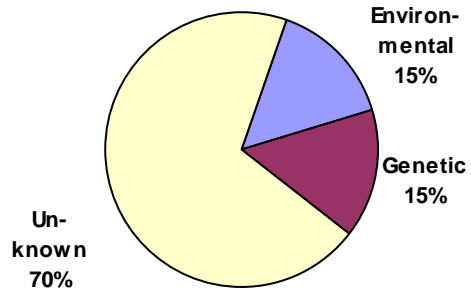


**Figure A11.4-1 What causes birth defects?**



**Figure A11.4-2 Pregnancy Outcomes, Down Syndrome**
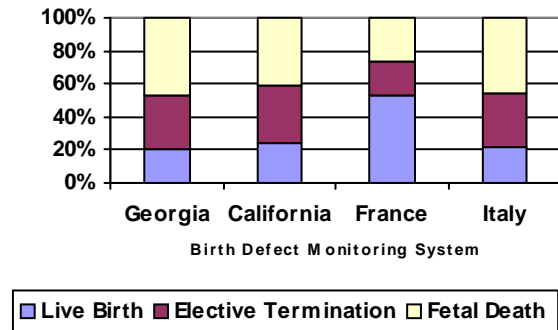
*A 100% stacked bar chart* can be used to compare proportions between two or more data sets (see Figure A11.4-2). However, be careful about presenting too many data points as the graph may become too busy to convey information effectively. An alternative would be a series of several pie charts, although 100% stacked bars allow for more consistent comparisons.

¹**Trend is significant; details are given in Table 3**

Time series are nearly always demonstrated using a *line chart*, with a marker at each year. Figure A11.4-3 includes a regression line indicating that the change in rates is indeed statistically significant, thereby adding important information to this chart.

**Figure A11.4-3 Rates of Hirschsprung disease 1989–2001**

Certainly the most common need for graphical representations of birth defects data is the comparison of rates of cases among persons and places. *Bar charts* are an ideal choice for this because they give an impression of relative differences but, unlike line charts, do not give the impression that moving left-to-right is a time progression (see Figure A11.4-4). (Note: This chart also demonstrates the use of white breaks in the bars in lieu of gridlines across the whole plot area.)
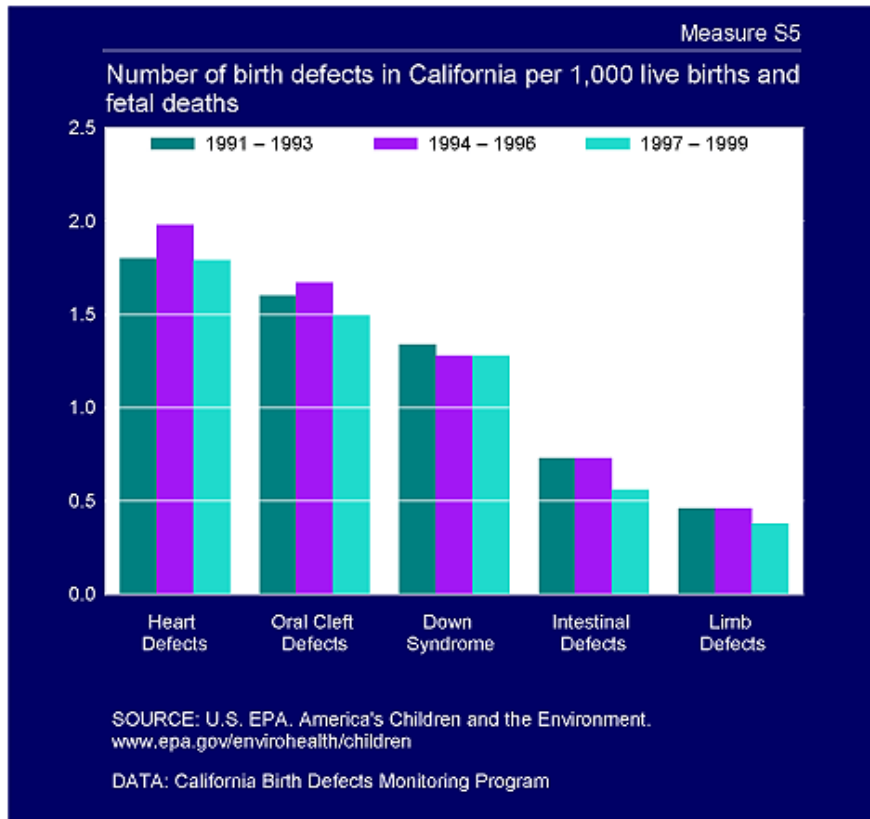


**Figure A11.4-4 Bar chart demonstrating cases per 1,000 live births**

Bar charts can also be used to convey information about the statistical significance of rates by using drop lines to represent confidence limits, as in Figure A11.4-5. Note: the best way to ensure that confidence limits are represented correctly is to import the results directly into your graphic software from your analysis software. However, it is also possible to produce the irregular confidence limits found when using Poisson regression in Microsoft Office products (see the document "Plotting Irregular 95% Confidence Intervals" on the Members Only section of the NBDPN website).

**Turner Syndrome 1999-2004**
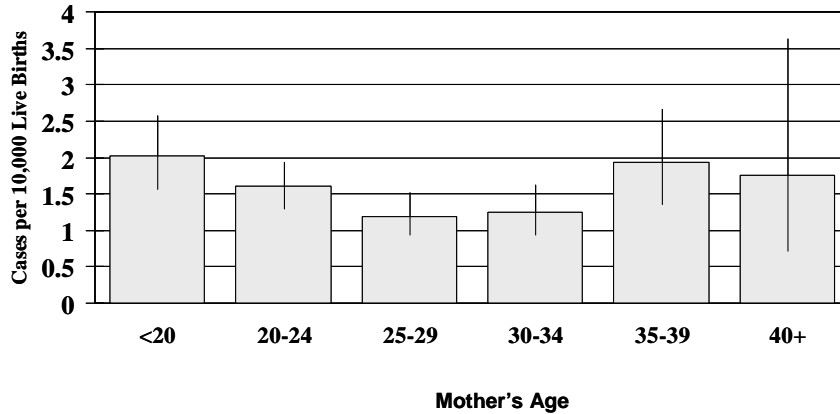


**Figure A11.4-5 Bar chart with confidence limits**

Figures A11.4-6 and A11.4-7 below, respectively, present examples of maps of epidemiological data. For further detail on the use of Geographic Information Systems see Appendix 11.2.
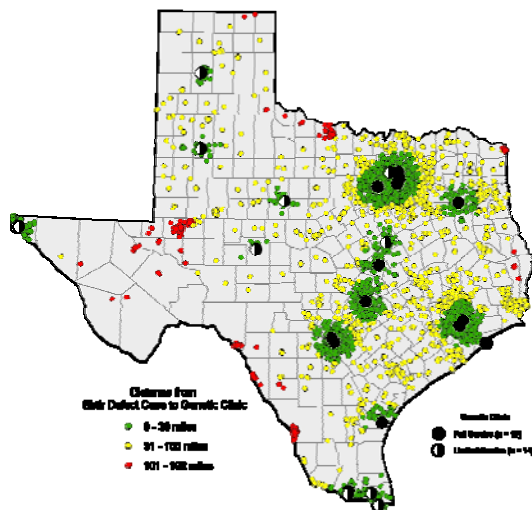


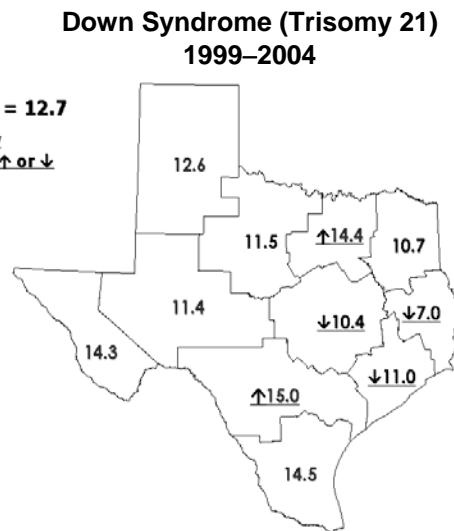**Figure A11.4-6 Spot Map**



**Figure A11.4-7 Area Map**

When places are not contiguous or for some other reason would be difficult to display on a map, a bar chart such as Figure A11.4-8 would be suitable.
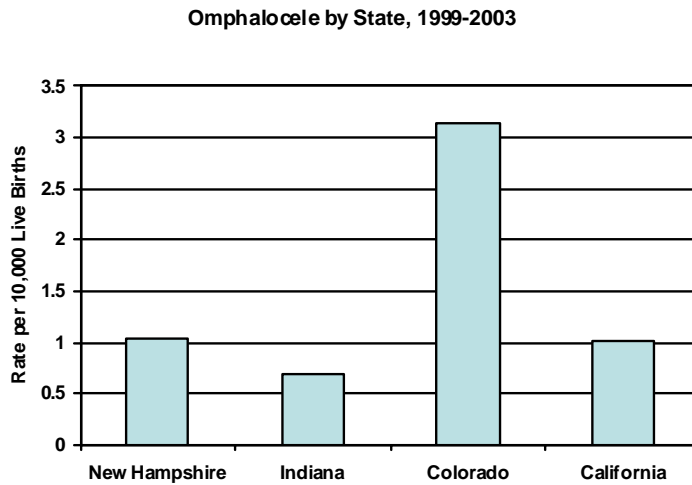
**Omphalocele by State, 1999-2003**



**Figure A11.4-8 Displaying geographic data with a bar chart**

When only the general place rather than a specific site is relevant (e.g., entire state versus specific regions or locales within the state), it is possible to use an area map (see Figure A11.4-9).
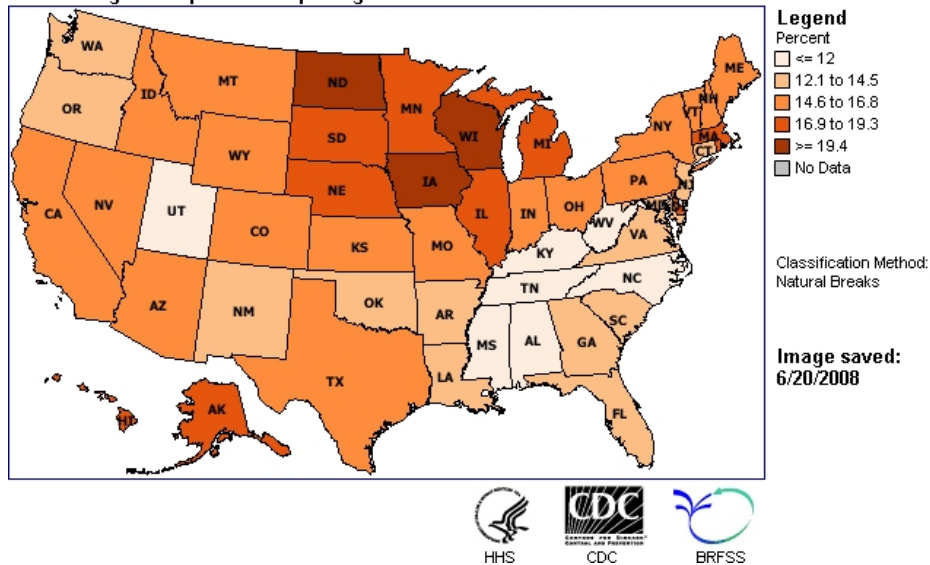


**Figure A11.4-9 Displaying geographic data in an area map**

In Table A11.4-1 below you will find one scheme for selecting the appropriate graphic representation given the type of data you will be presenting.

Before making your final decision, however, you should also ask yourself two questions that relate less to the nature of your data and more to your own personal preferences and the needs/interests of your audience:

➤ *Am I comfortable explaining this graph?* If the answer is no, find an alternative format with which you are more comfortable.

➤ *Given my audience, should I sacrifice detail for clarity, or clarity for detail?* For example, an audience of foster parents would probably benefit from clarity, whereas an audience of epidemiologists will readily comprehend your meaning and will rather be looking for additional detail about methods or sample characteristics.

**Table A11.4-1 Selecting a method of illustrating epidemiologic data (adapted from *Principles of Epidemiology*, 3rd edition, U.S. Department of Health and Human Services).**

| If Data Are: | | And These Conditions Apply: | | Then Choose: |
|---|---|---|---|---|
| Proportions | | < 6 data points | 1 series | Pie chart (Sample Figure 4) |
| | | | >1 series | 100% stacked bars (Sample Figure 5) |
| | | 6+ data points | 1+ series | Consider combining data point categories or table. (Sample Figure 5) |
| Time Series | | Numbers of Cases | | Line chart (Sample Figure 6) |
| Data with discrete categories | | | | Bar chart (Sample Figures 7, 8) |
| Place | Number of cases | Not readily identified on map | | Bar chart (Sample Figure 11) |
| | | Readily identified on map | Specific site important | Spot map (Sample Figure 9) |
| | | | Specific site unimportant | Area map (Sample Figure 10) |
| | Rates | | | Area map (Sample Figure 12) |

**Cited References on Graphic Presentation**

Hollands JG. The classification of graphical elements. *Canadian Journal of Experimental Psychology.* 2003;57(1):38-47.

Centers for Disease Control and Prevention, Office of Workforce and Career Development. *Principles of Epidemiology: An Introduction to Applied Epidemiology and Biostatistics.* 3rd edition. Atlanta, GA: U.S. Department of Health and Human Services, Centers for Disease Control and Prevention; n.d.