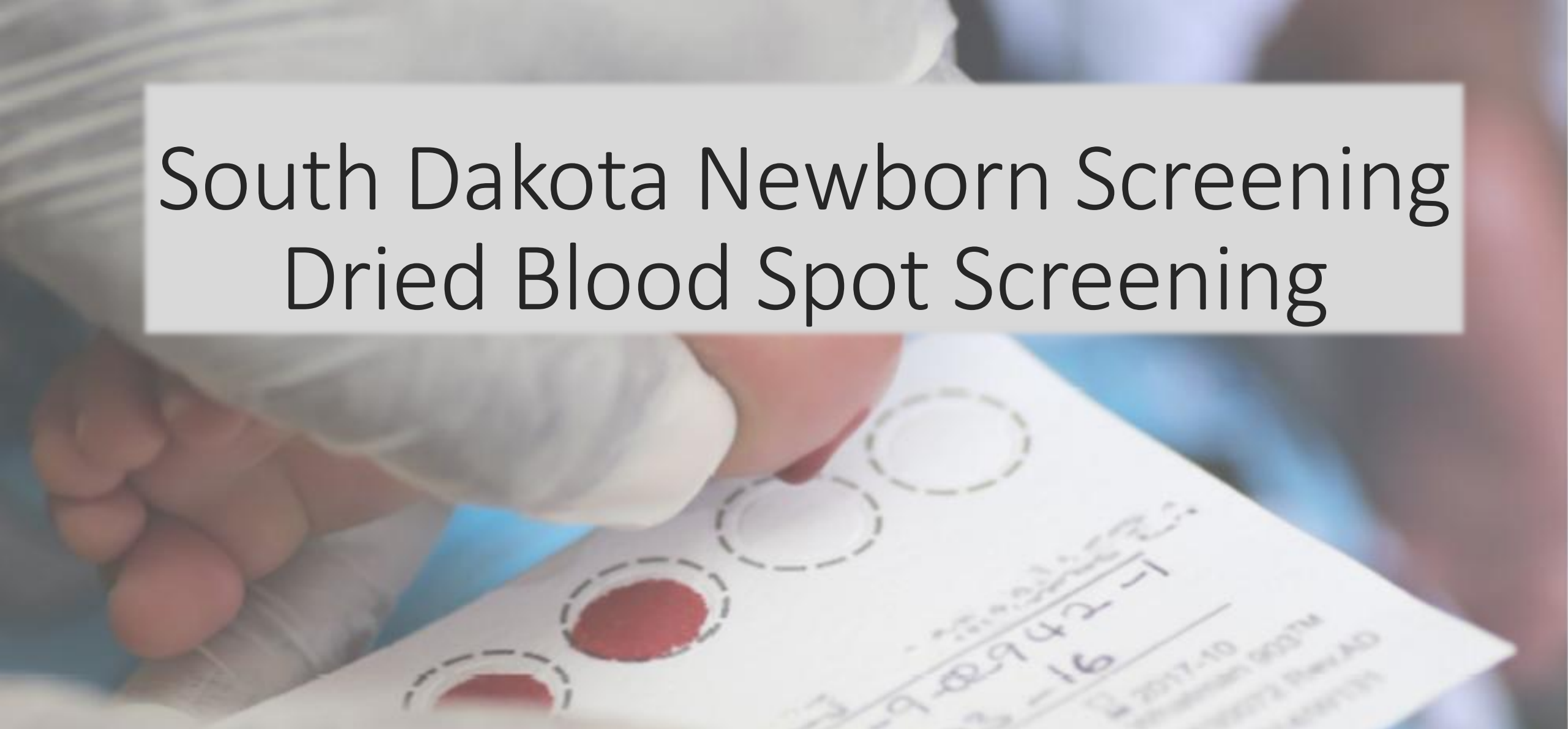
South Dakota Newborn Screening
Dried Blood Spot Screening



SOUTH DAKOTA DEPARTMENT OF HEALTH

THE UNIVERSITY OF IOWA | State Hygienic Laboratory

University of Iowa Stead Family Children's Hospital

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What is Newborn Screening?

Newborn screening is a way to identify babies who may have serious medical conditions. These conditions are often treatable but may not be visible at birth. Early treatment of these conditions can prevent against more serious illness, disability or death.

Newborn screening tests include:

- ❑Dried Blood Spot Screening (Genetic or Congenital Disorders)
- ❑Hearing Screening
- ❑Pulse Oximetry Screening (Critical Congenital Heart Disease)

Newborn screening is a collaborative effort!

Timely screening of South Dakota's newborns is only possible due to the collaborative effort from all the following:

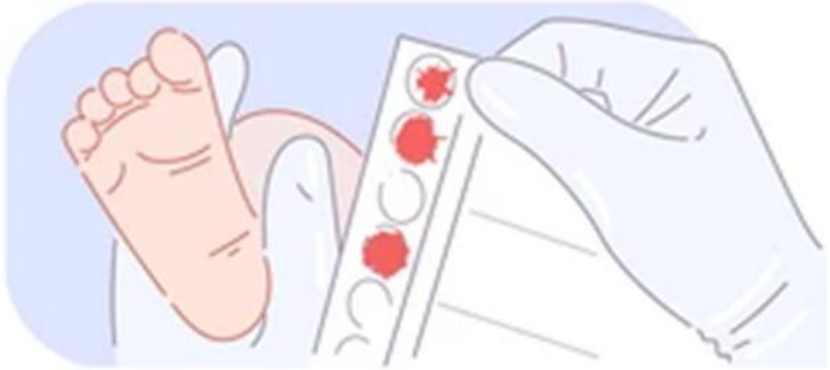
- State Hygienic Laboratory
- Iowa Department of Health
- South Dakota Department of Health
- All submitting South Dokata hospitals, clinics and midwives
- Health care workers
- Parents
- Courier drivers
- Follow up teams
- Medical geneticist

The South Dakota in collaboration with Iowa Newborn Screening Program (INSP) has the distinction of being known as one of the top timeliest newborn screening program in the United States!

Dried blood spot screening

Each year the South Dakota Newborn Screening Program blood spot screening identifies numerous newborns with one of the disorders screened for by the program.

Many of the disorders we screen for are extremely rare (e.g., 1 in 48,000 live births for classic galactosemia), but without newborn screening many of these would remain undetected until it became too late for treatment to prevent, reduce, or reverse the health problems associated with these disorders.



Newborn screening.
Blood sample from heel.

The State of South Dakota screens for over 50 genetic or congenital disorders!

ORGANIC ACIDEMIAS

- (GA-1) Glutaric acidemia type I*
- (HMG) 3-Hydroxy 3-methylglutaric aciduria *
- (IVA) Isovaleric acidemia*
- (3-MCC) 3-Methylcrotonyl-CoA carboxylase*
- (Cbl-A,B) Methylmalonic acidemia (cobalamin disorders, vitamin B12 disorders)*
- (βKT) βeta-Ketothiolase*
- (MUT) Methylmalonic Acidemia (methylmalonyl-CoA mutase)*
- (PROP) Propionic acidemia*
- (MCD) Holocarboxylase synthase*

•AMINO ACIDEMIAS AND UREA CYCLE DISORDERS

(ASA) Argininosuccinic aciduria*

(CIT) Citrullinemia, type 1 or ASA Synthetase Deficiency*

(HCY) Homocystinuria (cystathionine beta synthetase)*

(MSUD) Maple Syrup Urine Disease*

(PKU) Classic Phenylketonuria*

(TYR-1) Tyrosinemia, type I*

FATTY ACID OXIDATION DISORDERS

- (CUD) Carnitine uptake defect (Carnitine transport defect)*
- (LCHAD) Long-chain L-3 hydroxyacyl-CoA dehydrogenase*
- (MCAD) Medium chain acyl-CoA dehydrogenase*
- (TFP) Trifunctional protein deficiency*
- (VLCAD) Very long-chain acyl-CoA dehydrogenase.

ENDOCRINE

- (CAH) Congenital adrenal hyperplasia *
- (CH) Primary Congenital hypothyroidism *

HEMOGLOBINOPATHIES

- (Hb SS) S,S Disease (Sickle Cell Anemia)*
- (Hb S/C) S,C Disease*
- (HB S/βTh) S, βeta-thalassemia*

OTHER

- (BIOT) Biotinidase deficiency *
- (CF) Cystic Fibrosis *
- (GALT) Classic Galactosemia *
- (SCID) Severe Combined Immunodeficiency *
- (SMA) Spinal muscular atrophy
- (POMPE) GSD II Glycogen Storage Disease Type II

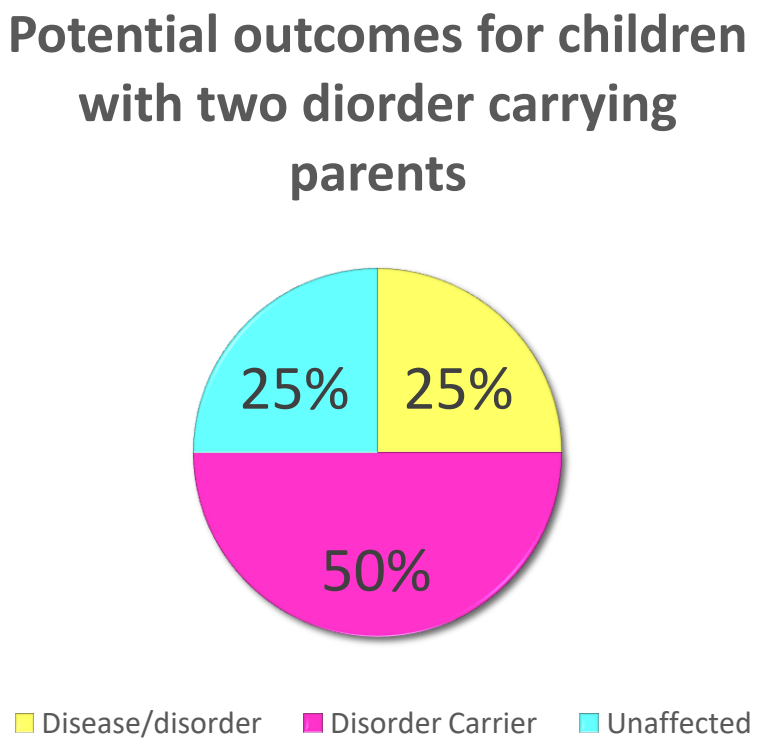
* Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) Recommended Uniform Screening Panel - Core Pane

Hidden Recessive disorders

Most disorders screened for are recessive in nature and go undetected through family bloodlines!

Recessive disorders require two bad genes to create a disorder. If only one gene is present the patient will be physically unaffected but remain a disorder carrier.

When two parent carriers have a child there is 25% chance the child will have inherit the disorder.



Time critical disorders

Some of the disorders we screen for are life threatening. We refer to these disorders as “time critical” disorders, because babies with one of these conditions may have only hours before the onset of a health crisis that can cause death or permanent disability.

What are the time critical disorders?

- Congenital Adrenal Hyperplasia
- Congenital Hypothyroidism
- Galactosemia
- Metabolic Disorders (most)

This is why collecting the screen as close to 24 hours is so important. With prompt collection and testing we can intervene and help minimize or prevent any serious complications for the baby!

How do we decide what to test for?

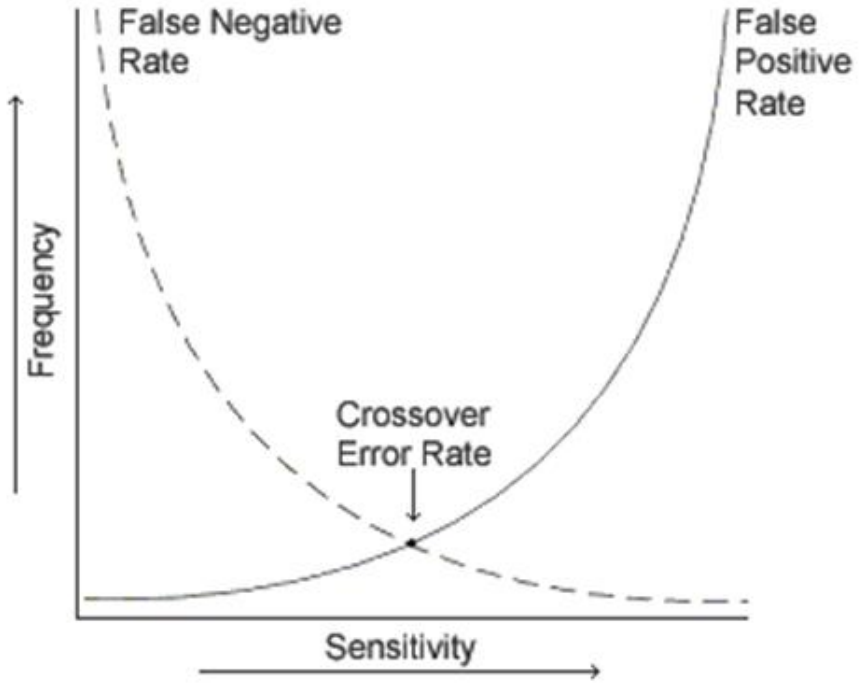
Decisions for including a disorder in the newborn blood spot screening program are based on the following criteria:

1. There is evidence of substantial public health benefit and acceptance by the public and the medical community.
2. Screening is feasible and cost effective.
3. Satisfactory test methods and laboratory facilities are available.
4. Resources exist to provide counseling and follow-up, and to address other consequences of screening.
5. The disorder is treatable and generally not easily identifiable without screening.
6. If untreated the disorder is likely to result in significant costs to the individual, the family and society.

Who decides which disorders are included on the South Dakota's newborn metabolic screening panel?

South Dakota Department of Health, Office of Child and Family Services is responsible for managing the newborn screening panel. The South Dakota Newborn Screening Advisory Committee advises which disorders to include, from the recommendations of Recommended Uniform Screening Panel (RUSP) that is provided by the Federal Advisory Committee on Heritable Disorders in Newborns and Children.

It’s a Screening Test Not a Diagnosis



Newborn screening is a screening test- not a diagnostic test!

A diagnosis is made from a combination of signs, symptoms, and test results from a doctor or primary care giver.

False positives and negatives are possible

- Continual quality improvements and refinements are made to decrease false results and lessen stress on infants and families.

Who is screened? And when?

By law **every** infant in born in South Dakota is screened.

Blood is drawn for the screen prior to hospital discharge.

- Optimal time being when the infant is 24 hours + 1 minute old.
- If a hospital transfer or blood transfusion is necessary a screen should be drawn prior to transfer or transfusion, even if < 24 hours old. A second screen drawn at a later time will be necessary in these situations.

Midwives also collect and submit newborn screenings.

South Dakota's Newborn Screening Coordinator, follow up nurse along with Iowa's follow up team work together that no baby is without a valid useable newborn screen.

Parent Education

All parents should be informed on the importance and benefits of screening prior to collection by providing the screening brochure.

- Available at here: Parental newborn screening brochure

Stress the importance of having correct contact information for the family and the primary care giver, including cell phone numbers.

- It could save a life!

Encourage parents to follow up with the baby's doctor at the first checkup on the results of the newborn screening.

Parent Education

Share the specific abnormal/positive screening result and associated condition(s) with the family.

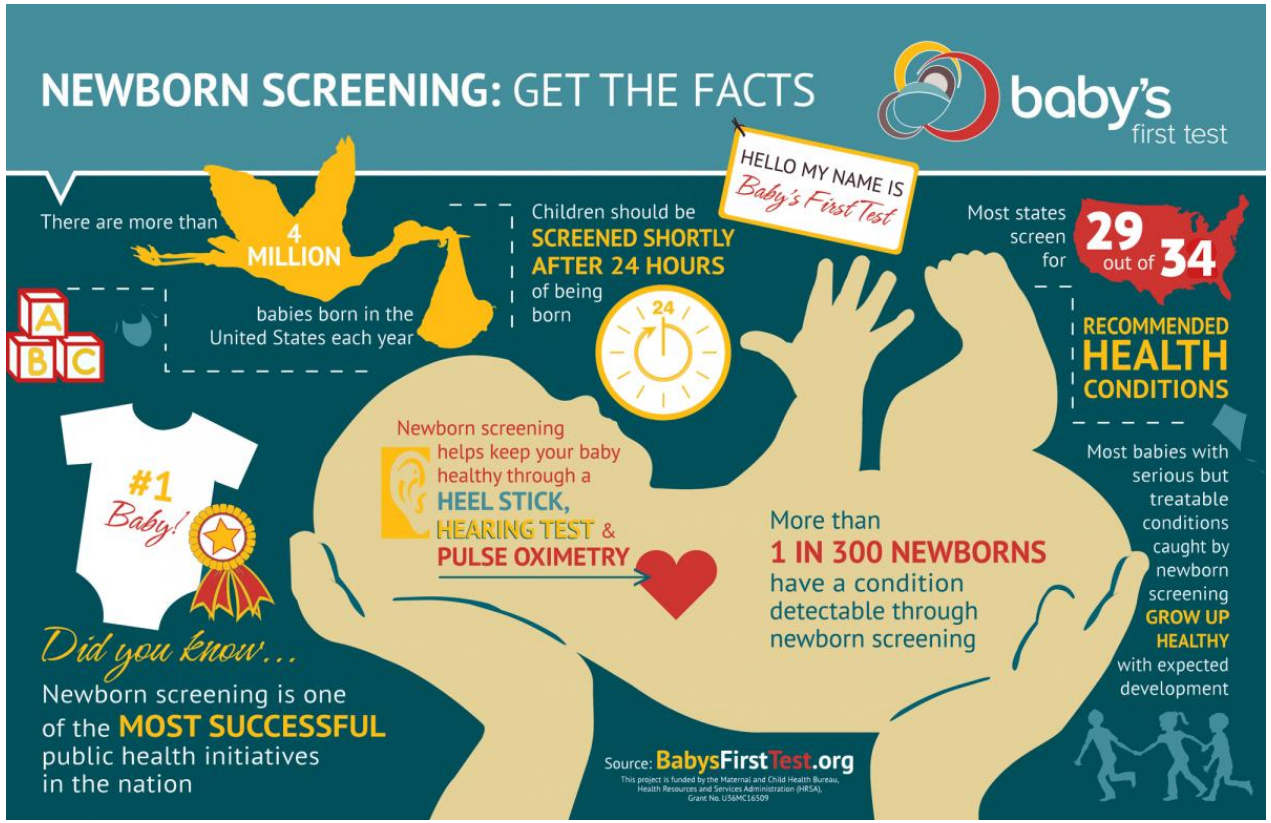
Comprehension: Assess the family's understanding of newborn screening.

Reiterate what screening is and is not.

Engage with the family and provide information at their desired level and pace.

Explore the family's emotions

Next steps: a shared plan and provide resources.



For more information about the Advisory Committee on Heritable Disorders in Newborns and Children, please visit https://www.hrsa.gov/advisory-committees/heritable-disorders

Other resources include BabysFirstTest.org for more informational guides and videos

What happens after testing is complete?

- All samples are assigned non-identifying barcode numbers upon arrival and are only tracked using that information.
- The newborn screening laboratory will destroy your baby's blood sample once it is no longer needed for testing.
- It will not be used for any purpose other than newborn screening.
- If you have questions about how your baby's blood sample is handled, contact the South Dakota Newborn Screening Program at (605) 773-3361.

If newborn blood spot screening is not done for some reason in the first week of life, is it worthwhile to still screen the baby later?

Yes!

While some disorders may begin to be expressed and some damage may have already occurred, treatment begun at any time will always be beneficial to the infant. Additionally, the family should be made aware of the infant's metabolic disorder, its genetic implications, and given appropriate counseling. Ideally, all babies should be screened in the first week of life, but screening a baby later is better than never screening at all.

Can newborn screening be done if a baby is born at home?

Yes!

Parents should arrange with their doctor, nurse, hospital, or midwife to have a newborn screening specimen collected. The specimens should be collected between 24 hours and 5 days of age.

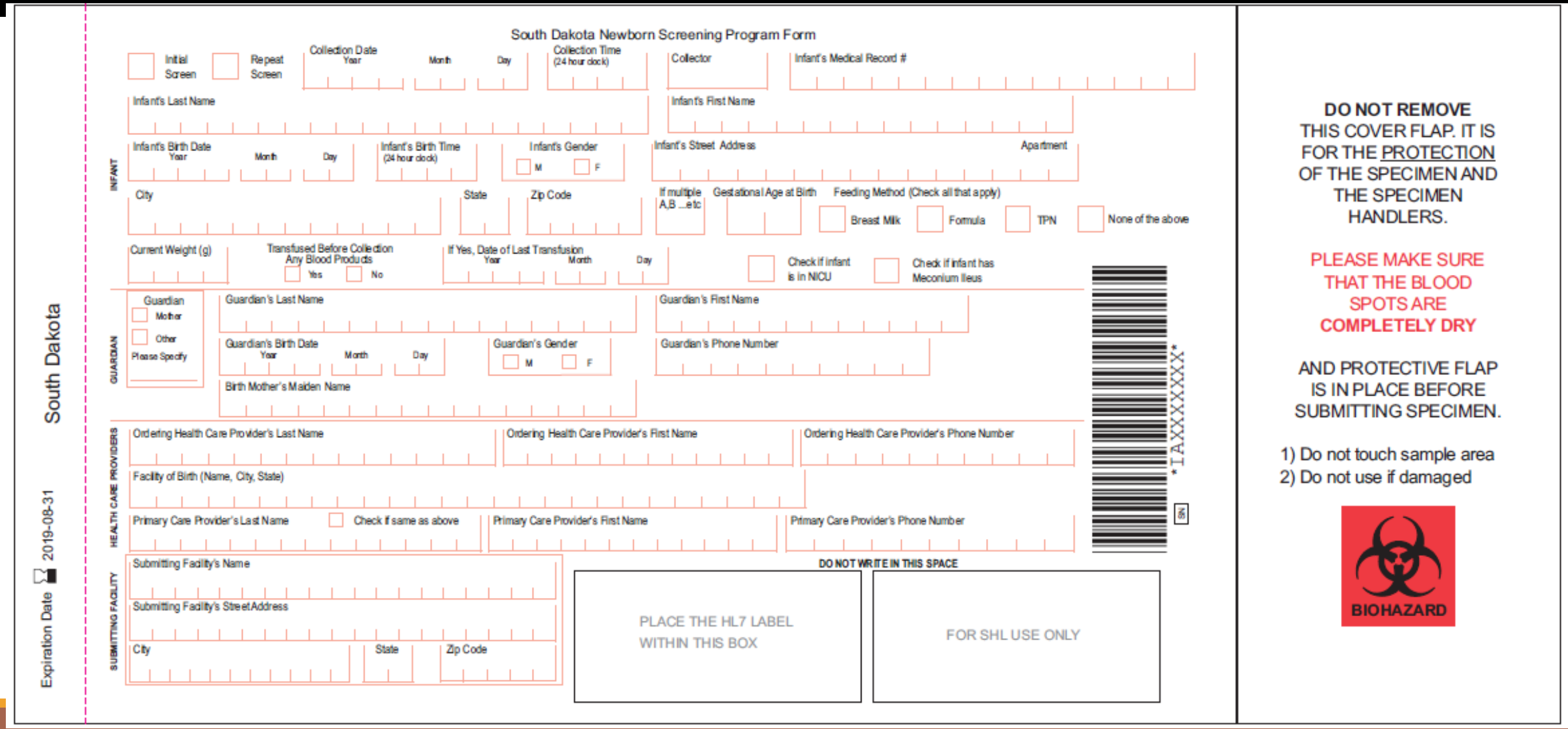
Properly completing a newborn screening form saves lives!

Forms provide vital information about the infant to help determine severity of potential problems.

- Weight
- Age
- Feeding method

Accurate contact information for the provider and parents, including a cell phone number, can help our follow up team quickly relay time sensitive information.

HOW TO COMPLETE SOUTH DAKOTA NEWBORN SCREENING CARDS



Completing Screening Card- General Information

It is extremely important to fill out the screening card accurately and completely. *Inaccurate or missing information may adversely affect screening results and/or the ability to quickly contact the infant's care provider in the event of an abnormal screening result.* ***Any delay may put the child's health at risk.***

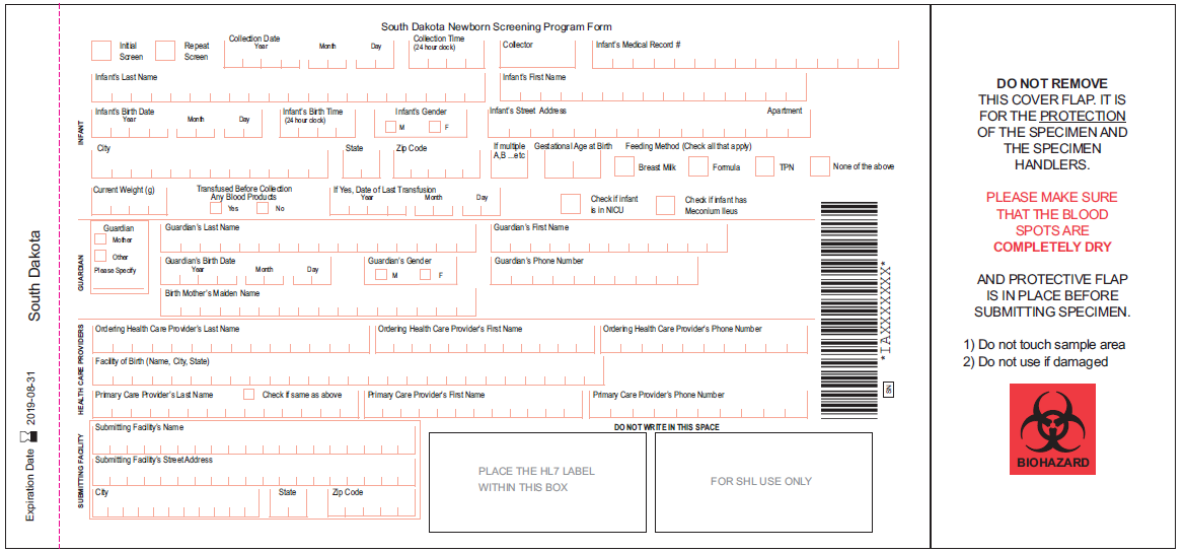
The specimen submitter is legally responsible for the accuracy and completeness of the information on the newborn screening card.

Remember:

Write firmly in blue or black ink to ensure that all information is transferred between carbon copies.

Remove the second ply for the facility's records.

For Questions, please call the State Hygienic Laboratory at 515-725-1630
Hours: Monday- Friday 8:00 a.m. to 4:30 p.m.



Completing Screening Card- Sample Information

Do not place stickers/labels or write in the lower right-hand side of the card in the area that says "FOR SHL USE ONLY"

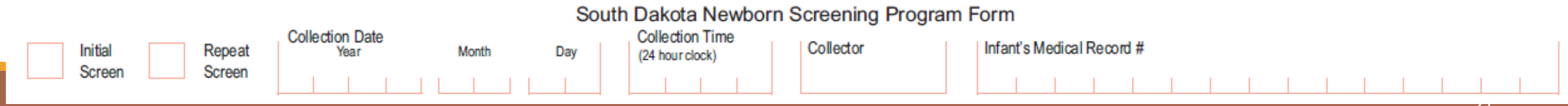
Place the HL7 (Health Level 7) label in the designated box if your facility electronically orders newborn screening tests. Leave this box empty if your facility does not electronically order newborn screening tests.
The box "**For SHL Use Only**" is used by the newborn screening lab. Do not write or apply stickers/ labels in this area.

Initial Screen vs. Repeat Screen:
Check the appropriate box: "Initial" or "Repeat."
Initial screen is the first submission.
Repeat screen(s) are any subsequent submission(s) received after the initial screen, even if the resubmission is due to poor quality/specimen rejection, prior early collection samples, etc.

Collection Date:
Use an eight-digit format (yyyy/mm/dd) for the newborn's date of collection. For example, a sample collected on March 9, 2015, would be recorded as 2015 03 09.

Collection Time:
Always use 24-hour clock (HH:MM) when entering the time of collection.
Validity of test results are specific to the exact age (in hours) of the infant, so an accurate time of collection is crucial.

Collector:
Use unique identifier (initials, last name, employee ID number, etc.) for the person collecting the sample. Each facility can determine its own unique identifier for internal use.



Completing Screening Card- Infant Information

Infant Medical Record Number:
Write the infant's medical record number.
Do not record the mother's medical record number.

Infant's Last Name:
Write the infant's last name.
It is important to list the infant's last name regardless of whether the guardian(s) has chosen a first name.
Do not assume that the infant's last name is the same as the mother's last name. Record the last name the infant will go by at discharge.
Providing an incorrect name could potentially cause a delay in reporting abnormal results and impact the health of the infant.

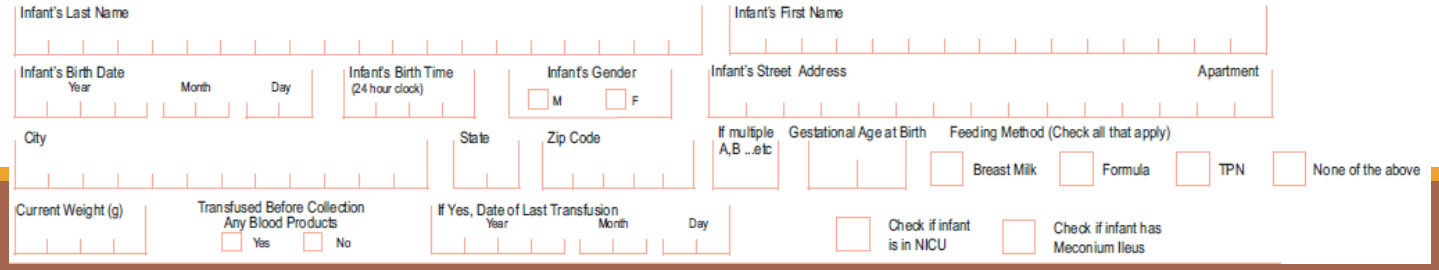
Infant's First Name:
Record infant's first name, if known.
If the guardian(s) have not yet chosen a first name, leave this field blank.
Providing an incorrect name could potentially cause a delay in reporting abnormal results and impact the health of the infant.

Infant's Birth Date:
Use an eight-digit format (yyyy/mm/dd) for the infant's date of birth. For example, an infant born on March 9, 2015, would be recorded as 2015 03 09.

Infant's Birth Time:
Always use 24-hour clock (HH:MM) when entering the time of birth. For example, the time for a baby born at 4:15 p.m. would be recorded as 16:15.
Validity of test results are specific to the exact age (in hours) of the infant, so an accurate birth time is crucial.

Infant's Gender:
Mark "M" for male or "F" for female. If unknown or ambiguous genitalia, write "Unknown" in the Infant's Gender box.
This helps with the identification of the baby.

Infant's Street Address:
Record where the infant will reside.
Use complete address, city, state and zip code.
In the event of an adoption or other guardianship, record the address where the infant will reside.
Accurate contact information is crucial for contacting the guardian in the event of an abnormal result or a need for retesting.

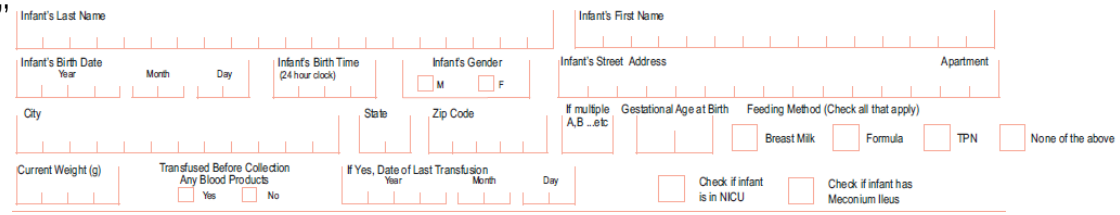


Completing Screening Card- Infant Information

If Multiple A, B…etc.:
If the infant is one of a set of multiple births (twins, triplets, etc.) record the birth order of the infant. For example, if the infant was the first born in a set of triplets, write "A", in the box. For the third born infant, write "C" in the box.
If single birth, leave blank, put a line through the field or cross it out.
This field is not in reference to the birth order of ALL pregnancies but the birth order of this one pregnancy.

Gestational Age at Birth:
Record the infant's week of gestation at time of birth. Record in completed weeks only, no rounding up.
Accurate gestational age is critical for analyzing the results of newborn screening tests. This includes all collections - initial and repeat screens.
If unknown, write "Unknown."

Feeding Method:
Check all types of feeding that apply within the last 24 hours. For example, if the infant has received both Total Parenteral Nutrition (TPN) and breast milk in the last 24 hours, check both boxes.
Breast milk includes milk sourced from biological mother or donor milk.
TPN includes, but is not limited to, Neonatal Venous Nutrition (NVN), Peripheral Parenteral Nutrition (PVN), Hyperalimentation (Hyperal), Starter TPN, any supplementation that includes amino acids, and/or any additional TPN products not mentioned.
If infant is receiving fluids only and/or no other feeding method listed, check "None of the above."
Formulas include all special formulas and additives (e.g. Human Milk Fortifier, Beneprotein, etc.).



Current Weight (g):
Record the infant's weight in grams at time of specimen collection.
Do not leave blank. It is important to correctly record the infant's weight for accurate test results.

Transfusion (Any Blood Products):
This field **MUST** be marked "Yes" or "No" because transfusion status affects results. Missing information could lead to delays. If the infant was given any blood product BEFORE newborn screen collection, check "Yes." If the infant was NOT transfused or transfused after collection check "No."
Write the date of the most recent transfusion. If infant has received multiple transfusions, you only need to record the most recent date of transfusion.
Use an eight-digit format (yyyy/mm/dd) for the most recent transfusion date. For example, infant was last transfused on March 9, 2015, recorded as 2015 03 09.
Transfusion includes ALL blood products including, but not limited to, red blood cells, plasma, immunoglobulins and platelets.
If baby received a transfusion before delivery (intrauterine), mark "Yes" and record the date of the most recent transfusion.

Check if infant is in NICU:
Check the box if the patient is in Neonatal Intensive Care/ Pediatric Intensive Care Unit (NICU/PICU) or another high-acuity level care unit at time of collection.
If infant is not in NICU/PICU, leave blank.

Check if infant has Meconium Ileus:
Meconium ileus is known to interfere with the screening for cystic fibrosis. If meconium ileus is suspected, the screening algorithm for cystic fibrosis will change.
Check the box ONLY IF the infant has or is suspected of having meconium ileus.
If no meconium ileus is suspected, leave blank.

Completing Screening Card- Guardian Information

Guardian is considered the person with the legal authority to care for the infant. In most cases, this is the birth mother but can include other legal guardian relationships if birth mother is not the legal guardian.

Guardian Box:

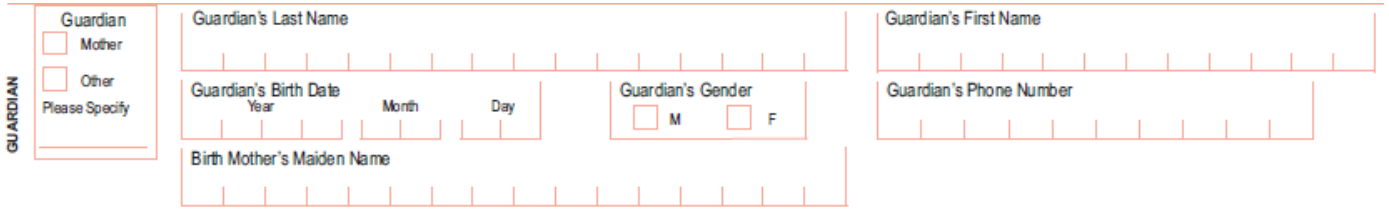
Mother is in reference to biological mother. If biological mother is legal guardian, check "Mother."
If legal guardian is any other relation other than biological mother, mark "Other."
If the infant is in the custody of the biological mother, provide the mother's information as the guardian. If the mother is not a legal guardian, provide legal guardian information.
If other, record relation under "Please Specify." Examples of "other" include adoptive parent, human services, adoption agency, grandparent, etc.

Guardian Last Name and First Name:

Record the guardian's last name followed by first name.
In the event of an adoption, record the name of the legal guardian (adoptive parent, adoption agency, social worker, etc.).
If infant was born via surrogacy, provide the name of the legal guardian who will take care of infant post-delivery.
Accurate identifying information is crucial for contacting the guardian in the event of an abnormal result or a need for retesting.
In the event that the infant will be held in protective services, record the name of the infant's social worker or legal guardian.

Guardian's Birth Date:

Use an eight-digit format (yyyy/mm/dd) for the guardian's date of birth. For example, a guardian born on March 9, 2015, would be recorded as 2015 03 09.
In the event of an adoption, write the date of birth of the adoptive parent.



Guardian's Gender:

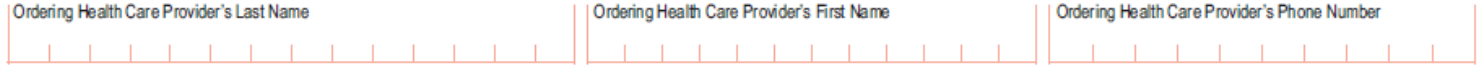
Check "M" for Male or "F" for Female.

Guardian's Phone Number:

Record the guardian's phone number (including area code) at which he/she most easily can be reached in case of emergency.
In the event that infant is not in the custody of birth parents, provide contact information for the legal guardian.
In the event of an adoption, record the phone number of the case worker here.
In the event that the infant will be held in protective services, record the phone number of the legal guardian or social worker. *Make sure the number provided will be answered on weekends and holidays in case of emergencies*.
Accurate contact information for a guardian is important to ensure that the infant can receive follow-up testing and/or care in the event of an abnormal result. Make sure the guardian's number provided will be answered on weekends and holidays in case of emergencies.

Completing Screening Card- Health Care Provider Information

Ordering Health Care Provider's Name:
Record the name of the health care provider ordering the infant's newborn screen, using last name followed by first name.

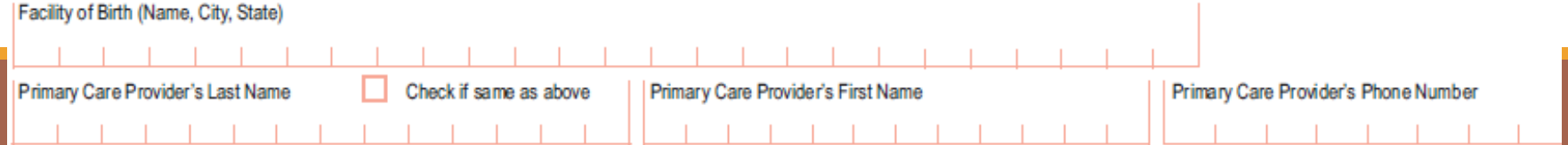


Ordering Health Care Provider's Phone Number:
Provide the phone number (including area code) for the health care provider ordering the infant's newborn screen.
This information may be used to contact the provider with abnormal test results and follow-up information.

Ordering Health Care Provider's National Provider Identifier Number (NPI)
Provide the Ordering Health Care Provider's National Provider Identifier number to help correctly identify the correct provider.
This information may be known by lab staff or billing staff at your facility.

Primary Care Provider Responsible for Infant Follow-Up After Discharge:
If the Primary Care Provider is the same as the Ordering Health Care Provider, check the box "Check if same as above."
If the Primary Care Provider is different from the Ordering Health care provider, record the name of the Primary Care Provider, using last name followed by first name.
If the provider is not known at the time of specimen collection, be sure to write down the name of the clinic where the guardian(s) plan to take the newborn for his or her first well child check.
Do not write the name of the provider who completed rounds on the newborn in the hospital.
Correctly recording this information is critical. The Newborn Screening Program needs the name of the primary care provider to make sure follow-up of abnormal results is completed.

Primary Care Provider's Phone Number:
Provide the phone number (including area code) for the infant's primary care provider.
This information is used to contact the provider with abnormal test results and follow-up information.



Completing Screening Card- Submitter Information

Apply the pre-printed labels supplied by the State Hygienic Laborato with the collection forms.

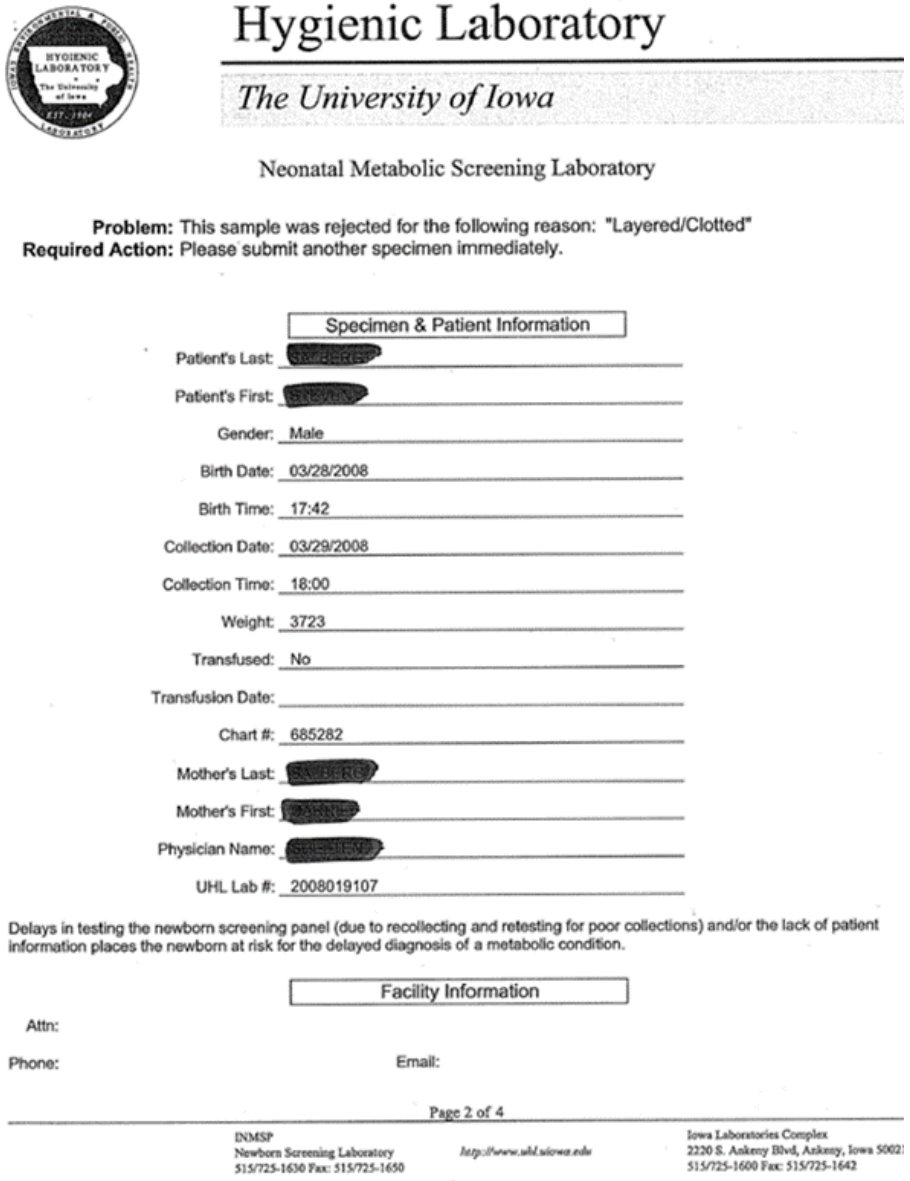
Verify that the label matches your facility name and address.
- Do not share forms or labels with other facilities as this can lead to results being sent to wrong organizations.
- The submitter information provided is used for result reporting purposes as well as billing. Provide accurate and complete information.

If no label is available**:**
Submitting Facility Name:
Record the name of the hospital, clinic or midwife who collected the specimen.

Submitting Facility's Complete Address:
Write the street address of the submitter (vital because many institutions have the same name and/or are part of a larger affiliation).
Write the city, state and zip code.

Missing Information request forms

Faxes are sent out daily to facilities requesting missing information or informing them of the need for recollection. Completion and immediate return is required for reports to be submitted back to facilities.



Hygienic Laboratory

The University of Iowa

Neonatal Metabolic Screening Laboratory

Problem: This sample was rejected for the following reason: "Layered/Clotted"
Required Action: Please submit another specimen immediately.

Specimen & Patient Information

Patient's Last: [redacted]
Patient's First: [redacted]
Gender: Male
Birth Date: 03/28/2008
Birth Time: 17:42
Collection Date: 03/29/2008
Collection Time: 18:00
Weight: 3723
Transfused: No
Transfusion Date:
Chart #: 685282
Mother's Last: [redacted]
Mother's First: [redacted]
Physician Name: [redacted]
UHL Lab #: 2008019107

Delays in testing the newborn screening panel (due to recollecting and retesting for poor collections) and/or the lack of patient information places the newborn at risk for the delayed diagnosis of a metabolic condition.

Facility Information

Attn:

Phone: Email:

Page 2 of 4

DMSP
Newborn Screening Laboratory
515/725-1630 Fax: 515/725-1650

http://www.uhl.uiowa.edu

Iowa Laboratories Complex
2220 S. Ankeny Blvd, Ankeny, Iowa 5002
515/725-1600 Fax: 515/725-1642

Factors affecting newborn screens

	Early Collection	Transfusion	Heat/ Humidity	TPN	Steroids/ Other Medication (ex: certain antibiotics)	Prematurity	Weight
CH	x				x		
CAH	x						x
BT		x	x		x		
GALT		x	x		x		
HB		x			x		
IRT		x			x		
TMS	x			x	x	x	
TREC						x	

Early collection before 24 hours of age could give false positives or negatives. Blood specimen should be collected between 24-48 hours. With exceptions being encouraged to draw before transfusion, transfer, or discharge.

Factors affecting newborn screens

	Early Collection	Transfusion	Heat/ Humidity	TPN	Steroids/ Other Medication (ex: certain antibiotics)	Prematurity	Weight
CH	x				x		
CAH	x						x
BT		x	x		x		
GALT		x	x		x		
HB		x			x		
IRT		x			x		
TMS	x			x	x	x	
TREC						x	

Red Blood cell transfusion interfere with the interpretation of some metabolic screening results. Whenever possible a screen should be drawn prior to transfusion. If this is not possible, a follow up screen will be requested 8 weeks from the last transfusion date.

Factors affecting newborn screens

	Early Collection	Transfusion	Heat/ Humidity	TPN	Steroids/ Other Medication (ex: certain antibiotics)	Prematurity	Weight
CH	x				x		
CAH	x						x
BT		x	x		x		
GALT		x	x		x		
HB		x			x		
IRT		x			x		
TMS	x			x	x	x	
TREC						x	

Because some of our lab assays directly measure enzyme activity, exposure to direct sunlight, heat, and/or high levels of humidity can cause these enzyme levels to degrade too rapidly to allow for accurate testing. A dried blood spot specimen should never be left to dry in a vehicle or other place where it can be exposed to extreme temperature and humidity variations.

Factors affecting newborn screens

	Early Collection	Transfusion	Heat/ Humidity	TPN	Steroids/ Other Medication (ex: certain antibiotics)	Prematurity	Weight
CH	x				x		
CAH	x						x
BT		x	x		x		
GALT		x	x		x		
HB		x			x		
IRT		x			x		
TMS	x			x	x	x	
TREC						x	

Infants on some types of TPN may show elevated levels of amino acids (e.g., phenylalanine). Indication of TPN status on the collection form is necessary for clarifying some test results..

Factors affecting newborn screens

	Early Collection	Transfusion	Heat/ Humidity	TPN	Steroids/ Other Medication (ex: certain antibiotics)	Prematurity	Weight
CH	x				x		
CAH	x						x
BT		x	x		x		
GALT		x	x		x		
HB		x			x		
IRT		x			x		
TMS	x			x	x	x	
TREC						x	

Steroids administered to the mother during pregnancy, or to the infant immediately after birth, can interfere with congenital adrenal hyperplasia test results. Contact the endocrine consultants regarding management for these situations, (319) 356-2838.

Factors affecting newborn screens

	Early Collection	Transfusion	Heat/ Humidity	TPN	Steroids/ Other Medication (ex: certain antibiotics)	Prematurity	Weight
CH	x				x		
CAH	x						x
BT		x	x		x		
GALT		x	x		x		
HB		x			x		
IRT		x			x		
TMS	x			x	x	x	
TREC						x	

SCID screening in Iowa is done through T-cell receptor excision circle (TREC) analysis. SCID is a lack of T cells which makes an infant extremely susceptible to infections. TRECs (T-cell receptor excision circles) are pieces of DNA produced in the thymus. Premature infants frequently have lower TREC levels compared to term infants. This finding can be secondary to several factors including time for T-cell maturation in the thymus, medications given prior to delivery which might reduce T-cell numbers, and/or dilutional factors related to sample collection from an indwelling catheter.

Factors affecting newborn screens

	Early Collection	Transfusion	Heat/ Humidity	TPN	Steroids/ Other Medication (ex: certain antibiotics)	Prematurity	Weight
CH	x				x		
CAH	x						x
BT		x	x		x		
GALT		x	x		x		
HB		x			x		
IRT		x			x		
TMS	x			x	x	x	
TREC						x	

Transient elevations of 17-OHP (the analyte for the congenital adrenal hyperplasia - CAH screen) may occur in pre-term and low birth weight babies. Because of this, four weight related 17-OHP ranges are in place to minimize the number of false positive results. Without a weight indicated on the collection form, CAH results cannot be reported. If the weight is inadvertently omitted you can fax the weight at time of collection to the lab and we will reissue the report based on the new information. The fax number is 515/243-3071

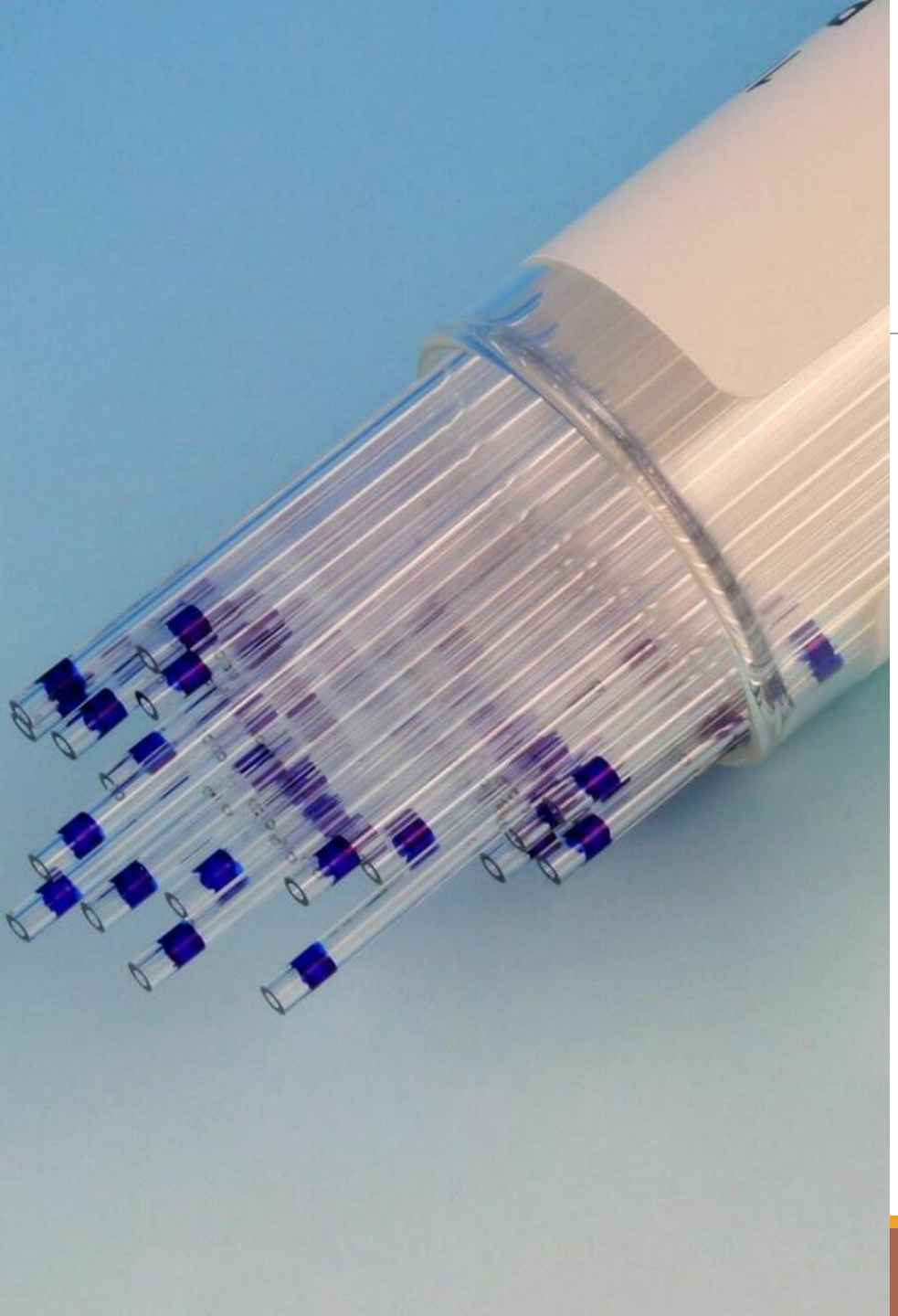
Unacceptable Collection Sites

- Arch of the foot
- Infant's fingers and toes
- Earlobes
- Previously punctured sites
  - If a recollection is needed a new puncture must be used.
- Intravenous lines contaminated with interfering substances

Alternative blood collection techniques not recommended

Venous	Syringe	Umbilical catheter	Umbilical cord blood
• More invasive than a heel stick • Veins may be required to administrate fluids or medications	• Causes clotting, settling, and lysing of cells	• Causes clotting, settling, and lysing of cells	• Maternal blood contamination • Must collect within first 5 minutes of birth, breaks the 24 hour age rule

*All listed methods may be appropriate under certain circumstances (e.g. NICU).



Alternative blood collection techniques not preferred

Capillary tubes are frequently used but we do not prefer their use.

- High risk of collection error
- Scratching or denting of filter paper makes the spots unusable.
- Do not “color in” the filter paper circles
- EDTA and heparin interfere with test results

If used a new anticoagulant tube must be used for each printed circle and the capillary tube should not touch the paper.

Preferred Method is a Heel Stick!

Heel Stick/Direct application method is the preferred method of collection and standard in newborn screening!

Summarized from the Clinical and Laboratory Standards Institute (CLSI) guidelines. Please refer to these guidelines for further information.

CLINICAL AND LABORATORY STANDARDS INSTITUTE®

How to collect an acceptable Blood Spot Specimen

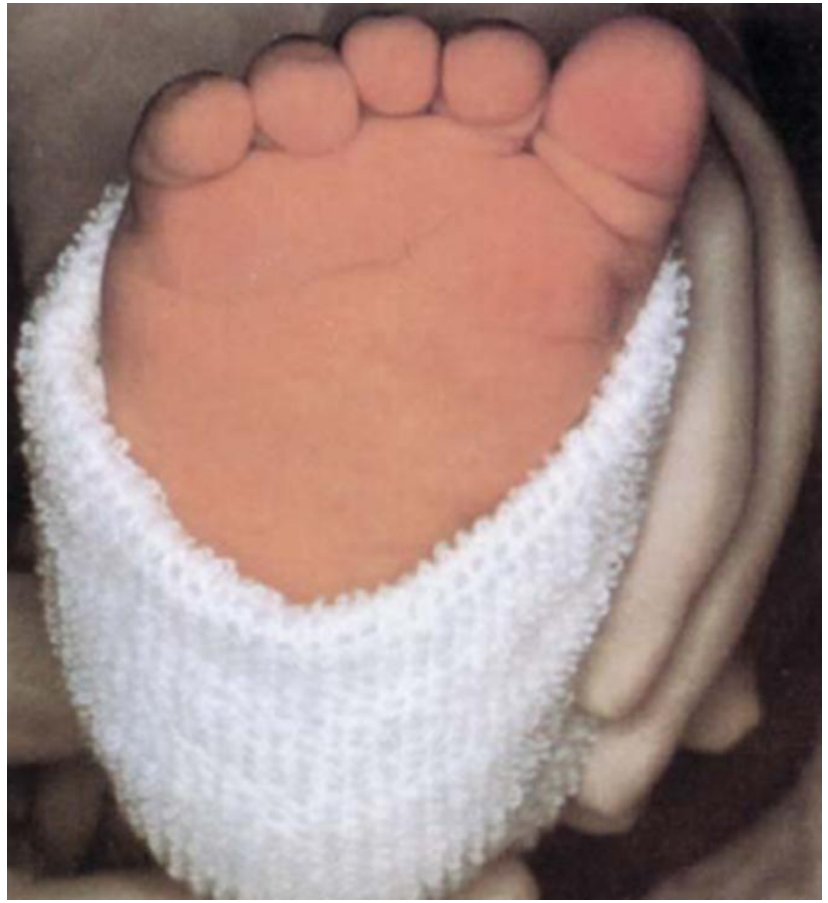
Preliminary and Precautionary Steps

- Check expiration date for the collection form
- Confirm infant's identity and ensure that all areas of collection form are completely and accurately filled in
- Wash hands vigorously
- Wear powder free gloves and change gloves between newborns

How to collect an acceptable Blood Spot Specimen

Site Preparation

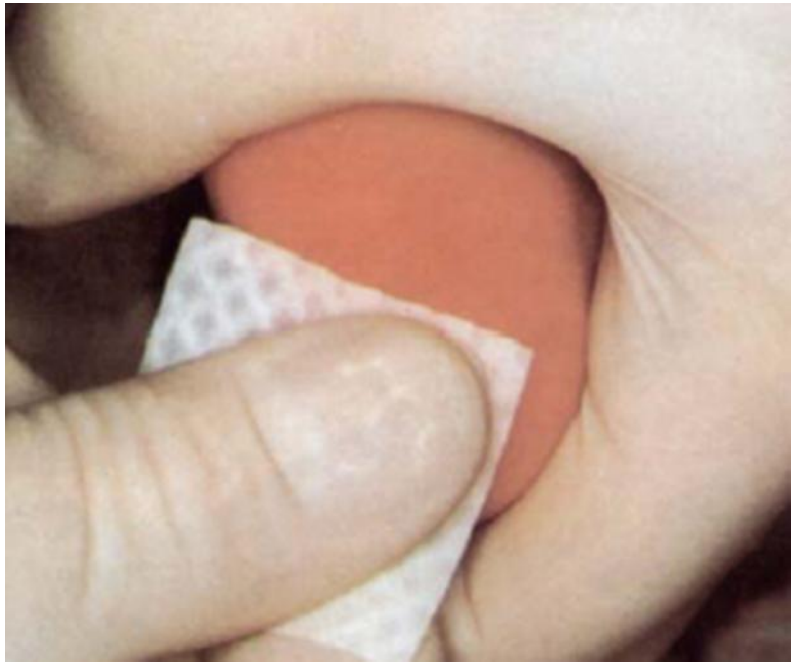
Warm the newborn's heel around the intended area of the skin puncture site or cover the puncture site 3-5 minutes with a moist towel heated no more than 42° C



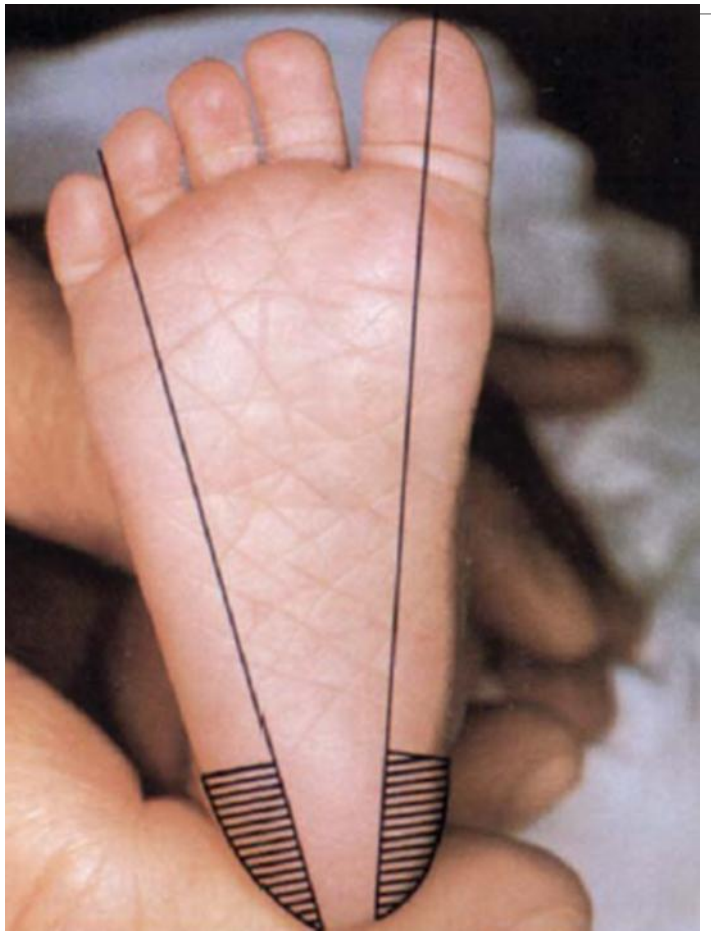
How to collect an acceptable Blood Spot Specimen

Site Preparation

- Position the leg lower than the infants heart to increase venous pressure
- Wipe heel clean with 70% isopropanol or chlorhexidine gluconate (<2%)
- Allow heel to completely air dry.



How to collect an acceptable Blood Spot Specimen



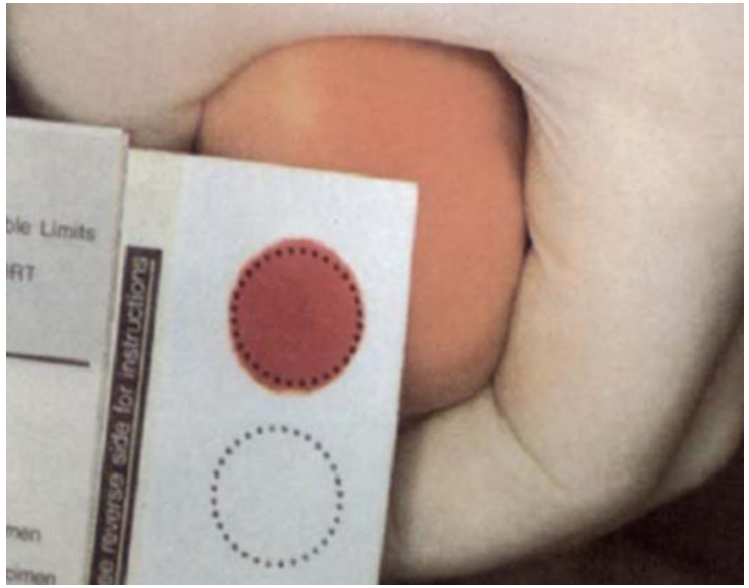
Puncture

- Use a sterile retractable puncture device (such as a lancet)
- Position the puncture device over the medial or lateral planter surface of the heel (within the shaded areas)
- Puncture the heel to a depth of:
  - <0.85mm for preterm newborns
  - <2.0 mm for term newborns

How to collect an acceptable Blood Spot Specimen

Direct Application & Collection

- Wipe away first blood droplet with sterile gauze pad
- Gently squeeze heel to allow a large round blood drop to form
- Apply gentle pressure to the heel with your thumb and ease up pressure as a drop of blood begin to form. Do not excessively “milk” the puncture site.



How to collect an acceptable Blood Spot Specimen

Direct Application & Collection

- Touch the filter paper to the blood drop and fill each preprinted circle with a single application of blood on one side of the filter paper. Be sure to not touch the filter paper to the heel.
- Examine both sides of the filter paper for proper saturation.
- Elevate the newborn's foot above the heart and hold gentle pressure to the puncture site with sterile gauze until bleeding has stopped. Do not use adhesive bandages.
- Dispose of all used items in a biohazard container.

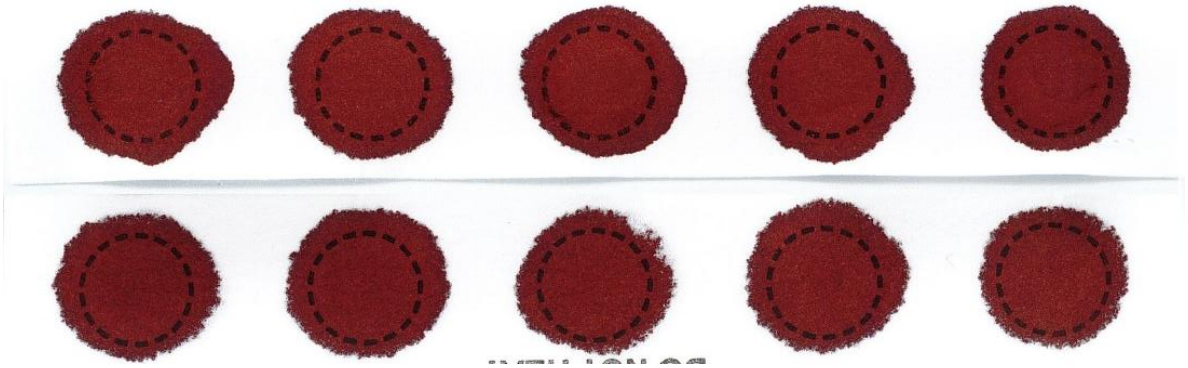
How to collect an acceptable Blood Spot Specimen

<u>Preparation for Transport</u>

- On a level, horizontal, dry location (like a drying rack) allow the blood spots to thoroughly air dry for at least three hours.
- After completely dry place the specimen in an approved container for transport after reviewing for from completion and blood spot quality.

Acceptability Criteria

- ❑Single drop applied to one side of the filter paper for each circle.
- ❑At least ½ inch or 9mm in diameter
- ❑Spots can be outside of the outlined areas on the filter paper but must not touch other spots.
- ❑Minimum of **three** quality spots per sample are required to perform all testing.



Are all 5 blood spots needed?

- If a result flags as a borderline or presumptive positive testing will be repeated in duplicate for the flagging test to confirm results before releasing.
- Resting may be required due to instrument malfunctions or other errors.
- Lot qualifications require comparison to previously run samples.
- Disorders are continually being added to the newborn screening panel.

Rejection/Poor Quality Samples

Samples may be rejected for several reasons including:

- Layering/ clotting
- Didn’t Soak through
- Quantity not Sufficient (QNS)
- Serum Separation
- Contamination/ diluted
- Scratched/ Abraded
- No blood applied
- > 14 days past collection
- Expired collection forms

All received specimen are tested (except cards with no blood) for all disorders

- If the child is considered at high risk with presumptive positive results, results will be relayed to follow up teams and care givers.
- If no test indicates high risk then the results will go out as a poor quality and request a retest.

Poor Quality Specimen adverse effect

Any sample that has been rejected due to failure to meet quality criteria MUST be recollected which can:

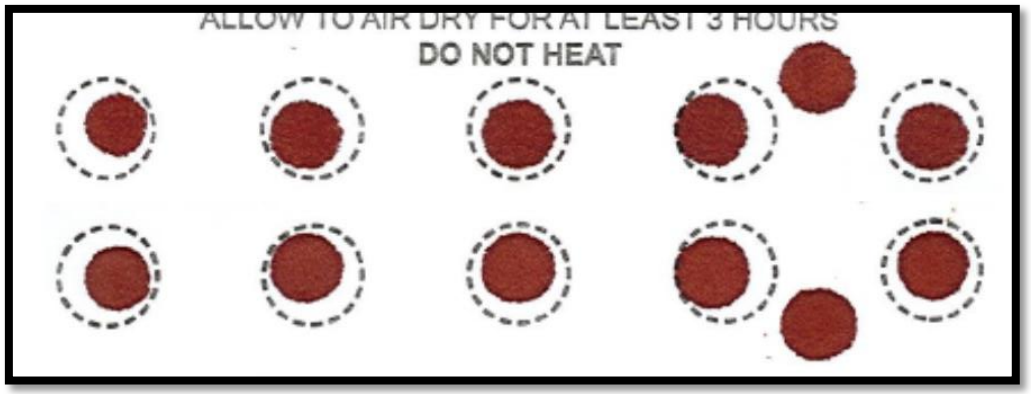
- Add trauma to the infant
- False positive results can cause anxiety for the parents
- Creates additional work for collecting facility and testing laboratory
- Delay potential diagnoses and treatment for the infant

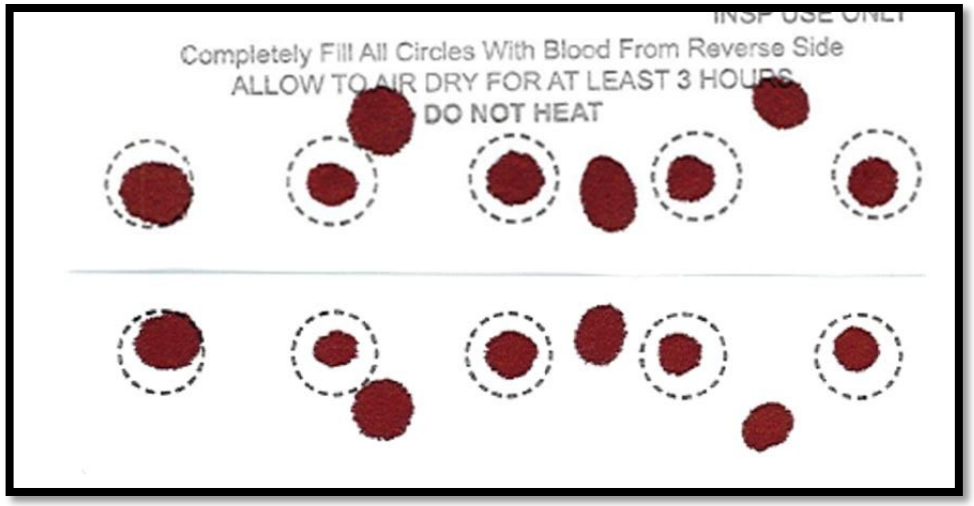
Quantity Not Sufficient (QNS)

Spots smaller than 9 mm or ½ inch in diameter can not be used to produce reliable results.

False negatives and positives are common from using small spots.

Lack of enough initial sample to complete all testing





Both examples have good spots but the size of the spots are too small to meet quality standards.

Quantity Not Sufficient (QNS) Prevention

- ❑Warm baby's heel prior to collection via a warm cloth or heel warmer for 3-5 minutes to encourage blood flow.
- ❑Position the baby with the heel lower than the heart for collection.
- ❑Patience! Wait for a large blood droplet to form on the heel before applying to the filter paper.
- ❑Use the pre-printed circles as a guide. To meet size requirement the spot may only be slightly smaller than the printed line.
- ❑Add large droplets to the filter paper in an area not being utilized, if this isn't possible collect a new specimen.

Why spot size matters

- George, Roanna S., and Stuart J. Moat. "Effect of dried blood spot quality on newborn screening analyte concentrations and recommendations for minimum acceptance criteria for sample analysis." Clinical chemistry 62.3 (2016): 466-475.
- Published December 8, 2015

- "Smaller bloodspots produced significantly lower results (15%–24% for 10-µL vs 50-µL sample size) for all analytes at all concentrations measured"
- "We recommend that samples of <20 µL (<8 mm diameter) be rejected, secondary to the observed negative bias."

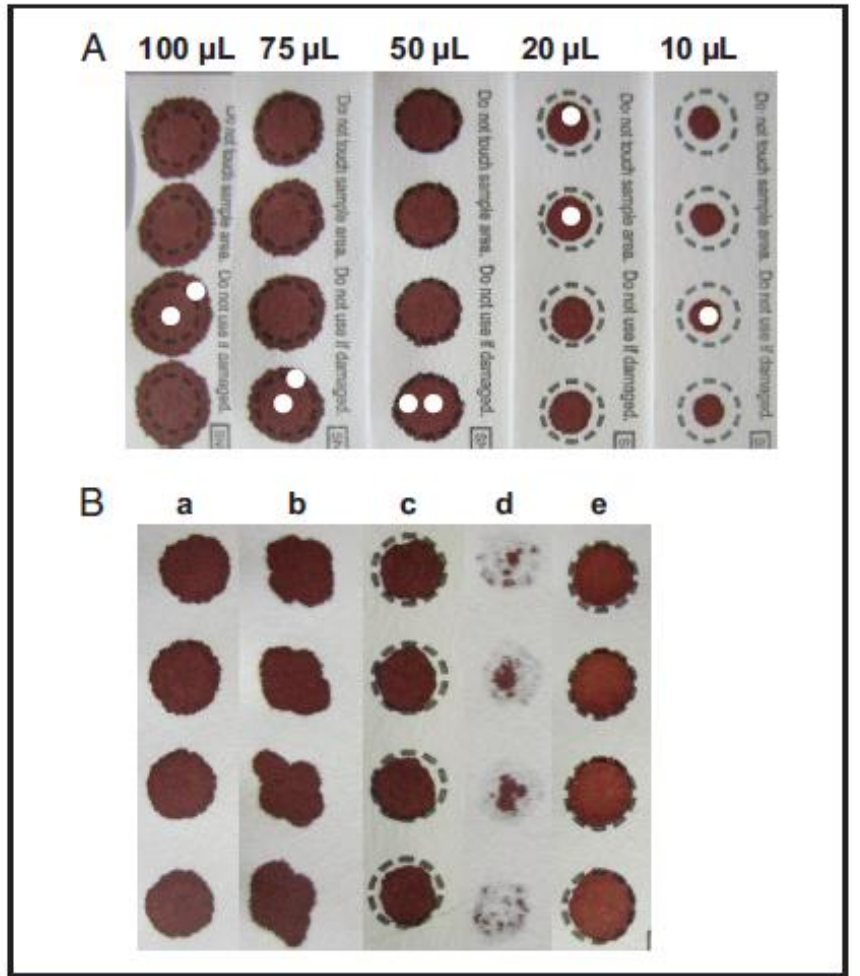
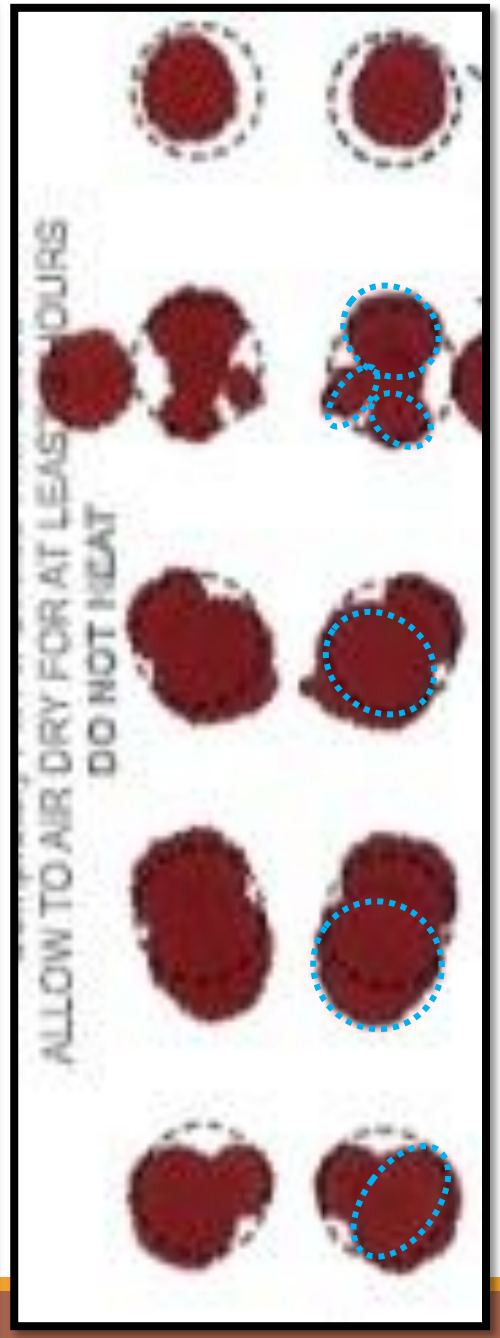
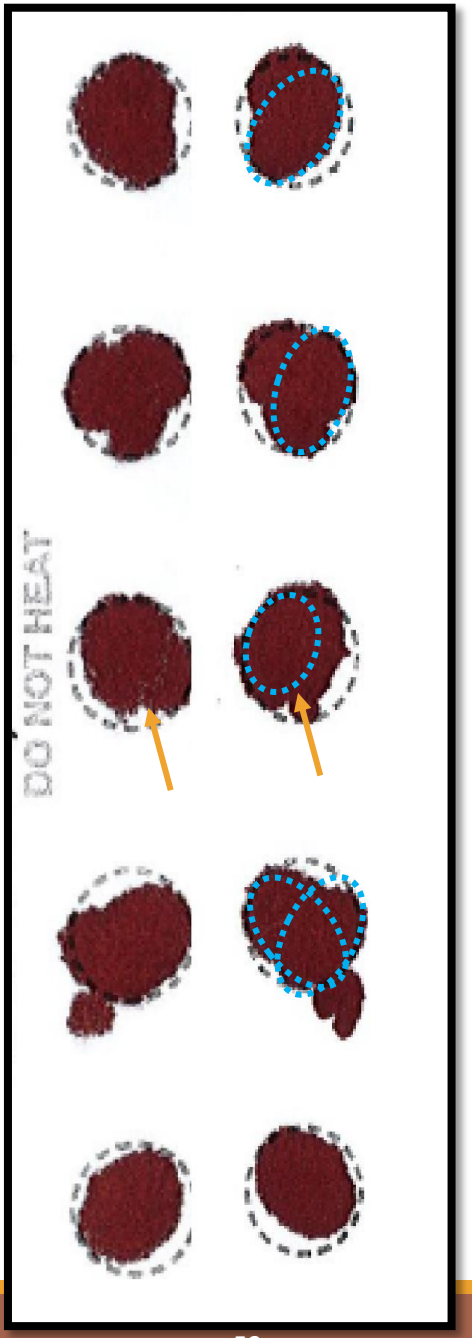


Fig. 1. Sample volume and quality factors.
(A), Effect of sample volume on bloodspot diameter. The white circles represent central and peripheral punch locations taken from each bloodspot during this study. (B), Examples of poor-quality bloodspots: a, double layered/applied to both sides of card; b, multispotted samples; c, insufficient sample applied (view of front of card); d, insufficient sample applied (view of back of card); e, 20-µL spots compressed.

Layering

- Presence of more than one spot, spots stacked on top of each other, overlapping spots, spots running into others, or samples applied on both sides of a filter paper.
- Impossible to tell where analytes disperse when spots touch
- Inaccurate concentrations of analytes among the blood spots lead to false positives



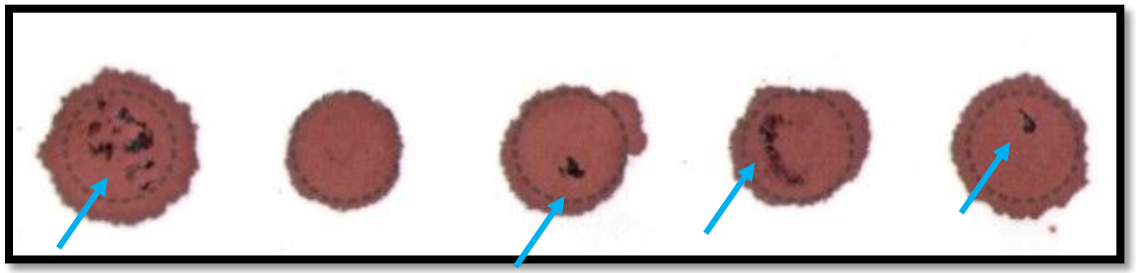


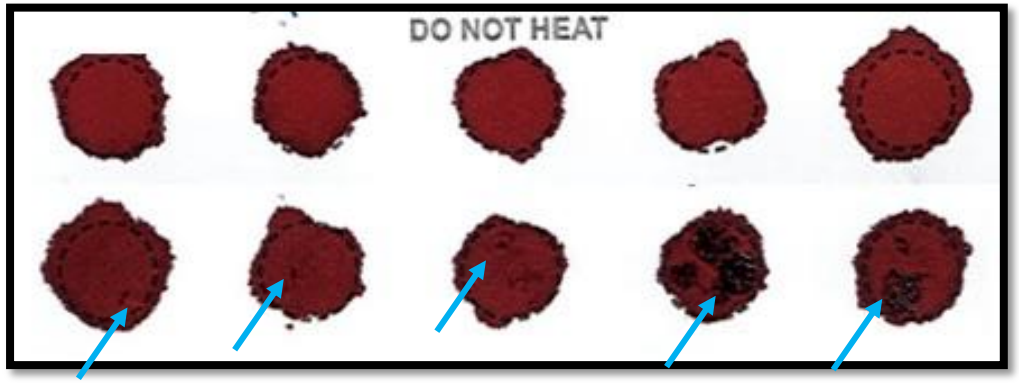
Layering Prevention

- ❑Apply only one large droplet of blood to each pre-printed circle!
- ❑Patience! Wait for a large drop to collect on the heel before applying to the filter paper.
- ❑Do not let spots touch.
- ❑If needed add spots outside of the pre-printed circles.
- ❑If layered spots are observed, add spots to the card where they will not touch or overlap. If this is not possible the collect a new specimen.

Clotting

- Scabs, blood clots, and obvious heel touches that have left a small area of densely layered blood.
- Caused by any of these issues: holding blood droplets too long before applying to the paper, improper drying of specimen, baby having high hematocrit, usage of capillary tubes, or touching the paper with the heel
- False positives

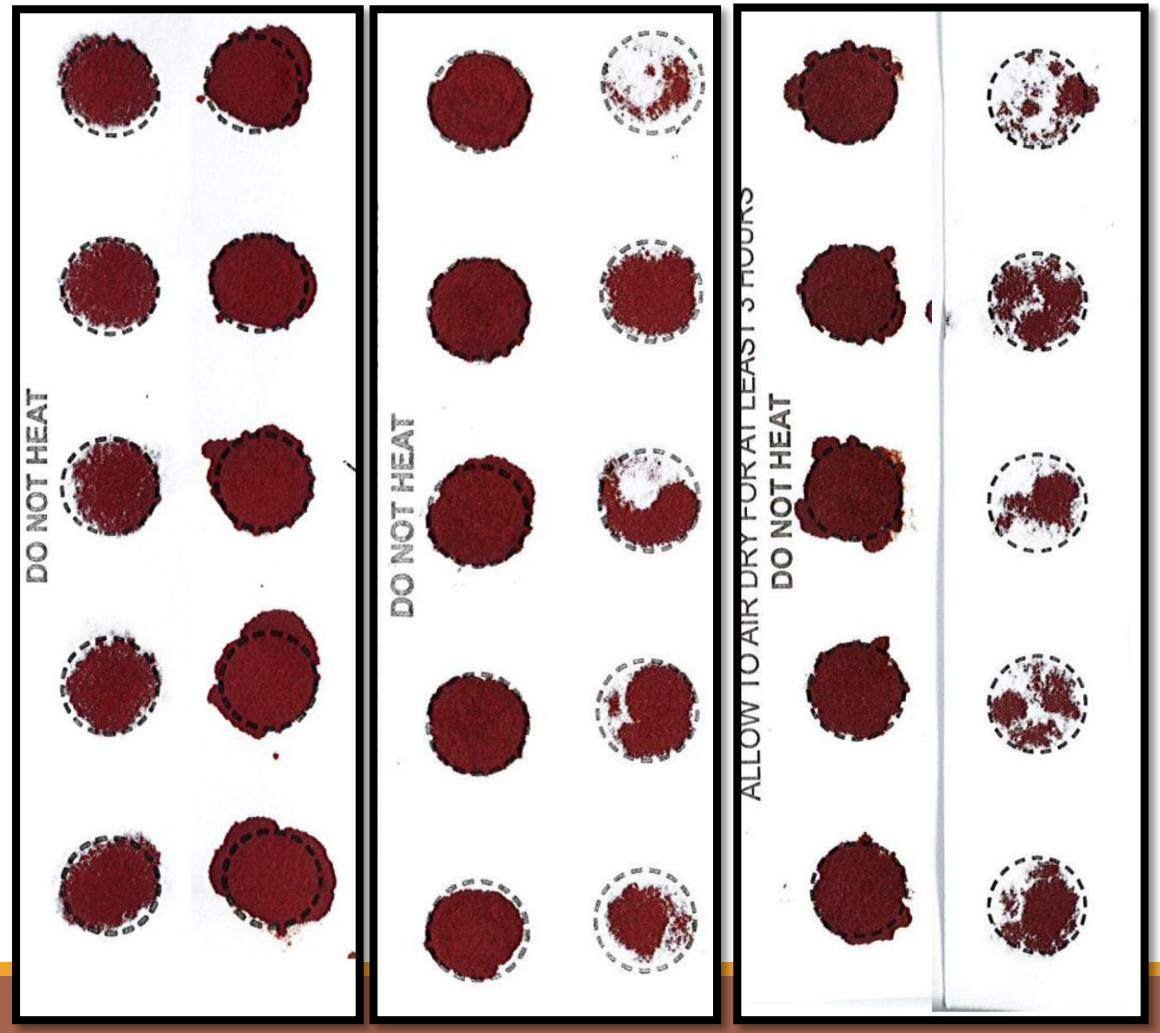




Clotting Prevention

- ❑Avoid using capillary tubes as they dramatically increase the likelihood for clotting.
- ❑Allow specimen to completely dry before packing for shipping. Contact from the paper on wet or damp blood cause clots and touch marks.
- ❑Do not touch the heel to the filter paper.
- ❑Avoid allowing blood droplets to age excessively before applying to the filter paper.
- ❑If clots are observed on the specimen before shipping collect a new specimen.

Didn’t Soak through



Blurry edges, white flecks and blank sections seen throughout the blood spot.

Often seen with high hematocrit children, or when small blood droplets are used.

False positives and negatives for all testing.

Didn’t soak through Prevention

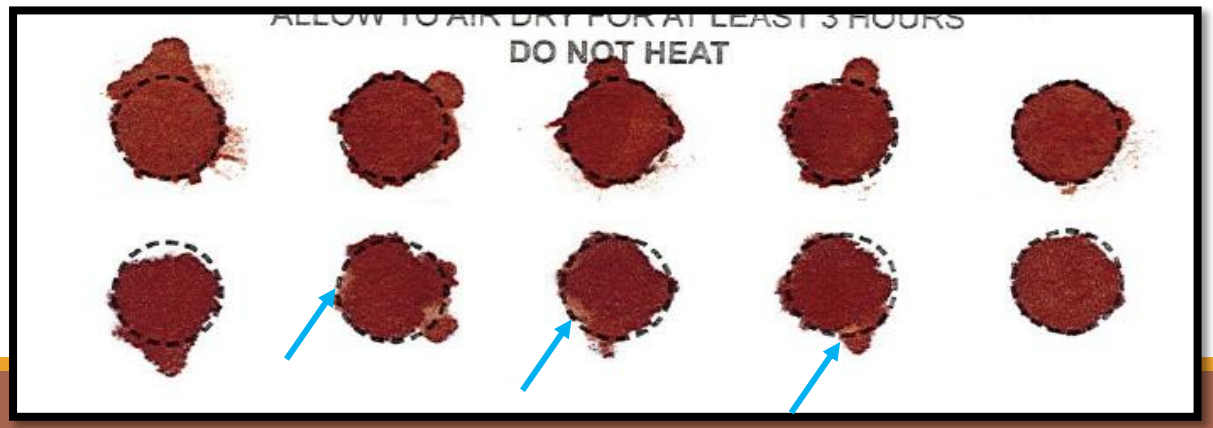
- ❑Wait for a single large blood droplet to form on the heel before applying to the filter paper.
- ❑Warm the infant’s heel before collection to improve circulation.
- ❑Do not touch the heel to the filter paper. Contact with the filter paper can contaminate or prevent uniform absorption.
- ❑Review specimen for proper absorption before shipping. If there is white filter paper visible in the blood spots collect a new specimen.

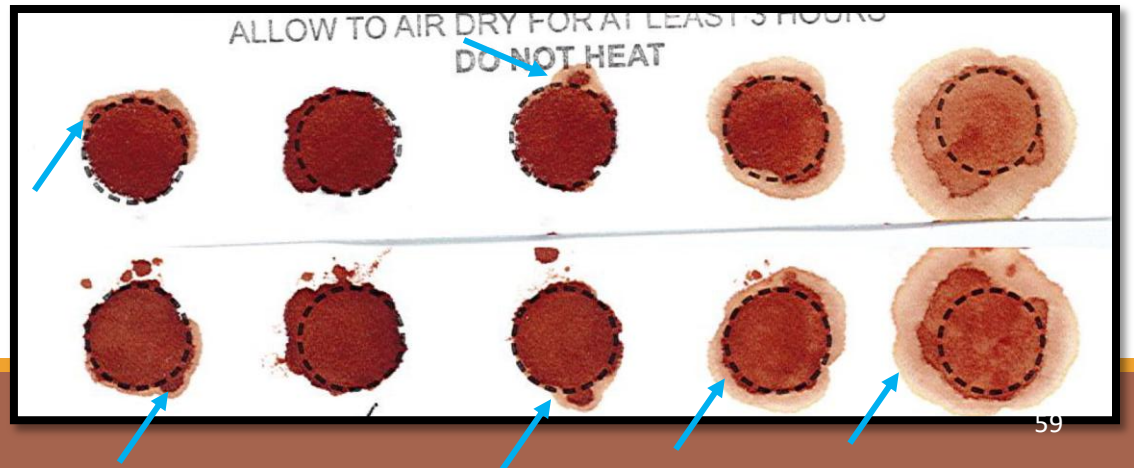
Serum Separation

“Halo rings” or lightening around the outside of a bloodspot where the red blood cells and plasma began to separate.

Use of capillary tubes with EDTA, drawing from a unproperly flushed line, or presence of Heparin greatly increases the possibility of serum separation.

EDTA and Heparin both interfere with reagents used for molecular testing and create inaccurate results.



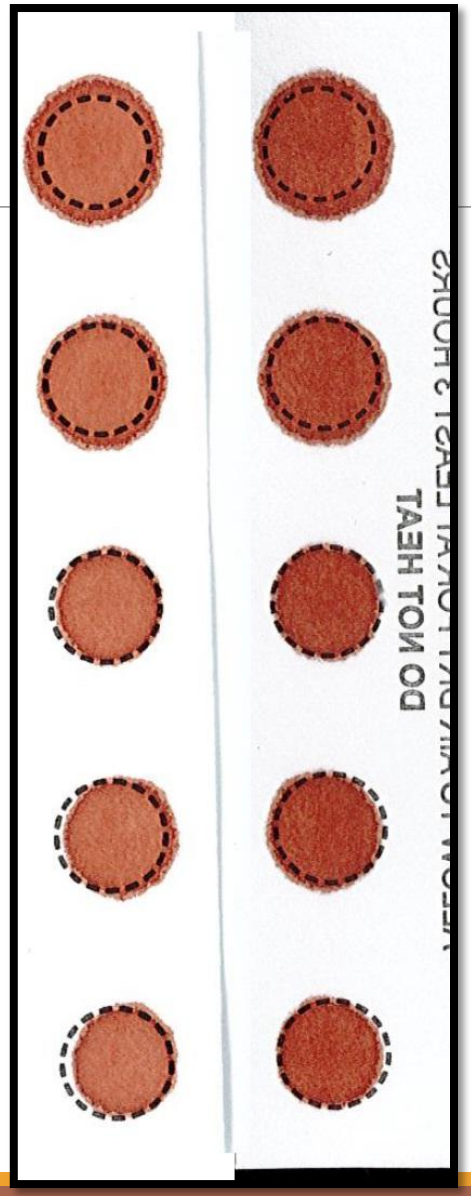


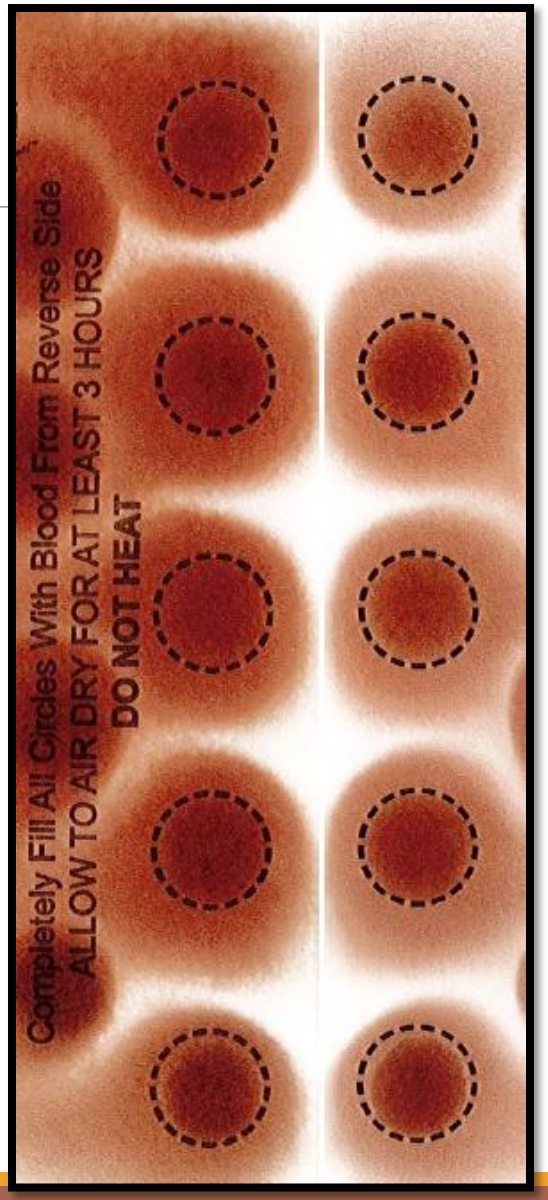
Serum Separation Prevention

- ❑Ensure the heel is clean and dry before collection.
- ❑Always use clean and dry gloves during collection and place the specimen to dry in area with no contaminates.
- ❑Do not squeeze/milk the heel.
- ❑Avoid using capillary tubes.
- ❑If pulling from an IV line ensure that the line has been properly flushed of all other fluids.
- ❑If halo spots are observed, then collect a new specimen.

Contamination/ Dilution

- ❑Exposure to ethanol, water, saline, or any other substance during collection, shipping or transportation that causes the specimen to become unusable or cause extreme lightening of blood.
- ❑Drawing from an IV line that has been improperly flushed will cause diluted specimens.
- ❑Severely anemic children may be flagged as diluted due to the lightness of the blood, a recollection is the best option.



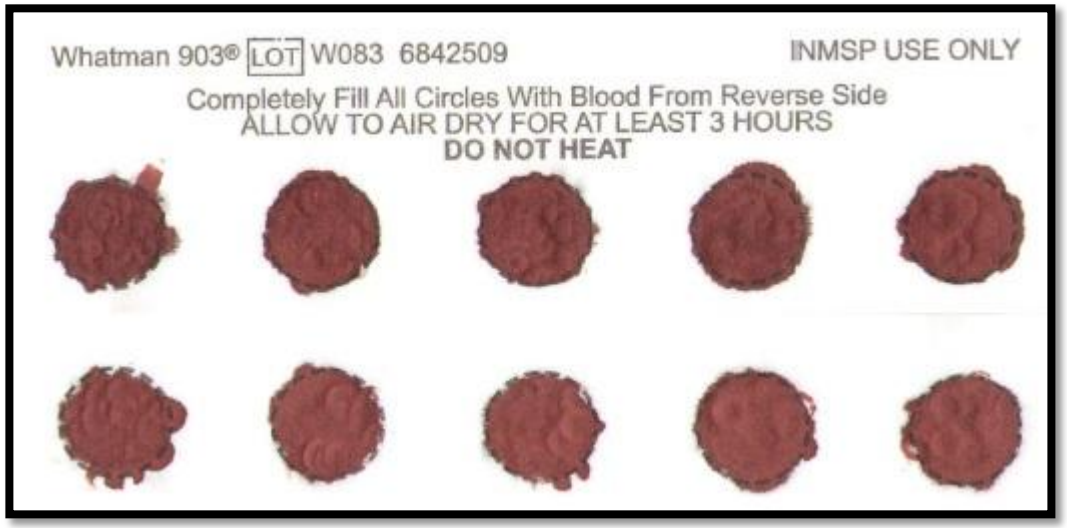


Contamination/ Dilution prevention

- ❑Always use clean dry gloves.
- ❑Ensure the heel is clean and dry before collection.
- ❑Do not use capillary tubes.
- ❑If collecting from an IV line be sure to clear line of all other fluids and contaminates.
- ❑Place the filter paper in a clean dry horizontal place to dry at least 3 hours prior to shipping.
- ❑Review specimen prior to shipping and if contamination is seen collect a new specimen.

Scratched/ Abraded

- ❑Capillary tube scratch marks, folds, rips and tears in the card
- ❑Use of capillary tubes to draw circles of blood create non-uniform analyte collections.

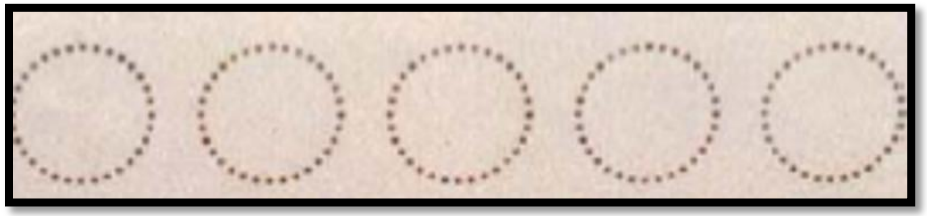


Scratched/ abraded prevention

- ❑Do not use capillary tubes.
- ❑Do not fold or bend the filter paper during storage, collection, or shipping.
- ❑Avoid over saturation as it increases the chances of tears in the filter paper.
- ❑Lay to dry in a flat horizontal location.

Other Reasons for rejected specimens

No blood applied



Failure to collect a specimen

Expired Form

- Filter paper past expiration date

Specimen older than 14 days past collection

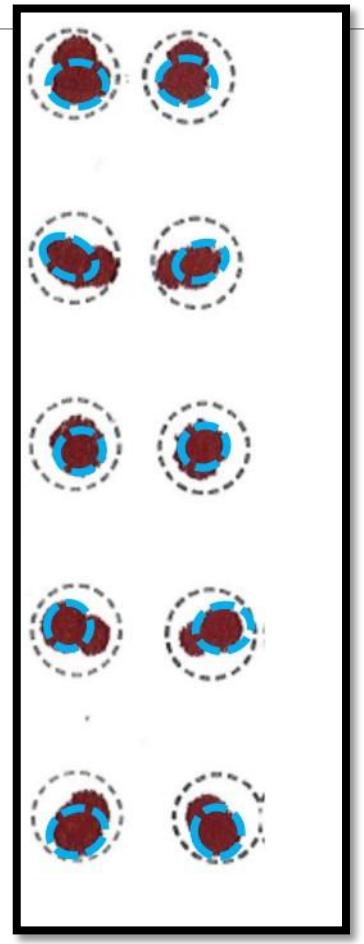
- Sample not sent in to SHL within appropriate time and is too old by date of reception

Specimens often have more than one reason for rejection

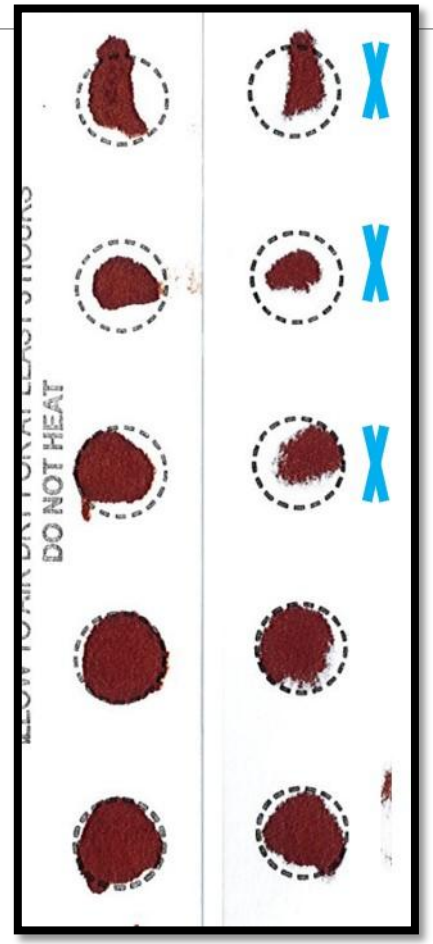
Only one rejection code can be assigned

- We use the code that applies to the most spots or is the root cause of the problems.

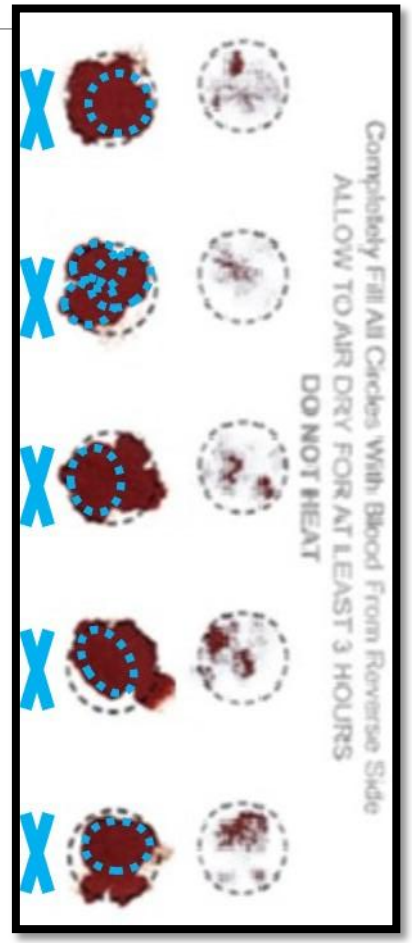
Not all spots need to be affected in the same manner, but if there are not 3 quality spots it will be rejected.



Layered + QNS



Didn't Soak through + QNS



Layered + Didn't Soak Through

How Poor Quality spots affect testing

Test	17-OHP	Phe	C8	T4	TSH	Galac	Biotin	Hgb
QNS	FN	FN	FN	FP	FN	FN/FP	FN/FP	FN/FP
Diluted	FN/FP	FN/FP	FN/FP	FN/FP	FN/FP	FN/FP	FN/FP	FN/FP
Serum Rings	FN/FP	FN/FP	FN/FP	FN/FP	FN/FP	FN/FP	FN/FP	N/A
Clotted/ layered	FN/FP	FN/FP	FN/FP	FN/FP	FN/FP	FN/FP	N/A	N/A

FN = False Neg., FP = False Positive

Recollecting immediately

Scenario: You've noticed that the first collection was unsatisfactory and drew a new screen immediately and aren't sure what to send to the lab or fill out on the new screen.

Awesome! Good job on properly reviewing specimen before shipping!

Answer: Completely fill out both specimen forms and send both forms. Mark both forms as the initial screen. The lab will automatically assign the better/passable of the screens as the one with reportable results.

Don't:

- Do not tear the filter paper off the demographic portion and attach to the original screen. Without the demographic information attached we can not use any of the blood from the attached filter paper.
- Do not send in an uncompleted form with blood collected on the filter paper. We have no way to verify who the blood belongs to. We can not trust that because two screens are paperclipped or stapled together that they belong to the same patient.

NOTE:

We can not use blood from two different cards for the same patient/barcode ID. We need a minimum of 3 good quality spots from an individual collection.

Second person verification

We strongly suggest that all forms are reviewed by at least two people for completion and quality of blood spots prior to shipping!

Verifier should check for:

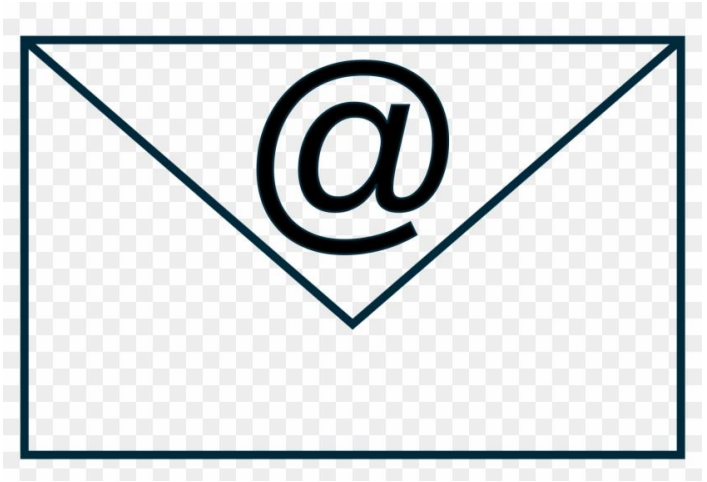
- Completion
- Legibility
- Accuracy
- Blood spot quality
- Internal record keeping- Does your facility keep the middle copy or have a log to track date of birth, collection, and shipping information? How do you track the specimen until results are received?

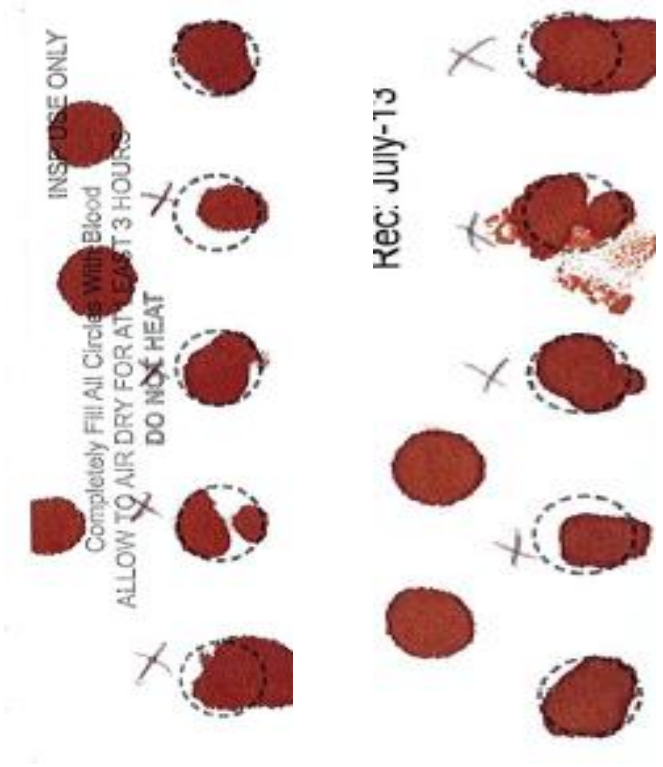
Is there a charge for repeat screening?

Although there is a charge for the initial screen, the South Dokata Newborn Screening Program does not charge for repeat screens. However, facilities collecting the repeat screen may have specimen collection charges, such as lab drawing fees.

Unsatisfactory Specimen Follow-up

Scanned images of rejected samples are emailed back to facilities along with an explanation of why it was unsatisfactory for education purposes. Retraining is encouraged for collectors repeatedly receiving rejection notices.





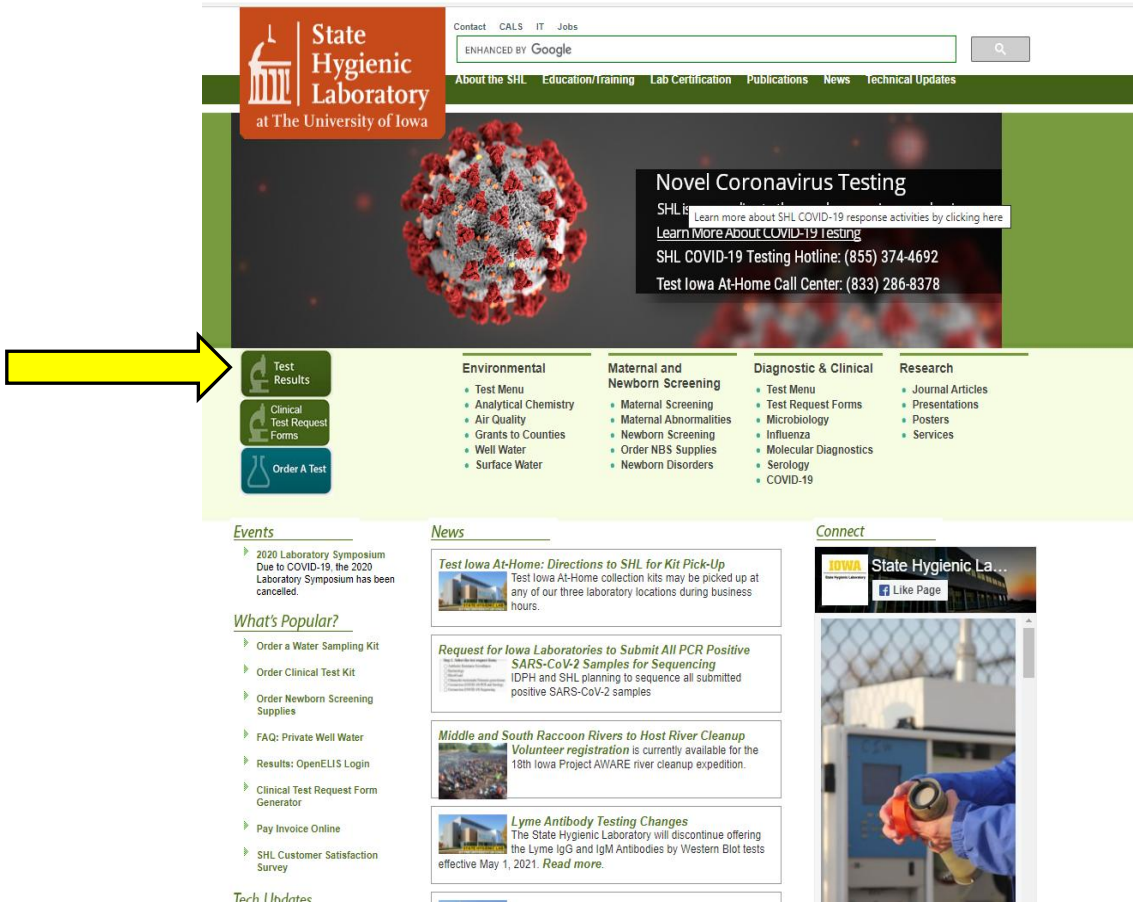
Sending Specimens with the Courier

- ❑South Dakota uses a daily courier.
- ❑Courier make collections every day of the year including holidays.
- ❑Plan collection times accordingly with your facilities pickup time so that specimen can be collected, fully dried and shipped on the same day if possible.
- ❑Same day pickups should occur if requests are made prior to 11 am.

Reporting Options are available based on needs

Screening results will be reported to the submitting facility once available. Reports can be sent in 3 ways:

- Paper reports- reports are mailed out USPS.
- Web Access reporting and Paper
- Paperless Web Access



South Dokata Newborn Screening Program Contacts

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South Dakota Newborn Screening Program Coordinator
South Dakota Newborn Screening Program
Office of Child and Family Services
South Dakota Department of Health
Gregory, SD
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