Newborn Bloodspot Screening Specimen Collection for Birth Facilities

Newborn Screening Follow-up Program
Prevention and Health Promotion Administration
Office of Genetics and People with Special Health Care Needs
443-681-3916

MARYLAND
Department of Health

MISSION AND VISION

MISSION

The mission of the Prevention and Health Promotion Administration is to protect, promote and improve the health and well-being of all Marylanders and their families through provision of public health leadership and through community-based public health efforts in partnership with local health departments, providers, community based organizations, and public and private sector agencies, giving special attention to at-risk and vulnerable populations.

VISION

The Prevention and Health Promotion Administration envisions a future in which all Marylanders and their families enjoy optimal health and well-being.

What is Newborn Bloodspot Screening?

•Newborn Bloodspot Screening (NBS) is a service provided by the Maryland Department of Health and Mental Hygiene for families with new babies.

•NBS identifies newborn babies with certain rare, serious disorders of body chemistry that if not detected early can cause mental retardation, serious illness or even death of the baby.

•NBS tests for over 50 possible disorders of body chemistry.

Newborn Bloodspot Screening: It's not just a "PKU" anymore

- Calling the newborn bloodspot screening a "PKU" is misleading
- PKU is just 1 of over 50 disorders included in the newborn screen.
- NBS screens for certain endocrine disorders; hemoglobin disorders; cystic fibrosis; and disorders breaking down lactose *(sugar in milk-based formulas and in breast milk)* and proteins.
- Screening for Severe Combined Immunodeficiency Disorder (SCID) was added in April 2016. New disorders are scheduled to be added in the near future.
- Some of the disorders on the newborn bloodspot screen are life-threatening and need immediate intervention.

Parents need to understand that this screening could save their baby's life!

Newborn bloodspot screening must be offered to all babies born in Maryland.

In 2008, the State of Maryland changed from an informed consent policy to an informed dissent policy.

Under the informed dissent policy, parents must be informed about the purpose of newborn bloodspot screening. It is the birth facility's responsibility to educate parents about the purpose for newborn bloodspot screening. ***Written consent is no longer required.*** Documentation is only needed ***if the parent refuses*** newborn screening.

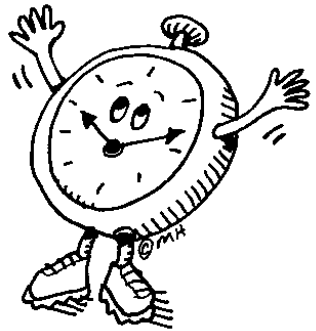
What Should Parents Know About NBS?

- NBS is designed to identify a possible disorder in their baby before any symptoms are present.
- Most of the disorders are rare. The most common disorders identified on newborn screening in Maryland are congenital hypothyroidism and sickle cell disease.
- Symptoms of hypothyroidism are usually not evident in a baby until irreversible brain damage has occurred.
- Early treatment with antibiotics can save the life of a baby with sickle cell disease.

The most important teaching point.….*NBS can help prevent developmental delays and may save their baby's life!!*

When is Newborn Bloodspot Screening Performed?

The first NBS should be collected after the baby has had at least 24 hours of feeding. This specimen is usually collected in the birth hospital.



- *If baby is born at home, discharged prior to 24 hours of age, or had less than 24 hours of feedings prior to the specimen collection, an initial NBS will need to be performed in the provider office.*

In Maryland, our screening process is based on two specimens. The second NBS should be collected between 7-14 days of age. This specimen is usually collected in the provider's office.

When is Newborn Bloodspot Screening Performed for NICU Babies?

•There is a different schedule for babies who are in the special care nursery (SCN) or neonatal intensive care unit (NICU). Specimens are usually collected:

- ✓ upon admission to the NICU
- ✓ at 2-3 days of age
- ✓ 10 days of age
- ✓ 1 month of age or discharge if baby is able to go home before 1 month of age

•*If baby has been in the NICU and has had a satisfactory and normal 10 day/discharge specimen, the provider office does not need to collect a repeat specimen.* ***If there are no normal results for hemoglobin, biotinidase and GALT prior to a transfusion****, the baby needs a repeat specimen collected at the provider office 4 months after the last transfusion.*

What conditions are included in Newborn Bloodspot Screening?

Metabolic Disorders:

- Biotinidase Deficiency
- Galactosemia

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Biotinidase Deficiency

•Biotin is one of the B vitamins that helps break down proteins.

•Biotinidase removes biotin from the protein after the protein has been broken down so the body can reuse the biotin.

•In biotinidase deficiency, the biotin cannot be re-used so the supply in the body decreases.

•Individuals with biotinidase deficiency can not obtain enough biotin through their diet alone.

•Treatment involves taking biotin supplements. If not treated, individuals can have developmental delays and deafness, as well as hair and skin problems.

•This disorder is suggested by NBS if there is an abnormal Biotinidase level.

Galactosemia

•Galactosemia is a disorder in which a baby cannot metabolize galactose, which is broken down from lactose (the sugar in milk products).

Normal metabolic pathway: Milk ⇒ Lactose ⇒ Galactose ⇒ Glucose ⇒ Energy

•A baby with classical galactosemia has an abnormal amount of the GALT enzyme and the galactose level is elevated. ***Classical galactosemia is a life threatening condition. Save lives through prompt screening!***

Galactosemia Pathway: Milk ⇒ Lactose ⇒ ↑Galactose ~~⇒~~ Glucose ⇒ Energy

•To help test for galactosemia, it is very important to note whether baby is breast feeding, feeding with a lactose formula (milk-based) or a non-lactose formula (soy-based) at the time of the collection of the NBS.

Galactosemia

•Classical galactosemia is the complete lack of the GALT enzyme. This form of galactosemia is life threatening and can cause death in the first week of life, usually from sepsis. Too much galactose can also cause liver and kidney problems, as well as cataracts.

•The GALT enzyme can be normal and the baby may still have a form of galactosemia. Other enzymes are also used to break down galactose. If the baby has a normal GALT and elevated galactose on NBS, further testing is needed.

•Treatment for galactosemia involves use of a soy based formula like Isomil® or ProSobee®

•NBS gives a qualitative measure of GALT and also measures the amount of galactose in the blood.

What conditions are included in Newborn Bloodspot Screening?

Fatty Oxidation Disorders:

•Some babies are missing an important enzyme to break down fatty acids.

• Different enzymes are needed to break down long, medium and short chain fatty acids.

•Some babies cannot break down stored fats and some babies can not tolerate any fat.

•Affected infants require a constant source of energy and **CANNOT** go without eating. Fasting will cause brain damage and eventually death.

•These disorders are suggested in NBS by elevations in the Acylcarnitine Profile.

What conditions are included in Newborn Bloodspot Screening?

Amino Acid and Organic Acid Disorders:

•Some babies are missing an enzyme for breaking down certain amino acids or proteins

• Inability to break down proteins causes toxic levels of organic acids and/or amino acids to build up in the body

•The name of disorder is often based on the name of the amino acid or organic acid involved.

•These disorders are suggested in NBS by either elevations in the Amino Acid Profile or by certain elevations in the Acylcarnitine Profile.

What conditions are included in Newborn Bloodspot Screening?

Amino Acid and Organic Acid Disorders:

•Phenylketonuria (PKU) is an amino acid disorder. It is caused by the inability to completely break down the protein called *phenylalanine*. PKU needs to be treated promptly to prevent irreversible brain damage.

•Maple Syrup Urine Disease or MSUD is also an amino acid disorder. MSUD is caused by the inability to completely break down the branched chain amino acids – *leucine, isoleucine* and *valine*. ***It is very important to know MSUD is a life-threatening condition.*** The baby's urine or ear wax can have a sweet smell like maple syrup. If any baby is noted to smell like maple syrup, the baby needs to be evaluated immediately for possible MSUD, regardless of NBS results.

•All of the amino acid and organic acid disorders need to be treated promptly. Treatment involves providing the infant with a special diet and/or supplements.

What conditions are included in Newborn Bloodspot Screening?

Urea Cycle Disorders:

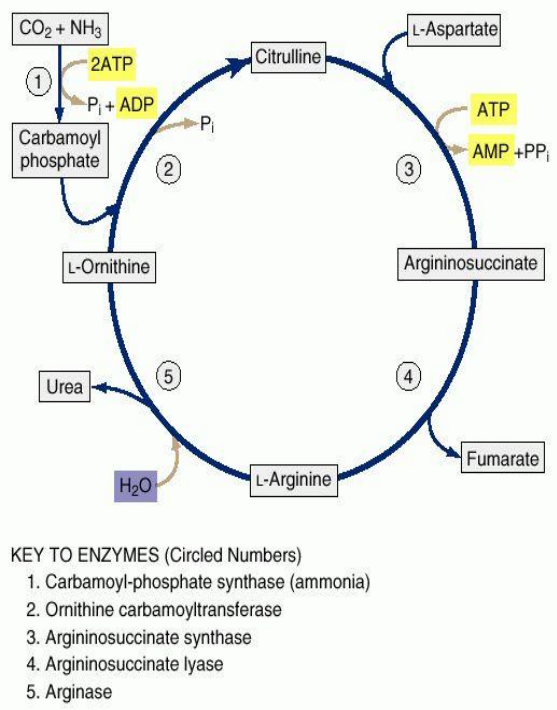
•Some babies are missing enzymes for breaking down certain amino acids used in the urea cycle.

• Inability to break down these proteins causes ammonia levels to increase.

• Baby may become lethargic and begin to seize prior to discharge from the hospital.

• All of these disorders need to be treated promptly. Treatment involves giving the baby a special diet and/or supplements.

• It is important to note that not all Urea Cycle disorders are picked up by NBS



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What conditions are included in Newborn Bloodspot Screening?

Endocrine Disorders:

- Congenital Adrenal Hyperplasia
- Congenital Hypothyroidism

Congenital Adrenal Hyperplasia (CAH)

•Caused by defects in enzymes used for hormone production, including sex hormones and aldosterone.

•Females may present at birth with ambiguous genitalia causing gender misassignment. Males may appear physically normal at birth.

•Classical CAH is life threatening. Crisis symptoms can occur soon after birth. Within a few weeks the baby may present with dehydration, vomiting, lethargy, poor feeding and shock.

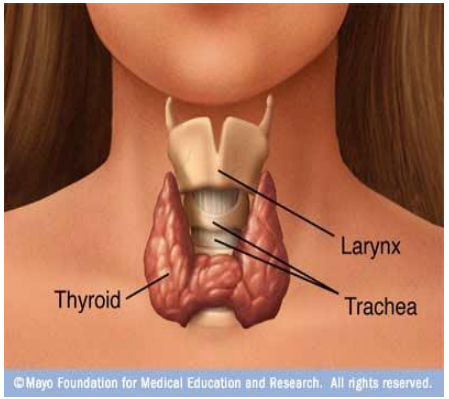
•A normal sodium level does not rule out all forms of CAH.

•Other forms of CAH may result in virilization of females and premature bone growth.

•This disorder is suggested in NBS by an elevated 17-OHP level. The 17-OHP level can be elevated in sick and premature infants as well.

Congenital Hypothyroidism

- One of the most common disorders identified through NBS
- Caused by the inability to secrete or produce thyroxine
- Most common cause of preventable brain damage
- Baby may have an enlarged tongue or lump in the neck



- Primary reason for 2nd NBS since baby may not have a low thyroxine level in the first 24-48 hours of life due to maternal thyroxine levels or medication
- Goal is to initiate treatment and have appropriate thyroxine levels by 3 weeks of age
- This disorder is suggested through NBS by a low Thyroxine level and an elevated TSH level.

What conditions are included in Newborn Bloodspot Screening?

Hemoglobin Disorders:

•NBS screens for Sickle cell disease and other hemoglobinopathies

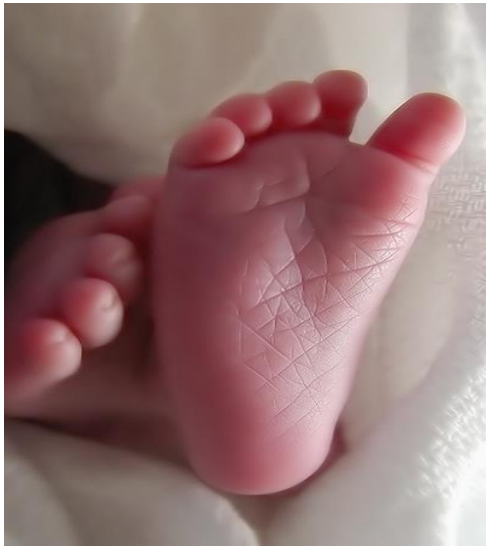
•Some hemoglobin disorders are caused by a defect in the structure of hemoglobin in red blood cells

•In sickle cell disease the shape of the red blood cell transforms into a “sickle” shape and causes blood to back up in the body, causing pain and possible damage to body organs. There is also a greater risk for infection in babies and young children.

•Effective treatment helps reduce problems associated with sickle cell disease.

What conditions are included in Newborn Bloodspot Screening?

Cystic Fibrosis:



•NBS screens for Cystic Fibrosis (CF) by measuring immunoreactive trypsinogen (IRT) levels in the blood. Elevations in IRT can indicate CF. ***All babies who have a meconium ileus or plug must be tested for CF, regardless of IRT results.****

•Cystic Fibrosis causes thick, sticky mucus in the lungs which leads to problems with breathing and frequent lung infections.

•Babies may have problems digesting food, causing slow growth.

•Treatment includes breathing treatments (including chest percussion) as well as antibiotics for the lungs. Medicine and a special diet are given to babies with digestive problems. Early identification and treatment provides a better quality of life and a longer life span.

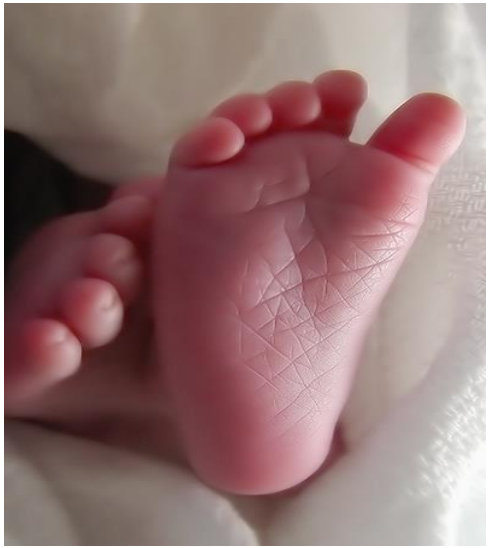
What conditions are included in Newborn Bloodspot Screening?

Severe Combined Immunodeficiency Disorder:

•NBS screens for Severe Combined Immunodeficiency Disorder (SCID) by identifying the amount of TREC s (t-cell receptor excision circles) in the blood.

•T-cells help fight infections. If T-cells are low, the baby cannot fight infections well.

•There are other conditions, such as DiGeorge syndrome, prematurity and use of steroids, that may cause lower number of T-cells.



Delays in Newborn Bloodspot Screening

- Newborn Screening is a life saving practice
- Timing is crucial in newborn bloodspot screening
- Delays for any reason can cause infants with certain disorders to die before parents realize something is wrong
- The sooner a disorder is identified, the earlier treatment can be started
- Infant outcome depends on newborn screening

Specimen Quality

- The Maryland State Newborn Screening Laboratory receives hundreds of specimens each *day* - both from birth hospitals and provider offices.
- Unsatisfactory specimens slow down the testing.
- The specimen may be unsatisfactory secondary to missing vital information on the lab slip or blood spots which are not acceptable for testing.

Frequently Missing Information

- Birth Date and Collection Date

  Used to determine age of infant at the time of collection

  Date of collection also determines age of blood at the time of analysis

- Baby's current weight

This information is very important for proper laboratory analysis of the results*

- TRAB (Test Request Authorized By) – the name of the provider ordering the test must be on the form in order for the lab to release the result

“Unsatisfactory Specimen”

- If the blood specimen does not give reliable results, it will have to be repeated
- Getting a repeat specimen can take weeks, putting the newborn baby at risk
- Always check the specimen before sending to make sure it appears acceptable

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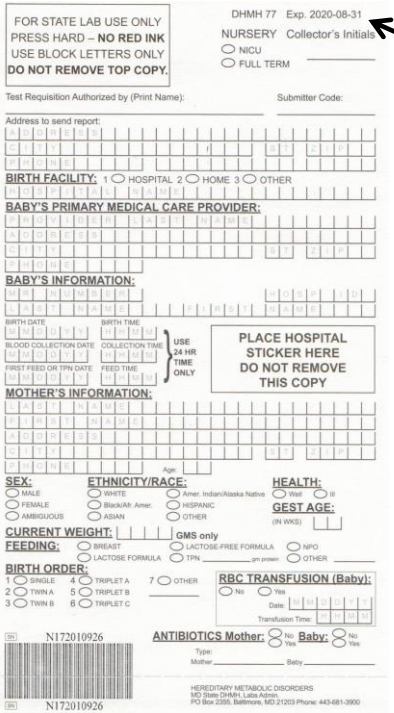
Collecting a Specimen

Completion of Collection Form



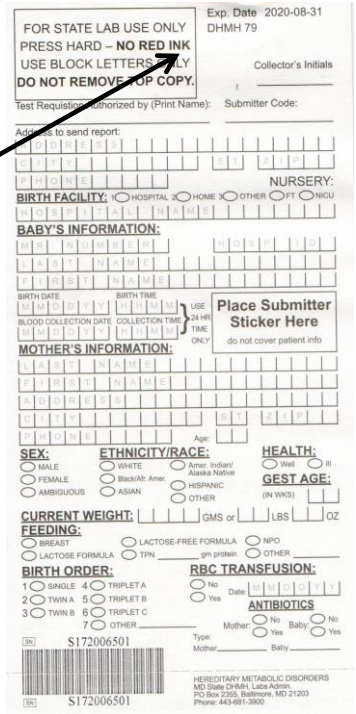
Use DHMH 77 for babies <7 days of age

Use DHMH 79 for babies >7 cays of age

Check expiration date prior to collection of blood. If expired, call 443-681-3900.

Complete requested information on card prior to collection of blood

Please remember that all requested information is important for identification of the baby or for evaluation of results.



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Completion of Collection Form

Specimen cards are legal documents.

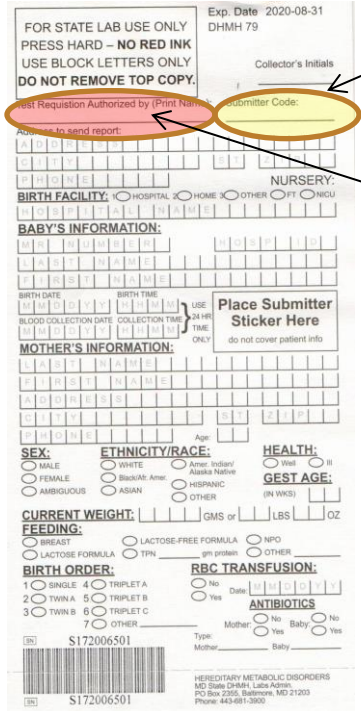
Accurate information is crucial for valid and reliable testing, as well as linking of initial and repeat specimens.

Use block letters in the spaces provided. Use of BLOCK LETTERS allows for faster and more accurate data entry into the State Lab computer system

Stickers **should not** be applied to the front of the collection form. Identification stickers, if used, should be placed on the back side of the top white slip.

Do not place any stickers either on the front or the back of the actual filter paper.*

Completion of Collection Form



Write in your Submitter Code. If you do not know your code or do not have a code, please call 443-681-3900.

Test Request Authorized By (TRAB) – this line must contain the name of the provider who is ordering the test. Results cannot be released if the TRAB is not completed

Using BLOCK LETTERS fill in all demographic information for provider's office and for mother.

The purpose of this information is to provide a way to link the baby's first and second specimens and to find the infant with a positive result quickly

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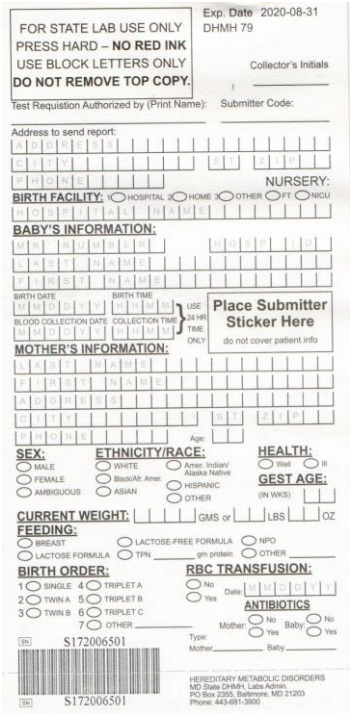
Completion of Collection Form



Occasionally, special circumstances arise such as in the case of an adoption, surrogacy or involvement of protective services

If baby is adopted, please write “adoption” on the lab slip and write in the adoptive parent’s name and phone number. If known, also indicate birth mother’s name on the slip.

If the birth mother is a surrogate, please write “surrogate” on the lab slip and write in the contact information for the custodial parents. If known, also indicate surrogate mother’s name.

If baby is in foster care, write “foster care” on the lab slip and indicate name and contact information for foster parent, along with birth mother’s name

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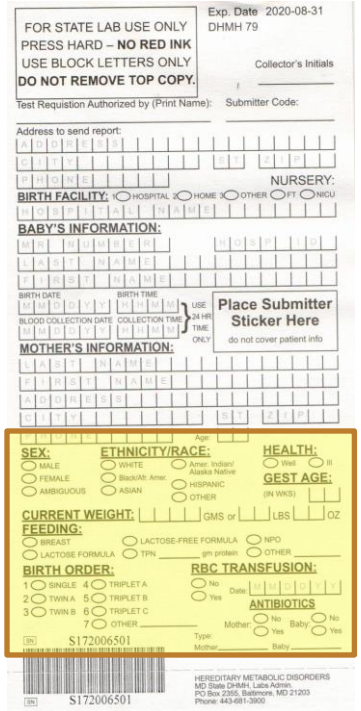
Completion of Collection Form



Health and gestational age is very helpful in interpretation of the results.

Infant's current weight is important because some cut-offs are based on the infant's weight

Type of feeding an infant is receiving is very important as well. Soy formulas are lactose-free. Most regular formulas are lactose formulas. If baby is nursing and being supplemented with formula, please indicate both breast and type of formula.

Indicate whether this is a single birth, twins, triplets, etc. If this was a multiple birth, it is very important to note which infant is Twin A, Twin B, Triplet A, etc. to help identify which subsequent specimen should be matched to the initial specimen.

It is also important to note if mom or baby have been on antibiotics within 48 hours of the specimen collection.

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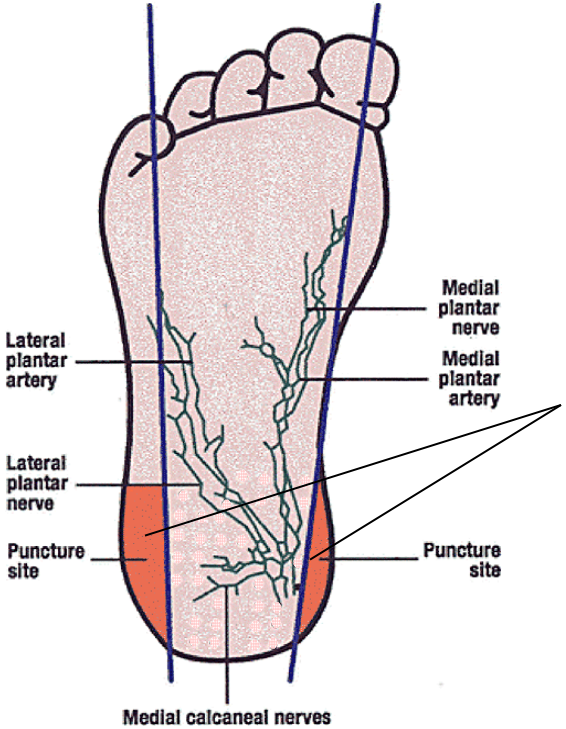
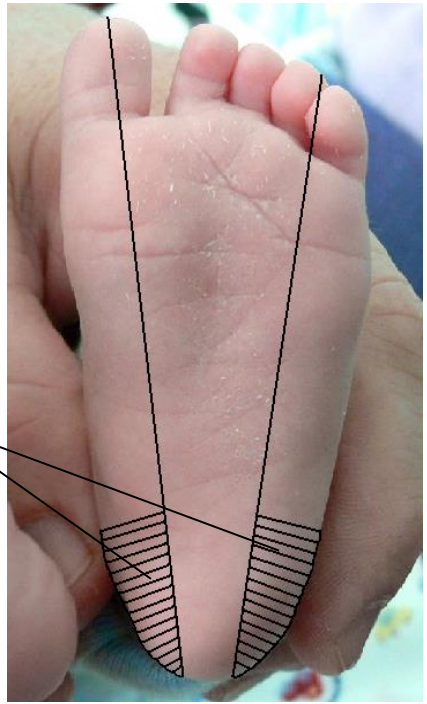
Recommended sites for neonatal capillary blood sampling



Puncture site should be on the medial or lateral portion of the plantar surface of the heel. (The plantar surface is the part of the foot that touches the floor if standing or walking)

The *shaded areas* show the best sites for puncture. Use of these areas will help prevent possible damage to the heel bone and the nerves and arteries noted on the diagram.



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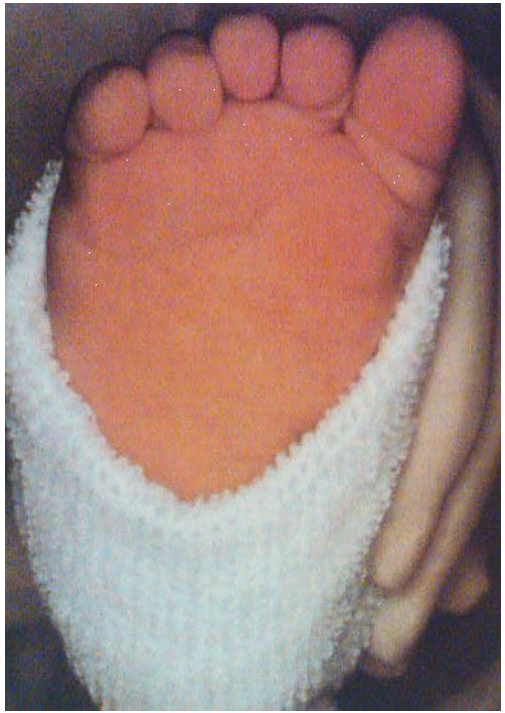
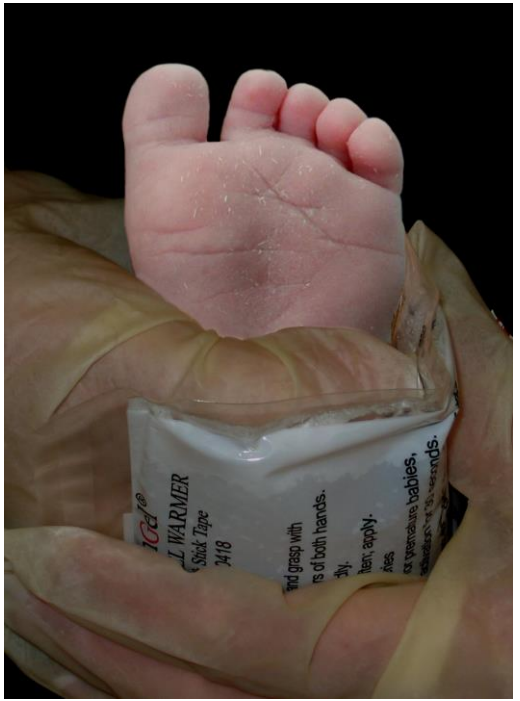
Preparation for neonatal capillary blood sampling



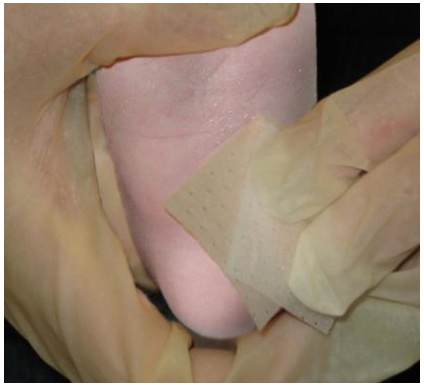
Warm infant's heel with soft cloth or diaper, moistened with warm water for 3 to 5 minutes.

You can also used a commercially prepared disposable heat pack. (make sure to follow your facilities guidelines for using heat packs)

Place infant's heel in a dependent position, such as Mom cradling infant with head higher than feet.

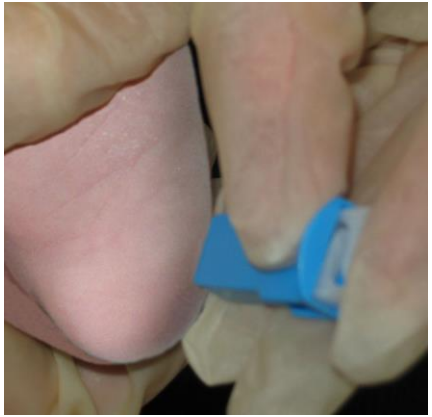


Neonatal capillary blood sampling



Cleanse site with alcohol prep and allow to dry.

Puncture heel with sterile lancet which has a puncture depth of less than 2.0 mm. The lancet should be labeled for use in newborn capillary specimen collection.



Wipe away first blood drop with sterile gauze pad.

Allow another LARGE blood drop to form

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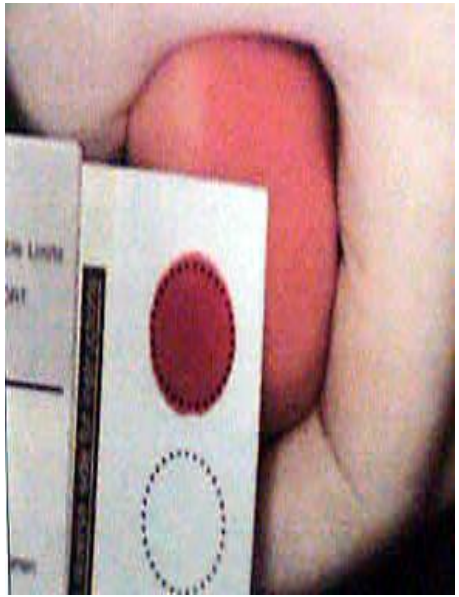
Neonatal capillary blood sampling



Direct application of blood onto the filter paper is recommended. Capillector tubes should not be used as they may scratch the top of the filter paper.

Lightly touch filter paper to LARGE blood drop.

Filter paper acts like a capillary tube, drawing blood into itself

Allow blood to soak through and completely fill circle with SINGLE application. *(To enhance blood flow, VERY GENTLE intermittent pressure may be applied to the heel. Do not milk or squeeze tissue next to the puncture site because this may cause serum to separate.)*

Apply blood to only one side of the filter paper

Fill remaining circles by lightly touching one LARGE blood drop to each circle.

If blood flow diminishes, clean a new site and repeat the process with a new sterile lancet.

Important Points to Remember

Do not touch the actual filter paper portion of the lab slip either before, during or after blood collection. Contamination of the filter paper with water, formula or powder from gloves will affect the results.

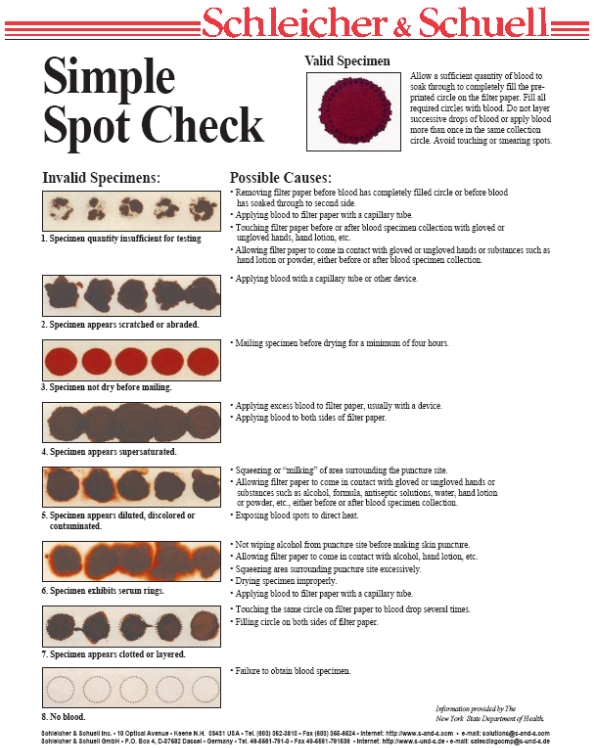
Check the specimen to make sure the blood saturated through the card and there is no overlapping of blood in the circles. If there is a problem with the specimen, test should be repeated.

Allow specimen to dry in on a clean flat non-absorbent surface for a minimum of 4 hours.

Completed specimen should be mailed to the Maryland State Laboratory within 24 hours of collection. Do not hold or “batch” specimens while waiting for several specimens to be mailed together.

If the office is contacted by a member of the Newborn Screening Follow-Up Unit regarding abnormal results, prompt action is needed because the baby has been identified as “at risk” for one of the conditions.

Common Problems with Specimen Collection



The most common problem with newborn bloodspot screening specimens is layering of blood on the circles.

Another common problem is not getting enough blood on the circles or letting the blood soak through the filter paper.

Scratching of the filter paper occurs frequently when capillector tubes are used and the blood is “colored” on the spots.

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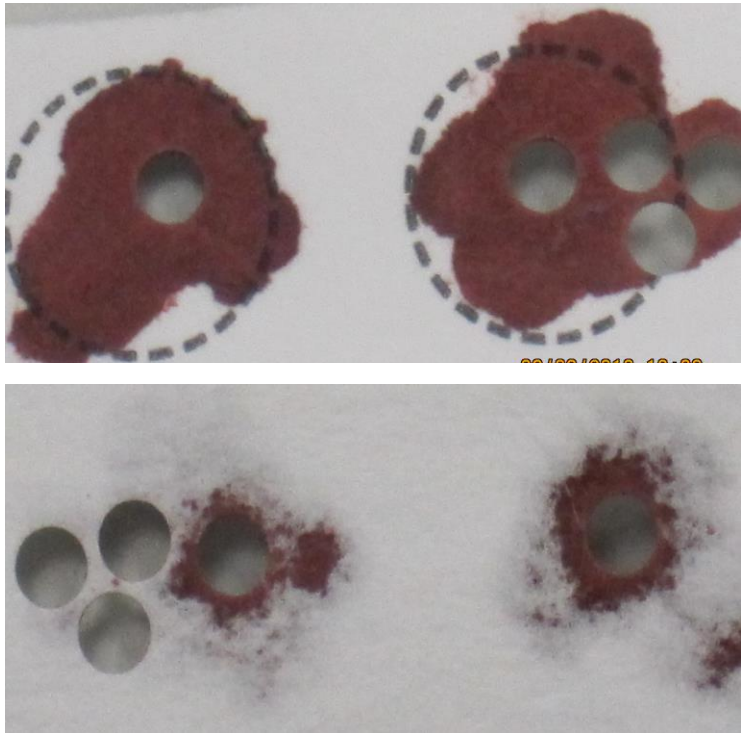
UNS 1- Insufficient Blood to Run All Tests



From the front of the card, this specimen appears to be satisfactory.

Turning over the same specimen card shows that the blood did not saturate through the filter paper to the back of the card.

If a specimen looks like this, start over with a new card. Never apply blood to both sides of the card.*

UNS 4 – Clots or Tissue on Surface

The darkened areas in the spot on the right indicate the presence of clots. These clots can occur if blood sits in a capillector tube prior to application to the filter paper. Direct application of blood from the heel is the preferred method for newborn screening specimen collection.

UNS 6 – Filter paper stretched or wrinkled

Possible causes: Bending filter paper during application

Applying too much blood to one spot

Newborn Screening Results

Results from the first screen are provided to the submitter, which is usually the hospital of birth

If the baby has an abnormal screen, the medical home/primary care provider (PCP) listed on the screening card is called with the results

Second screen results are mailed to the Medical Home/PCP who is listed as the submitter on the lab slip

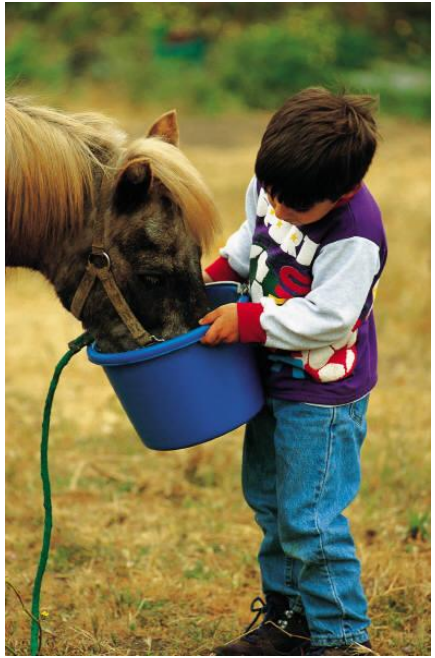
If there are abnormal results on the second screen, the PCP office will be called. A third screen or confirmatory testing may be requested

If results are not received within two weeks of submission, please contact the Maryland State Newborn Screening Laboratory at 443-681-3900 to check on status of testing

You Can Save a Life

Prompt and correct newborn screening practices can save the life of a baby

With early intervention and treatment, even babies with serious diseases can live a long, happy life



Newborn Screening Program

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phpa.health.maryland.gov/genetics

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Prevention and Health Promotion Administration

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