Facts about Congenital Central Nervous System / Neural Tube Defects

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Some congenital Central Nervous System (CNS) defects also are referred to as Neural Tube Defects (NTD). The neural tube forms and closes during the first month of pregnancy. The upper part of the neural tube forms the brain and skull, while the lower part forms the spinal cord and back bones. Congenital Central Nervous System defects can vary from mild (such as slightly smaller head size (mild microcephaly)) to severe (such as missing parts of the brain and skull (anencephaly)).

Signs and symptoms for congenital central nervous system/neural tube defects depend upon the type and severity of the particular defect. Anencephaly, and most cases of encephalocele, spina bifida, hydrocephalus, and microcephaly will be apparent on prenatal screening tests and ultrasounds. However, if prenatal care was limited, these congenital defects are usually readily visible at the time of birth. In some mild cases, for example, spina bifida occulta, diagnosis may not occur until adulthood.

Congenital Central Nervous System/Neural Tube defects

- Occur within the first 4 weeks of pregnancy.
- Risk has been greatly reduced by adequate intake of Folic Acid.
- May affect mental and physical abilities.

Diagnosis

Some congenital CNS/NTD defects may be diagnosed during pregnancy depending upon the severity of the defect and the level of prenatal care. During the first trimester a maternal blood test for AFP (alpha fetal protein) is used to screen for neural tube defects – this test is usually part of a larger maternal blood test, which may be called the "Triple Screen". An ultrasound also may show if a CNS/NTD defect is present. Once the baby is born and the doctor suspects a CNS/NTD defect, then the doctor will usually perform other imaging tests, such as X-ray, MRI, or CT to confirm the diagnosis.

Fetal development

The Central Nervous System begins to develop about 3 weeks after conception and will continue to develop throughout the entire pregnancy. It is the first organ system to begin developing.

Development begins with the formation of special cells that will eventually become part of the central nervous system. As the embryo continues to grow, a groove (neural groove) forms along the back of the embryo. This groove will close during week 4 to form the neural tube. The upper end of the neural tube will form the brain, while the lower end will form the spinal cord.

The early development, that is, before most mothers know they are pregnant, and the continued development of the CNS throughout the pregnancy, makes the CNS vulnerable to defects. Anencephaly, encephalocele, and spina bifida all result from the neural tube not properly closing causing part of the central nervous system to be exposed outside the body.

Known Causes

The cause of congenital Central Nervous System and Neural Tube defects among most babies is unknown. Some have defects due to changes in their genes or chromosomes. These congenital defects also are thought to be caused by a combination of genes and other risk factors, such as exposure to chemicals or viruses in the environment, maternal diet, or maternal medication use.

The Center for Disease Control and Prevention (CDC) continues to study congenital Central Nervous System and Neural Tube defects to learn how to prevent them. Many studies have shown that taking 400 micrograms per day of Folic Acid before becoming pregnant and continuing during pregnancy have greatly reduced the occurrence of Central Nervous System and Neural Tube defects.

Preventative Actions/Activities

Not all birth defects can be prevented, but studies have shown there are actions/activities a woman can take to increase her own chances of having a healthy baby. These actions/activities include:

- Take 400 micrograms (mcg) of folic acid every day for at least 1 month before getting pregnant to help prevent birth defects.
    - Folic acid is also in some fortified foods like cereal.
- Stop smoking and drinking alcohol.
- Do not use “street” drugs.
- Avoid overheating your body, as may happen if you use a hot tub.
- If you have a medical condition, be sure it is under control. Some examples of medical conditions include asthma, diabetes, oral health, obesity, or epilepsy. Also be sure that your vaccinations are up to date.
- Talk to a health care professional about any over-the-counter and prescription medicines you are taking. These include dietary or herbal supplements.
- Avoid contact with toxic substances, materials, or viruses (chicken pox, rubella (German measles), or cytomegalovirus) that could cause infection at work and at home. Stay away from chemicals and cat or rodent feces.
- If you have a fever, then treat right away with Tylenol® or equivalent store brand.
- See your health care provider if you are considering becoming pregnant or as soon as you think you may be pregnant
- Eat a healthy diet.

Examples of Specific Central Nervous System / Neural Tube Defects

- ***Spina Bifida***

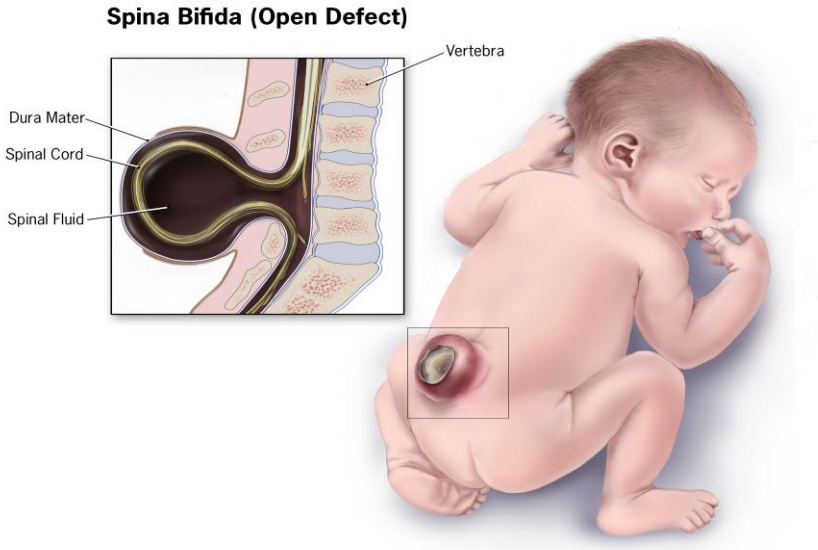
  Spina bifida is a Neural Tube Defect in which the neural tube does not completely close and may occur anywhere along the spine. Often there is damage to the spinal cord and nerves because the backbone, which protects the spinal cord and nerves, has not properly formed and closed. A child with spina bifida may have mild to severe mental and physical disabilities depending upon the size and location of the opening and its effect on the spinal cord and nerves.

  There are three common types of spina bifida. They are:

  - **Myelomeningocele:** This is the most serious type of spina bifida and usually results in moderate to severe disabilities. In myelomeningocele, part of the spinal cord and nerves are in the fluid filled sac that comes through the baby's back.
  - **Meningocele:** In this type of spina bifida the fluid filled sac does not contain the spinal cord or nerves. Hence, there is little or no nerve damage resulting in minor disabilities.
  - **Spina Bifida Occulta:** This is the mildest form of spina bifida with no visible opening or sac on the back. There usually are not any disbilities because the nerves have not been damaged. In many cases, this defect is not diagnosed until late childhood or adulthood.

  Diagnosis may be made prenatally depending upon the size of the defect and level of prenatal care through ultrasound or an AFP screening test. After the child is born, the spina bifida may be readily apparent or appear as a hairy patch or dimple on the child's back. An X-ray, MRI, or CT scan may be performed to finalize the diagnosis.

  Treatments for spina bifida depend upon the severity of the defect. In cases of spina bifida occulta, usually no treatment is needed. Myelomeningocele and meningocele cases require surgery. In some cases, surgery may be performed on the fetus (in utero) before being born. If surgery is performed after the child is born, the surgery is usually performed within the first 48 hours to minimize any nerve damage. Usually children with myelomeningocele will need ongoing care with a team of surgeons, physicians, and therapists.



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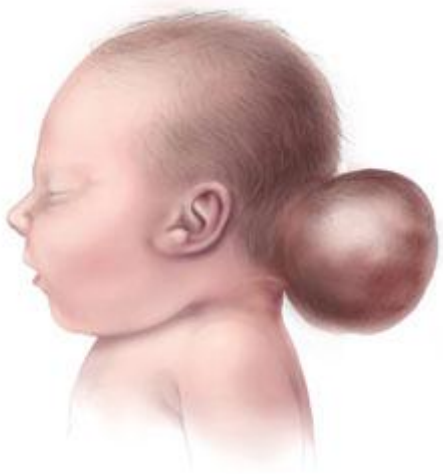
- ***Encephalocele***

  Encephalocele, similar to spina bifida, is caused when the neural tube fails to completely close. The sac-like protrusions, which contain part of the brain and the membranes that cover it, form along the midline of the skull, most notably in the back of the skull, the area between the nose and forehead, and upper part of the skull. If not diagnosed prenatally by ultrasound, an encephalocele is normally diagnosed immediately upon birth, however, a small encephalocele located in the nose and forehead region may go undetected.

  An encephalocele located in the back of the skull is usually associated with neurological problems. In addition, encephaloceles are often accompanied by other craniofacial abnormalities and brain malformations. Symptoms associated with encephalocele may include excess accumulation of fluid in the brain, a small head, paralysis of the arms and legs, difficulty in controlling voluntary muscle movements, developmental delays, vision problems, mental and growth retardation, and seizures.

  Surgery is the only treatment for an encephalocele. The exact type and extent of the surgery depends upon the location of the encephalocele and the type of tissue contained within the sac.



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- ***Anencephaly***

  Anencephaly occurs when the upper part of the neural tube fails to completely close. In most cases the child is born with the front part of brain and the part of the brain responsible for coordinating activities missing. The remaining parts of the brain that are present are not covered by the skull. Children born with anencephaly die shortly after birth.

  A child with anencephaly may be diagnosed during pregnancy depending upon the level of prenatal care. Anencephaly will be immediately diagnosed upon birth.

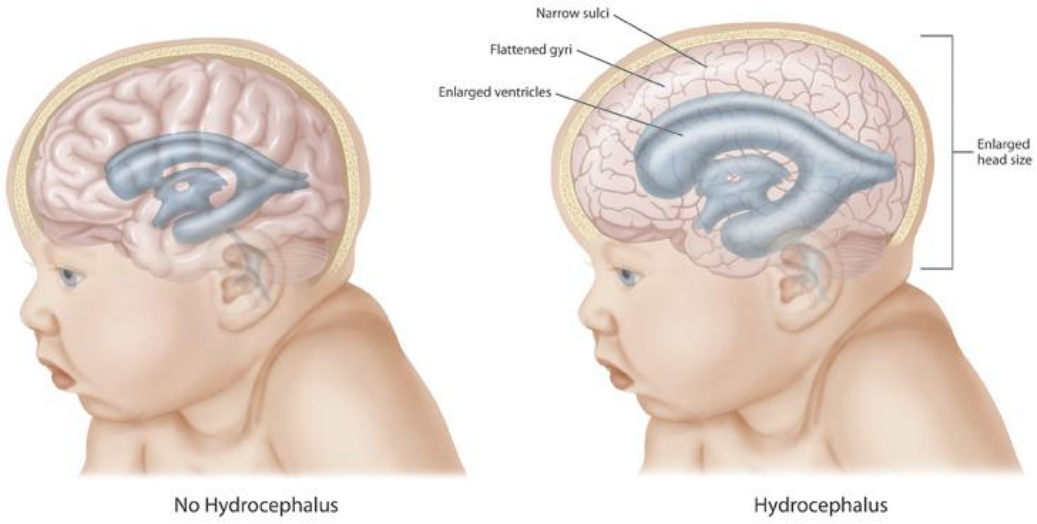
This image was provided by the Centers for Disease Control and Prevention, National Center on Birth Defects and Developmental Disabilities

- ***Hydrocephalus***

  Hydrocephalus occurs when more cerebral spinal fluid (CSF) is made than can be absorbed back into the body. This causes the ventricles within the brain to swell and put pressure on the brain. In general, there are two types of hydrocephalus: congenital and acquired. A child is born with congenital hydrocephalus, while acquired hydrocephalus develops after the child is born – this description will only address congenital hydrocephalus.

  Congenital hydrocephalus may occur by itself or be associated with other defects or syndromes. Some cases of hydrocephalus are linked to genetic causes. Hydrocephalus may be diagnosed prenatally by ultrasound or other imaging techniques such as ultrafast fetal MRI. Once the child is born, other imaging techniques may be used, too. A child with congenital hydrocephalus may be born with an enlarged head or the size of the head may grow rapidly after birth.

  Treatments for hydrocephalus and their prognosis depend upon the type of hydrocephalus and its cause. One treatment is a surgery that places a shunt (tube) within the ventricle to drain the excess CSF. If diagnosed prenatally, the doctors may decide to deliver the fetus at 32 weeks and perform the surgery to prevent further damage to the brain. A child with hydrocephalus has an increased risk to develop mental and physical disabilities that may require continuing professional help.



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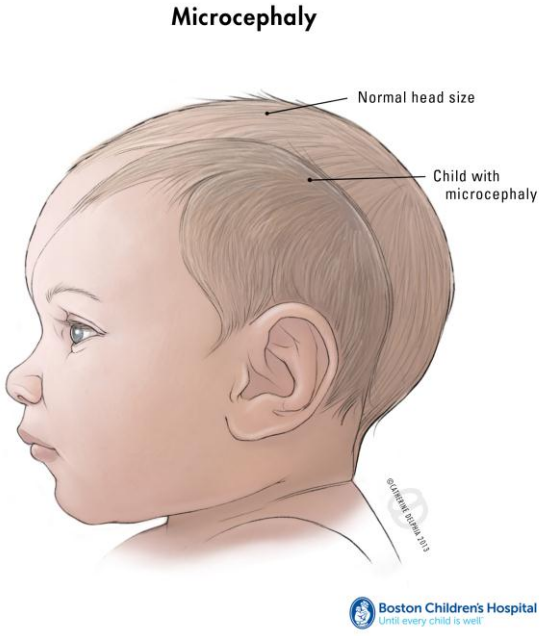
- ***Microcephaly***

  Microcephaly is usually present at birth and occurs because the developing brain does not grow properly resulting in a smaller than normal head size. The smaller growth of the brain may have been caused by exposure to hazardous substances or drugs, certain viruses, lack of vitamins or nutrients in the diet, or genetics. In many cases, a child with microcephaly will experience mental and/or physical disabilities. However, in some cases a child with microcephaly will develop normally with no restrictions on their abilities.

  A prenatal diagnosis may be made through ultrasound depending upon the level of prenatal care and the severity of the microcephaly. Most cases will be diagnosed at birth, though some cases may not be diagnosed until several months after the child was born. Besides a smaller head size, a child with microcephaly may have a high pitched cry, poor feeding or failure to thrive, and spastic movements of the arms and legs.

  There is no treatment for a child with microcephaly that will increase the child's head size. Treatments are based upon the individual child's needs with regard to his/her mental and physical disabilities.



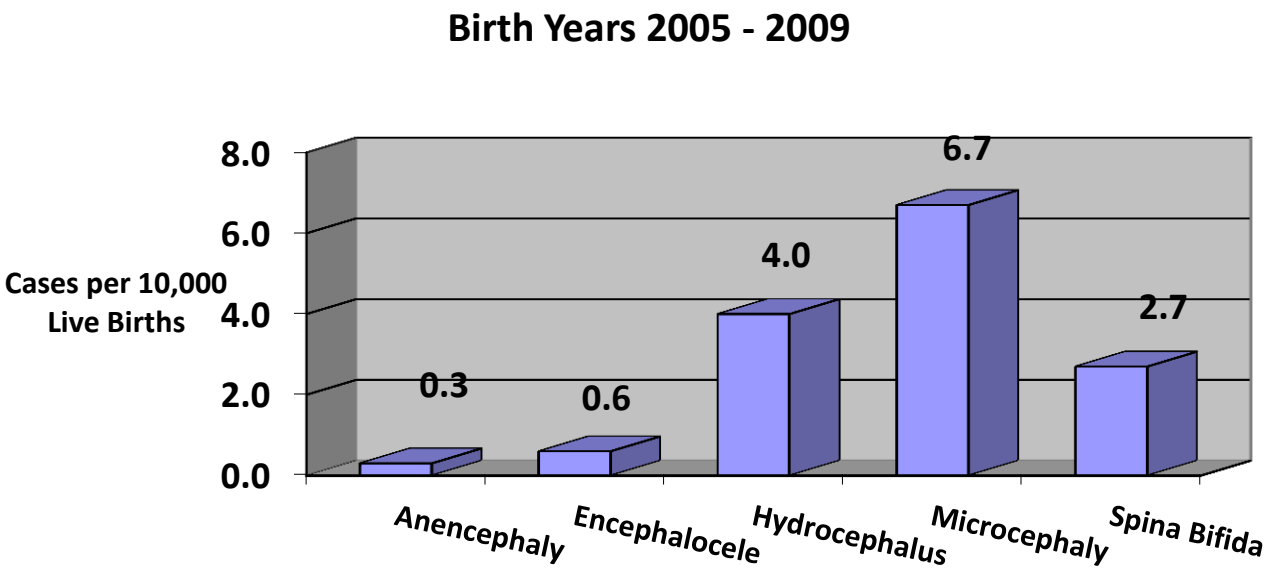
This image was provided by Boston's Children Hospital

New Jersey Data:

The Special Child Health Services (SCHS) Registry is a confidential record of infants and children who have birth defects and special health care needs or who are at-risk for developing such needs. Infants and children with a birth defect diagnosed through five years of age are required to be reported to the Registry. A child with a mandated condition identified after 5 years of age through 21years of age may also be reported but is not mandated by law. While not mandated to be reported, infants and children through 21 years of age with any chronic medical condition, especially those associated with developmental delay, are included in the Registry.

Children may be registered by hospitals, physicians, dentists, audiologists, certified nurse midwives, advanced practice nurses, cytogenetic laboratories and directors of clinical laboratories. SCHS case management units and Early Intervention Service providers may also register children. Postmortem examinations resulting in the identification of a mandated condition need to be reported to the Registry.

The figure below shows the number of cases per 10,000 live births of each Central Nervous System/Neural Tube defect for children born from 2005 through 2009 and registered with the New Jersey SCHS Registry.



Resources/Additional Information:

General information related to birth defects:

http://www.cdc.gov/ncbddd/birthdefects/index.html

General information related to anencephaly, encephalocele, and spina bifida:

http://www.cdc.gov/NCBDDD/birthdefects/types.html

General information related to hydrocephalus:

http://www.ninds.nih.gov/disorders/hydrocephalus/detail_hydrocephalus.htm

General information related to microcephaly:

http://www.ninds.nih.gov/disorders/microcephaly/microcephaly.htm

New Jersey Department of Health information for children with special needs, (Special Child Health Services Registry and services/resources for families):

http://nj.gov/health/fhs/sch/index.shtml