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## Newborn Screening (NBS)

Laboratory (615) 262-6352  
Results and Follow-Up (615) 532-8462



### I. Introduction

Tennessee law requires that a blood sample shall be obtained from each infant born in the state, regardless of age, before discharge from the hospital and tested for specific genetic disorders. These genetic disorders can cause mental retardation or death if not treated quickly. To prevent the early affects of these disorders, the blood sample should be drawn from the infant 24 hours after birth and less than 2 days of life (24-48 hours from birth is the optimum window for sample collection.) Infant's who are screened before they are 24 hours of age must be rescreened within 24 to 48 hours by a local health department, or private physician. Infant's who are born in a non-hospital setting must be taken to a hospital, local health department, or private physician between 24 and 48 hours of birth to have the blood sample collected. Drops of blood from the infant's heel are absorbed into a special filter paper attached to the Newborn Screening Form PH-1582 and sent to the Newborn Screening Laboratory at the Tennessee Department of Health (TDH) Laboratory Services. The current protocol includes testing the blood sample for the following disorders:

#### CONGENITAL HYPOTHYROIDISM (TSH)

Hypothyroidism occurs when the body does not produce enough thyroid hormone from the thyroid gland. This hormone is called thyroxine, which is needed for brain and body growth. A decreased amount of thyroxine can interfere with normal growth and can lead to mental retardation. If detected early and hormone replacement is initiated, normal growth and development can take place.

#### HEMOGLOBINOPATHIES

Hemoglobin is the part of red blood cells that carries oxygen. Hemoglobinopathies are diseases that affect the kind or amount of hemoglobin a person has in the red blood cells. Some hemoglobinopathies can cause anemias or thalassemias.

Infants of **Asian** or Mediterranean background, such as Italian, Greek, Arab, or Kurdish, should be rescreened for thalassemias at the 12-month routine child-health visit. These specimens can be sent to the Meharry Sickle Cell Center or other clinical laboratories.

Sickle cell disease is the most common hemoglobinopathy. The red blood cells are sticky and crescent or sickle-shaped and therefore do not move easily through the vascular system, decreasing the vital levels of oxygen carried throughout the body. There is a dramatic decrease in infant mortality and life-threatening complications when an infant is identified early and treated with antibiotics. Testing in Tennessee began in 1988 and the incidence rate nationally is 1:500<sup>(1)</sup> in the Africa-American population. People of Hispanic, Asian, Arabic, or Mediterranean decent are also more likely to have hemoglobinopathies.

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<sup>(1)</sup>Burtis, C., Ashwood, E., Bruns, D., Tietz Fundamentals of Clinical Chemistry, 6<sup>th</sup> edition, pages 825-835

## **Newborn Screening (Continued)**

### **GALACTOSEMIA (GAL)**

Infants with classical galactosemia lack an enzyme, uridyl transferase, needed to breakdown galactose, a kind of major sugar found in milk. Due to either an absent or low enzyme, galactose accumulates in the body leading to mental retardation, growth deficiency, blindness, overwhelming infection and death. Infants with galactosemia can rapidly become sick after only a few days of normal feeding. Infants who are detected early are placed on a lifelong special galactose-free diet.

### **CONGENITAL ADRENAL HYPERPLASIA (CAH)**

In order for this test to be reported accurately it is imperative that the **CURRENT weight** of the infant be recorded correctly on the Newborn Screening Form. This disorder results from a deficiency in one or another of the enzymes of steroid syntheses. The deficiency results in an abnormal accumulation of hormones important for normal growth and development of puberty. Most affected infants have a second defect that affects normal electrolyte balance. Infants having this deficiency can become very sick within 1 to 2 weeks of birth. Female infants with this disorder may be assigned the wrong sex at birth. Male infants are often not identified until they are in crisis. With early diagnosis and treatment it is possible to achieve normal growth and development of puberty. Because this disorder is complex, infants may need to be monitored by a pediatric endocrinologist.

### **BIOTINIDASE DEFICIENCY**

This disorder is caused by the lack of an enzyme in the baby's body called Biotinidase. Babies with Biotinidase Deficiency can have seizures, skin rash, or infection, developmental delays and hearing loss. Problems with the disorder can be prevented with treatment using biotin.

### **AMINO ACID DISORDERS**

Amino acid disorders are a group of conditions in which there is a problem with breaking down certain components of food called amino acids. These disorders are caused by a specific defect in one of the many enzymes that perform these tasks. The specific amino acid can build up in the blood and other organs, including the brain. This amino acid and any of its metabolites can cause serious health problems such as mental retardation, damage to vital organs, seizures or coma. The effects of the disorder will vary and depend on the age at which symptoms occur and the specific amino acid(s) elevated. Treatments vary and may include special dietary intervention, replacement medications, acute illness protocols and metabolic genetic and nutritional monitoring. Disorders include Phenylketonuria, Homocystinuria, Citrullinemia, etc.

### **ORGANIC ACID DISORDERS**

Organic acid disorders are a group of conditions in which there is a problem with breaking down protein and amino acids in foods. This is due to a specific defect in one of the enzymes that breaks down these substances. These organic acids can build up in blood and urine and can lead to problems such as low blood sugar, failure to thrive, developmental delays, infections and in rare occasions coma and death. The effects of the disorder will depend on the age at which symptoms occur. Delay in the recognition and treatment may have serious consequences. Treatment may include special dietary intervention, replacement medications, acute illness protocols and metabolic genetic and nutritional

monitoring. Some disorders are 3-Methylcrotonyl Co-A Carboxylase deficiency, Glutaric Acidemia Type I and II, etc.

## **FATTY ACID OXIDATION DISORDERS**

Fatty Acid Oxidation Disorders are a group of conditions that affect the breakdown of certain fats called fatty acids. A defect in a specific enzyme leads to a build up of fatty acids in the body. When a baby with one of these conditions "fasts" (goes for a long period of time without eating), problems can happen. This occurs because the baby cannot use the energy stored in the fats of the body. This kind of metabolic crisis can sometimes lead to seizures, failure to breathe, cardiac arrest and death. It is extremely important to identify a child with this disease so that crisis can be prevented. Treatment may include avoiding fasting, replacement medications, monitoring the diet for specific metabolic nutritional requirements and blood levels of certain metabolites. An example of Fatty Oxidation disorders is Medium chain Acyl Co-A Dehydrogenase Deficiency (MCAD).

## **CYSTIC FIBROSIS**

Cystic Fibrosis (CF) affects the lungs and digestive system. A defective gene causes the body to produce thick mucus that clogs the lungs causing difficulty in breathing. The mucus also blocks the pancreas and stops enzymes from helping the body break down and absorb food. Symptoms can include salty tasting skin, frequent lung infections, poor growth and frequent greasy stools. Patients are treated in CF centers where there is a team of physicians, nurses, nutritionists, respiratory therapists and social workers. CF is most common in Caucasians, but affects all races and ethnic groups.

**FOR A COMPLETE LISTING OF DISORDERS TESTED SEE THE SECTION ENTITLED REFERENCE RANGES.**

## Newborn Screening (Continued)

### II. Test Methods

- Galactosemia testing is performed by a quantitative enzymatic fluorometric method to detect Uridyl transferase enzyme activity and Total Galactose.
- Biotinidase screening is performed by colorimetric methodology. It is a semi-quantitative analysis for the determination of Biotinidase activity in dried whole blood spots.
- Thyroid testing is performed by a quantitative fluoroimmunoassay (FIA) method which detects the amount of thyroid stimulating hormone (TSH) present.
- Hemoglobin testing is performed by High Performance Liquid Chromatography (HPLC).
- Congenital adrenal hyperplasia employs a quantitative fluoroimmunoassay (FIA) methodology, which detects the amount of  $17\alpha$ -hydroxyprogesterone ( $17\text{-}\alpha\text{OHP}$ ).
- Organic Acid, Fatty Acid and Amino Acid tests are analyzed quantitatively by Tandem Mass Spectrometry. These analytes are detected by their mass to charge ratio.
- Cystic Fibrosis testing is performed by a quantitative fluoroimmunoassay (FIA) method which detects the amount of Immunoreactive Trypsinogen (IRT) present.

## Newborn Screening (Continued)

### III. Specimen Collection

#### A. Collection Form

Use the Newborn Screening Collection Form PH-1582. Forms are available from your local county health department. Health departments can order more forms from the Shipping Department of Laboratory Services (615-262-6391). The expiration date of the filter paper is printed near the bottom right corner of the form. It will have the year-month. Forms are good until the last day of the month printed on the form for the year specified. (Example: EXP. 2016-01 means do not use the form after 01/31/2016.) Blood collected on forms after the expiration date will be reported out as "*Unsatisfactory Filter Paper Expired*" and another specimen will have to be submitted.

#### B. Infants more than 6 months old

The test methods used by the Newborn Screening Laboratory are *not suitable for infants greater than 6 months of age*. If the infant is greater than 6 months of age, contact the Metabolic Center closest to the provider to inquire as to what tests need to be performed and where to send the specimens.

#### C. Procedures When Infants Are Transfused Prior to the Newborn Screening \*

If possible, collect a specimen for the newborn screen **before a transfusion** even if the infant is less than 24 hours of age. The hemoglobin and biotinidase will be accurate and if it is normal, there is no need for follow-up hemoglobin testing.

The TSH, CAH, Total GAL, CF, FA, OA, Biotinidase and AA test methodologies and results (i.e. measurement of the metabolite) can be affected by transfusions. Any infant that was transfused within 72 hours prior to specimen collection with any of the following blood products will need to have their screen repeated:

- Exchange transfusion
- ECMO procedure
- Whole blood
- Plasma

GAL, FA, OA, AA and Hemoglobinopathy results can be affected by red blood cell transfusions. CF results are not affected if the infant has been given packed red blood cells.

Biotinidase, FA, OA, CF and AA results can be affected by platelet transfusions.

\* If an infant has symptoms such as vomiting, diarrhea, dehydration, or jaundice, the newborn screen should be repeated immediately regardless of the number of days that have passed since the transfusion. In addition, the regional metabolic center or endocrinologist (depending on symptoms) should be contacted.

## **Newborn Screening (Continued)**

**Repeat a specimen for TSH, CAH, MCAD, OA, FA, AA, Biotinidase, CF and GAL** as soon as possible after the fourth day post transfusion. Send the specimen to the TDH Newborn Screening Laboratory.

**Repeat Hemoglobinopathy** at three months post transfusion. Send the specimen to Meharry Sickle Cell Center. See Newborn Screening Section XIII for information about submitting specimens to Meharry Sickle Cell Center.

If the infant has been transfused, mark transfusion on the collection form and give the last transfusion date.

### **D. Infants in the NICU and Premies**

Sick or premature newborns should have a specimen collected on or near the seventh day of age regardless of feeding status or before transfusion. Amino Acid results are based on the assumption that the infant has had protein feed. Galactosemia results are based on the assumption the infant is on a lactose feeding. If the infant is sick or not feeding well and the physician feels a test is not accurate due to the infant's feed status, the physician is encouraged to obtain a repeat specimen.

**Newborn Screening (Continued)**

**E. Instructions for Filling out the NBS Form PH-1582 (Rev. 01/13)**

It is important to fill in all of the blank lines on the form completely, legibly and accurately. Use a ballpoint pen to legibly print the information on the form. If critical areas on the form are left blank the Family Health and Wellness Follow-up Section will have to call to obtain the information. This may cause a delay in reporting results.

**FRONT**

**NEWBORN SCREENING** TO AVOID RECOLLECTION - Accurately complete the entire form. All information must be printed.

First Repeat: Prior Unsat Prior <24Hrs or Transf Prior Abnormal Previous TDH#

**HOSPITAL INFORMATION**  
 Hospital of Birth ID Hospital of Collection ID  
 Infant Medical Record No.

**MOTHER'S INFORMATION**  
 Mother's Current Last Name First Age  
 Address  
 City State Zip  
 Phone  
 Mother's Social Security No. County of Residence

**INFANT STATUS AT TIME OF SPECIMEN COLLECTION:** CURRENT WEIGHT: \_\_\_\_\_ Grams  
 TRANSFUSED: ( ) Y ( ) N If yes, Date of Last: \_\_\_\_\_  
 ANTIBIOTICS: ( ) Y ( ) N NICU: ( ) Y ( ) N  
 FEEDING: ( ) 1 Breast ( ) 2 Non-Lactose ( ) 3 TPN/Lipids ( ) 4 Lactose ( ) 5 NPO

**HEARING** PULSE OXIMETRY  
 Date: \_\_\_\_\_/\_\_\_\_\_/\_\_\_\_\_  
 Method: \_\_\_\_\_ ABR \_\_\_\_\_ OAE  
 Right Ear: \_\_\_\_\_ Pass 1 \_\_\_\_\_ Refer 2  
 Left Ear: \_\_\_\_\_ Pass 1 \_\_\_\_\_ Refer 2  
 \_\_\_\_\_ Unable to test 0 \_\_\_\_\_ Still in Hospital 7  
 \_\_\_\_\_ Declined 3 \_\_\_\_\_ Expired 8  
 \_\_\_\_\_ Transferred 6

**SEE BACK OF FORM FOR SCREENING INSTRUCTIONS**  
 Initial O2 Screen Date/Time: \_\_\_\_\_/\_\_\_\_\_/\_\_\_\_\_  
 Did both RH and foot need to be tested? ( ) Y ( ) N  
 Final Result: Passed ( ) Failed ( )  
 Referred to Cardiology: ( ) Y ( ) N  
 If not performed, reason:  
 \_\_\_\_\_ Refused (1) \_\_\_\_\_ Expired (2) \_\_\_\_\_ On O2 (3)

**PRIMARY CARE PROVIDER'S INFORMATION**  
 ( ) - \_\_\_\_\_  
 Name  
 Address  
 City State Zip

TENNESSEE DEPARTMENT OF HEALTH  
 LABORATORY SERVICES  
 830 HART LANE, NASHVILLE, TENNESSEE

SPECIMEN CONTROL NUMBER DATE REC'D/LAB NO. Lab Unsat:  
**E-349570**

DO NOT WRITE IN THIS AREA

2016-01 Form PH 1582 REV. 01/13

**BACK**

**Instructions for Blood Specimen Collection**

- Hold infant's limb in a dependent position to increase blood flow.
- Clean heel thoroughly. Wipe with alcohol and dry before puncturing.
- Puncture heel with sterile lancet to assure free flow of blood. Puncture should not exceed 2.0 mm in depth.
- Wipe away first drop and discard.
- Allow a large drop of blood to form on infant's heel. Apply the back side of the filter paper directly to the drop of blood at the puncture site (NOT the heel). The drop of blood should be large enough to approximately fill one circle and must completely saturate through the paper. It should look the same on both sides of the filter card.
  - DO NOT apply more than one drop of blood per circle.
  - DO NOT apply blood to both front and back of filter paper.
- Apply blood to all circles.
- Allow blood spots to completely dry in a horizontal position at room temperature (see diagram). Do not stack specimens while specimen is exposed. After drying, rewrap the cover sheet to its original position to protect specimen.
- DO NOT place specimens in a sealed plastic bag.
- Mail within 24 hours of collection by USPS priority mail:  
 Newborn Screening  
 Department of Health  
 Laboratory Services  
 P.O. Box 305130  
 Nashville, Tennessee 37230-5130

**Critical Congenital Heart Disease Screening Instructions**

- Perform screen at 24-48 hours of age or shortly before discharge if <24 hours old.
- Screen foot only (either foot) and document date and time in area for initial screen:
  - If 97-100%: Mark PASSED. No further testing needed.
  - If <90%: Mark FAILED.
  - If 90-96%: Add the screening of Right Hand.
- If both RH and Foot need to be screened, note it on the collection form:
  - Mark PASSED if:
    - ≥95% in either extremity with a ≤3% difference between the right hand and foot.
  - Mark FAILED if:
    - <90% in either the RH or the foot at anytime. Babies should have IMMEDIATE CLINICAL ASSESSMENT.
    - <95% in both the RH and the foot or a >3% difference between RH and foot on three measures each separated by one hour.

Example:  
 ♦ If measurements are <95% in both the RH and foot or >3% difference between the two:  
 → Perform a second screen in one hour  
 ♦ If the second measurements are <95% in both the RH and foot or >3% difference between the two:  
 → Perform a third screen in one hour  
 ♦ If the third measurements are <95% in both the RH and foot or >3% difference between the two:  
 → Then mark FAILED

Referred to Cardiology: Only mark Yes if the baby was referred after clinical assessment.

EASTERN LABORATORY SERVICES, PLLC

## Newborn Screening (Continued)

### 1. First \_\_\_\_\_

If this is the "First" specimen ever collected on the infant, place a mark on the "First" line. If the specimen is marked as a "First" all test are automatically performed.

### 2. Repeat: \_\_\_\_\_Prior Unsat \_\_\_\_\_Prior <24 hrs or Transf \_\_\_\_\_Prior Abnormal

If this is a repeat specimen and the provider or the parent was sent a letter indicating that the specimen should be repeated, a reason would be given on the letter.

If the specimen was unsatisfactory for any reason and this is why the repeat is being done place a mark on the "Prior Unsat" line.

If the first specimen was collected before the infant was 24 hours of age and this is why the repeat is being done or if the specimen was collected after the infant was transfused and this is why the repeat is being done, place a mark on the "Prior <24 hrs or Transf" line.

If the specimen had an abnormal test result and this is why the repeat is being done, mark on the "Prior Abnormal" line.

### 3. Previous TDH#

If this is a repeat specimen and the provider or the parent was sent a letter indicating that the specimen should be repeated, write the 11 digit unique TDH number included in the letter in this space. If possible enclose a copy of the letter with the repeat specimen.

### 4. Infant's Last Name:

Legibly print the infant's last name.

### 5. Infant's First Name:

If the infant has a first name, print it legibly here. If the infant does not have a first name at the time of collection and it was a single birth, write "BOY" for the first name if it's a male infant or "GIRL" for the first name if it is a female infant. If there are multiple births also indicate the birth order by using A, B, C, etc. EXAMPLE: GIRL "A", BOY "B", GIRL "C", BOY "D".

### 6. Previous Last Name:

If the infant has had a change in their last name, legibly print their previous last name here. *This is very important!* Without this information we are unable to identify when an infant has had a repeat specimen collected.

### 7. Birth Date:

The day the infant was born. Write the date as MM / DD / YY. The date should be the same day as recorded on the infant's birth certificate.

**8. Birth Time:**

The time the infant was born in military time. (See Chart IV-3 for a MILITARY TIME CONVERSION CHART.)

Example: Write 3:00 AM as 0300 and 3:00 PM as 1500. The use of strict military time will indicate AM or PM. The time should be the same time as recorded on the infant's birth certificate.

**9. Collect Date:**

This is the date the specimen was collected. Write the date as MM / DD / YY.

**10. Collect Time:**

The time the specimen was collected in military time. (See Chart IV-3 for a MILITARY TIME CONVERSION CHART.)

Example: Write 6:00 AM as 0600 and 6:00 PM as 1800. The use of strict military time will indicate AM or PM.

**11. Single Birth:**

If the infant was a single birth, mark  1. Single Birth.

**12. Twin ( )A ( )B**

If the infants were twins mark  2. Twin and mark either ( ) A for the first born or ( ) B for the second born.

**13. Other:**

If the delivery is triplets (or more), mark  3. Other, followed by the number and letter to indicate the birth order.

Example: Triplets would be written as 3A, 3B and 3C. Quadruplets would be written as 4A, 4B, 4C and 4D.

**14. Gender:**

If the infant is a boy mark  1. M. If the infant is a girl mark  2. F.

**15. Race:**

Place a mark next to the category which best reflects the race of the infant  
( ) 1. White, ( ) 2. Black, ( ) 3. Asian, ( ) 4. Am. Ind or ( ) 5. Other.

**16. Ethnicity:**

Place a mark next to the category which best reflects the ethnicity of the infant  
( ) 1. Hispanic or ( ) 2. Non-Hispanic.

**17. Birth Weight:**

Give the weight of the infant at birth. The weight must be recorded in grams. See reference Chart IV-2 Pounds and Ounces to Grams Conversions.

## 18. Gestation Age:

Indicate in weeks the age of infant at time of birth.

## 19. Status of the infant at time of collection:

**a. Current Weight:** \***NOTE**\* Give the weight of the infant at the time the specimen was collected. The current weight of the infant must be recorded accurately so the CAH test results are accurate. The weight must be recorded in grams. See Chart IV - 2 POUNDS AND OUNCES TO GRAMS CONVERSION.

**b. Transfused:** If the infant was not transfused mark (X) No. If the infant was transfused, in utero or after delivery, mark (X) Yes and then enter the **date of last** transfusion (MM / DD / YY) prior to the specimen being collected. The transfusion information must be recorded accurately so that the hemoglobinopathy, galactosemia and other test results are accurate.

**c. Antibiotics:** If the infants has had antibiotics prior to specimen collection mark (X) Yes otherwise mark (X) No.

**d. NICU:** If the infant is in the NICU at the time of collection mark (X) Yes otherwise mark (X) No.

**e. Feeding:** Place a mark next to the method by which the infant is currently being fed ( ) 1. Breast, ( ) 2. Non-Lactose, ( ) 3. TPN/Lipids, ( ) 4. Lactose, ( ) 5. NPO.

## 20. Hospital Information:

### a. Hospital of Birth ID and Hospital Collected ID:

**THE REPORT WILL BE MAILED TO THE HOSPITAL OF COLLECTION.** Enter the seven-digit hospital code that indicates the location of the infant's birth and the collection hospital. The hospital code is available from the hospital of birth. This blank **must be completed** regardless of the provider. Private physicians and county health departments must also give the code for the hospital of birth on **each specimen submitted to the TDH Laboratory**. If the infant has been transferred, the hospital of collection may be different from the hospital of birth. **Make sure you have the correct codes for both hospitals.**

If the infant was not born in a Tennessee hospital, indicate the location by giving the name and location of the hospital of birth.

If the infant was not born in a hospital, enter the two-digit code for the county and record "HOME" in hospital block as shown. See Chart IV-4 for a COUNTY CODE LIST FOR TENNESSEE. (For example Shelby County - Home Birth - |7|9| |H|O|M|E|.)

### b. Medical Record Number:

Give the unique patient number assigned to the infant in the hospital, doctor's office, or local health agency.

## 21. Mother's Information:

**Adoption:** If a newborn has been adopted please write "ADOPTION CASE" on the form and put either the adoptive parents or the adoption agency's information in the spaces

under Mother's Information. If a repeat specimen is required, letters will be sent to the mother listed on the form until the Newborn Screening lab receives a repeat specimen.

**a. Mother's Current Last Name:** Legibly print the mother's legal last name.

**b. First:** Legibly print the mother's legal first name.

**c. Age:** Legibly print the age of the mother at the time of the infant's birth.

**d. Address, City, State, Zip Code:** Legibly print the mother's complete address where she is currently living including the city, state and zip code + 4. If the mother's residence is not in the USA, name the country.

**e. Phone Number:** Legibly print the area code and home phone number where the infant's mother can be contacted. If the mother does not have a phone, give the area code and phone number of a relative or neighbor who could easily contact the mother.

**f. Mother's Social Security No.:** Legibly print the mother's social security number.

**h. County of Residence:** Legibly print the two-digit code that corresponds to the county in which the mother resides. (See Chart IV-4 for the COUNTY CODE LIST FOR TENNESSEE.)

## 22. Hearing:

**Note:** If you are only reporting hearing screens, do not use the newborn screening forms. Contact Family Health and Wellness at 615-532-8462 for instructions regarding the correct method to report and for additional questions regarding the hearing screens.

Hospitals that provide newborn hearing screening utilizing the physiologic methods of ABR and/or OAE are requested to complete the Hearing Screen portion of the form. If your hospital does not provide newborn hearing screening, leave the spaces blank.

**a. Method:** Place a mark next to \_ABR if the infant was tested by the Auditory Brainstem Response (ABR) or Automated Auditory Brainstem Response (AABR) method. Place a mark next to \_OAE if the infant was tested by the Otoacoustic Emissions (OAE) using the Distortion Product Otoacoustic Emissions (DPOAE) or the Transient Evoked Otoacoustic Emissions (TEOAE) method. If both methods were used, mark the method administered prior to discharge and the results.

**b. Right Ear:** Place a mark next to \_Pass 1 if the right ear passed the screen otherwise mark \_Refer 2.

**c. Left Ear:** Place a mark next to \_Pass 1 if the left ear passed the screen otherwise mark \_Refer 2.

**Note:** Pass is the term used for a test that indicates the hearing is within normal limits. Refer is the term used for referral of infants for further evaluation.

**d.** If the hearing test was not performed please indicate the reason by marking one of the following: \_Unable to test 0, \_Declined 3, \_Transferred 6, \_Still in Hospital 7, \_Expired 8

**e. Risk Factors:** Indicate risk factors by marking the appropriate box. See the back of the Hearing screening copy of the Newborn Screening form for a description of Risk Indicators for Hearing Loss.

### 23. Pulse Oximetry:

**a. Initial O2 Screen Date/Time:** Indicate date MM/DD/YY that test was conducted. Indicate the time the test was conducted in military time. (See Page IV - 23 for a MILITARY TIME CONVERSION CHART.)

Example: Write 6:00 AM as 0600 and 6:00 PM as 1800. The use of strict military time will indicate AM or PM.

**b. Did both RH and foot need to be tested?** Answer by marking either ( )Y or ( )N.

**c. Final Result:** Indicate the results of the test by either marking Passed( ) or Failed( ).

**d. Referred to Cardiology:** Indicate by marking either ( )Y or ( )N.

**e. If not performed, reason:** Indicate if the test was not performed by marking \_\_Refused (1), \_\_Expired (2), or \_\_On O2 (3)

### 24. Primary Care Provider's Information

*The report will be mailed to the Physician or Provider listed here. The report will also be mailed to the hospital of collection.*

**a. Phone:** Legibly print the area code and phone number of the physician or health care provider to be contacted if there is an abnormal test result.

**b. Name:** Legibly print the first and last name of the physician or health care provider.

**c. Address, City, State and Zip Code:** Legibly print the complete street address, city, state and zip code + 4 of the physician or health care provider.

### 25. Date Rec'd/Lab No (White area at bottom of form)

#### DO NOT WRITE IN THIS AREA

The laboratory will record the date and assign a specimen number to the specimen when it is received at the laboratory.

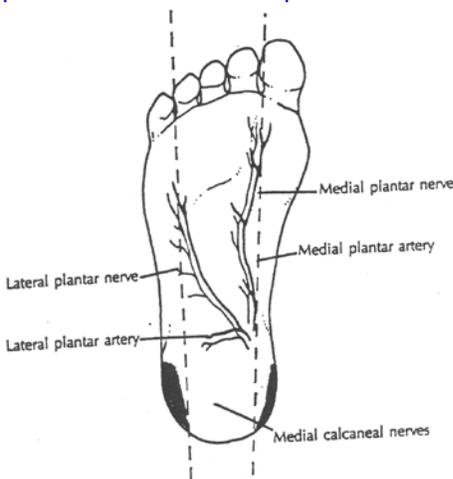
### F. Filter Paper Collection Procedure

#### PRECAUTIONS:

Improper storage of the collection forms can result in the filter paper being altered by the absorption of ambient moisture in the atmosphere, or by compressing the filter paper if they are stacked on top of each other. Store the forms on their side in a cool, clean, dry place. Touching the filter paper, either before or after collection, with gloved or ungloved hands or substances such as powder, hand lotion, feeding formulas, or other materials can alter the filter paper and also the test results. Use of highlighters on the filter paper or wearing strong perfumes may also interfere with testing procedures.

Verify the patient information on the specimen collection form with the infant's identification band and appropriate chart records before sticking the infant. Check the expiration date of the form. If it has expired do not use it. Appropriate biohazard precautions should be used. Wear powder free gloves and change gloves between infants. Never reuse lancets; place them in a puncture proof sharps container after use. Place biohazardous material in appropriate containers.

1. Select the puncture site on the heel. The shaded areas on the diagram indicate the preferred and least hazardous puncture sites. Draw an imaginary line between the fourth and fifth toes that runs parallel to the lateral aspect of the heel or a line running from the great toe that runs parallel to the medial aspect of the heel.



2. Warming the heel with a warm (Temperature not to exceed 42 °C) wet towel or diaper or a commercially available heel warmer for not more that 3 minutes along with positioning the leg lower than the heart may dilate the blood vessels and therefore increase blood flow.
3. Disinfect the skin with 70% isopropyl alcohol and allow it to air dry. Vigorous rubbing may also stimulate blood flow to the area.
4. Puncture the skin with one continuous motion using a sterile automated lancet. On a full term infant the depth of the puncture should not exceed 2.0 mm. On premature or small infants the puncture should be less than 2.0 mm. Lancets with tips longer than 2.5 mm may cause excessive tissue damage, bone damage, or long term problems with walking.
5. Wipe away and discard the first drop of blood since it may be contaminated by alcohol or tissue fluid.
6. Allow the second drop (approximately 75-100  $\mu$ l) to form by the spontaneous free flow of blood.
7. Touch the filter paper to the drop of blood as close to the center of the circle as possible. Allow the blood spot to expand within the circle and soak through to the other side. Turn the card over to make sure that the blood did soak through completely. DO NOT press or touch the filter paper to the puncture site.
  - a. DO NOT APPLY BLOOD TO BOTH SIDES, Apply blood to only one side of the filter paper. It does not matter which side, however, the blood must soak through from one side to the other.

- b.** It is essential that only one drop of blood be used to fill a circle. If the circle cannot be filled with only one drop, go on to the next circle. Do not apply two drops to the same circle.
  - c.** It is permissible for the blood to go outside the circles lines, but do not allow it to overlap on blood in an adjacent circle. Overfilling of the circle can cause supersaturation.
  - d.** Do not use capillary tubes or syringes to fill circles. Such devices may alter the filter paper by scratching it, cause supersaturation, or blood clots.
  - e.** Recollect the sample if tiny blood clots appear on the specimen or if any fluid or substance contaminates the specimen.
- 8.** Once the blood collection is completed, hold the infant's foot above the heart level and press a sterile gauze to the puncture site until the bleeding has stopped. Adhesive bandages are not recommended.
- 9.** Air dry the blood specimen horizontally at room temperature away from heat or direct sunlight for at least 3 hours. Do not allow the blood spots to touch any surface. Do not close the fold-over-paper protective flap for at least 3 hours and the specimen is completely dry.

## Newborn Screening (Continued)

### IV. Shipment of Specimens

After the specimen has had time to dry for at least 3 hours, close the protective paper flap over the top of the blood spots. **DO NOT tape the flap closed or fold the form.** The biohazard label should be placed on the outside of the flap of the form. Place the form in a **paper** envelope labeled "Dried Clinical Specimen". If you are sending more than one form, rotate the forms 180° so that the blood spots are not stacked directly over one another but are alternated. **DO NOT place the forms in any form of a plastic sealed bag.** This includes poly bags, ziplock bags, plasticene envelopes, or plastic shipping bags.

Mail or transport the specimen to the Tennessee Department of Health's Laboratory in Nashville within 24 hours of collection.

**If the specimens are going to be mailed  
United States Postal Service Address:**

Newborn Screening  
Tennessee Department of Health  
Laboratory Services  
P. O. Box 305130  
Nashville, TN 37230-5130

**If the specimens are going to be  
delivered by Courier, FedEx, or UPS:**

Newborn Screening  
Tennessee Department of Health  
Laboratory Services  
630 Hart Lane  
Nashville, TN 37216-2006

The U. S. Postal Service address is what appears on the back of the yellow fold-over flap. If you have your specimens sent by FedEx, UPS or another delivery company you must use the address on the right above.

### V. Reporting Procedure and Interpretation

The results of all specimens are reported to the physician or provider listed on the form.

The laboratory reports all positive, or suspected positive, results generally within 1 to 2 working days after the specimen is received in the laboratory. The NBS Family Health and Wellness follow-up program will notify the provider of abnormal results by email or fax to initiate treatment of the patient, confirmation testing and follow-up of the patient. The written report is mailed usually within 5 to 7 working days after receipt of the specimen.

Written reports of normal specimens are mailed within 5 to 7 working days after receipt in the laboratory.

All unsatisfactory specimens are tested, even though the integrity of the specimen is in question. If a positive, or suspected positive, is found, results are communicated to the provider so that treatment can be initiated. **However, all unsatisfactory specimens are reported as unsatisfactory and must be repeated.**

**Newborn Screening (Continued)**

<b>Reference Ranges</b> Cut-offs points are established through population-based studies. These values may change over time.									
<b>Galactosemia (GAL)</b>									
Total Galactose $\geq$ 15 mg/dL and Galactose Enzyme $\leq$ 40  It is assumed that the infant has had a lactose feeding and has not been transfused unless otherwise indicated on the Newborn Screening Collection Form.									
<b>Congenital Hypothyroidism (TSH)</b>									
<b><u>For infants 1 day through 7 days</u></b> < 33 $\mu$ U/ml serum Within Normal Limits 33 – 55 $\mu$ U/ml serum Borderline > 55 $\mu$ U/ml serum Positive <b><u>For infants 8 days through 6 months of age</u></b> < 13 $\mu$ U/ml serum Within Normal Limits $\geq$ 13 $\mu$ U/ml serum Positive <b><u>For infants &lt;24 hours of age</u></b> < 33 $\mu$ U/ml serum Within Normal Limits but MUST REPEAT FILTER TEST IMMEDIATELY $\geq$ 33 $\mu$ U/ml serum Inconclusive Results MUST REPEAT FILTER TEST IMMEDIATELY									
<b>Hemoglobinopathy</b>									
FA is a normal hemoglobin pattern for a young infant AF is a normal hemoglobin pattern for an old infant The result is reported as Within Normal Limits as long as no abnormal peaks are detected. It is assumed that the infant has not been transfused unless otherwise indicated on the Newborn Screening Collection Form.									
<b>Congenital Adrenal Hyperplasia (CAH)</b>									
<b>Birth Weight</b> <table style="width: 100%; border: none;"> <tr> <td style="width: 50%;"><b>&lt;1250 grams</b></td> <td style="width: 50%; text-align: right;"><b>&lt;97 ng/mL</b></td> </tr> <tr> <td><b>1251-1750 grams</b></td> <td style="text-align: right;"><b>&lt;62 ng/mL</b></td> </tr> <tr> <td><b>1751-2245 grams</b></td> <td style="text-align: right;"><b>&lt;42 ng/mL</b></td> </tr> <tr> <td><b>&gt;2250 grams</b></td> <td style="text-align: right;"><b>&lt;30 ng/mL</b></td> </tr> </table>		<b>&lt;1250 grams</b>	<b>&lt;97 ng/mL</b>	<b>1251-1750 grams</b>	<b>&lt;62 ng/mL</b>	<b>1751-2245 grams</b>	<b>&lt;42 ng/mL</b>	<b>&gt;2250 grams</b>	<b>&lt;30 ng/mL</b>
<b>&lt;1250 grams</b>	<b>&lt;97 ng/mL</b>								
<b>1251-1750 grams</b>	<b>&lt;62 ng/mL</b>								
<b>1751-2245 grams</b>	<b>&lt;42 ng/mL</b>								
<b>&gt;2250 grams</b>	<b>&lt;30 ng/mL</b>								
<b>Biotinidase Deficiency</b>									
$\geq$ 27 MRU Within Normal Limits $\geq$ 13 and <27 MRU Partial Deficiency < 13 MRU Deficient									
<b>Cystic Fibrosis (CF)</b>									
<b>For Infants 1 day through 7 days</b> <70 ng/mL of blood within normal limits <b>For infants 8 days through 6 months of age</b> <60ng/mL of blood within normal limits									

**Newborn Screening (Continued)**

**MS/MS REFERENCE RANGES (UPDATED 8/2013)**

**Amino Acid Disorders**

Metabolites	Normal Values	Disorder(s) Related
Arginine	Arg < 95 µmol/L	Argininemia (Arginase Deficiency)
Citrulline (low)	Cit > 3 µmol/L	Carbamoylphosphate Synthetase Deficiency
Citrulline (high)	Cit < 62 µmol/L	Citrullinemia Type I (Arginosuccinate Synthetase Deficiency) Citrullinemia Type II (Citrin Deficiency) Argininosuccinate Lyase Deficiency (Arginosuccinic Aciduria)
Cit/Arg Ratio	Cit/Arg < 6.00	Citrullinemia Type I (Arginosuccinate Synthetase Deficiency) Citrullinemia Type II (Citrin Deficiency) Argininosuccinate Lyase Deficiency (Arginosuccinic Aciduria)
Argininosuccinic Acid (Asa)	Asa < 0.33 µmol/L	Argininosuccinate Lyase Deficiency (Arginosuccinic Aciduria)
Asa/Arg Ratio	Asa/Arg < 0.13	Argininosuccinate Lyase Deficiency (Arginosuccinic Aciduria)
Glycine	Gly < 915 µmol/L	Nonketotic Hyperglycinemia
Methionine	Met < 61 µmol/L	Homocystinuria or variant forms of Hypermethioninemia
Ornithine	Orn < 377 µmol/L	Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH) Hyperornithinemia with Gyral Atrophy
Orn/Cit Ratio	Orn/Cit < 18.9	Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH) Hyperornithinemia with Gyral Atrophy
Phenylalanine	Phe < 152 µmol/L	Phenylketonuria Hyperphenylalaninemia due to: Phenylalanine Hydroxylase Deficiency GTP Cyclohydrolase I Deficiency Pterin-4-Alpha Carbinolamine Dehydratase Deficiency 6-Pyruboyltetrahydropterin Synthase Deficiency Defects of bipterin co factor biosynthesis Defects of bipterin co factor regeneration
Phe/Tyr Ratio	Phe/Tyr < 2.01	Phenylketonuria Hyperphenylalaninemia due to: Phenylalanine Hydroxylase Deficiency GTP Cyclohydrolase I Deficiency Pterin-4-Alpha Carbinolamine Dehydratase Deficiency 6-Pyruboyltetrahydropterin Synthase Deficiency Defects of bipterin co factor biosynthesis Defects of bipterin co factor regeneration
Tyrosine	Tyr < 330 µmol/L	Transient Tyrosinemia Tyrosinemia Types I, II and III
Valine	Val < 274 µmol/L	Maple Syrup Urine Disease Types IA, IB, II
Val/Phe Ratio	Val/Phe < 4.13	Maple Syrup Urine Disease Types IA, IB, II
Leucine	Leu < 288 µmol/L	Maple Syrup Urine Disease Types IA, IB, II
Leu/Phe Ratio	Leu/Phe < 5.53	Maple Syrup Urine Disease Types IA, IB, II
Succinylacetone	SA < 1.56 µmol/L	Hepatorenal Tyrosinemia Type I

**Newborn Screening (Continued)**

**Organic Acid Disorders**

<b>Metabolites</b>	<b>Normal Values</b>	<b>Disorder(s) Related</b>
C3	C3 < 6.35 $\mu\text{mol/L}$	Propionic Acidemia Methylmalonic Acidemia due to: Methylmalonyl-CoA Mutase Deficiency Deficient Synthesis of 5-Prime Deoxyadenosylcobalamin Defects in the MMAA gene Methylmalonic Acidemia with B12 defect and Homocystinuria Multiple CoA Carboxylase Deficiency
C3-DC + C4-OH	C3-DC + C4-OH < 0.46 $\mu\text{mol/L}$	Malonic Aciduria (MA) 3-Hydroxyacyl CoA Dehydrogenase Deficiency (M/SCHAD)
C4	C4 < 1.33 $\mu\text{mol/L}$	Isobutyryl CoA Dehydrogenase Deficiency (IBCD)
C5	C5 < 0.62 $\mu\text{mol/L}$	Isovaleric Acidemia (IVA) 2 Methylbutyryl CoA Dehydrogenase Deficiency (2MBCD) 2 Methyl 3 Hydroxybutyric Aciduria (2M3HBA)
C5:1	C5:1 < 0.08 $\mu\text{mol/L}$	2 Methyl 3 Hydroxybutyric Aciduria (2M3HBA)
C4-DC + C5-OH	C4-DC + C5-OH < 0.60 $\mu\text{mol/L}$	Multiple CoA Carboxylase Deficiency 2 Methyl 3 Hydroxybutyric Aciduria (2M3HBA) 3 Hydroxy 3 Methylglutaryl CoA Lyase Deficiency (HMG) 3 Methyl Crotonyl CoA Carboxylase Deficiency (3 MCC) 3 Methylglutaconyl CoA Hydratase Deficiency (3MGA) Methylmalonic Acidemia due to: Methylmalonyl-CoA Mutase Deficiency Deficient Synthesis of 5-Prime Deoxyadenosylcobalamin Defects in the MMAA gene Methylmalonic Acidemia with B12 defect and Homocystinuria
C4-DC + C5-OH/C8 Ratio	C4-DC + C5-OH/C8 < 7.49	Multiple CoA Carboxylase Deficiency 2 Methyl 3 Hydroxybutyric Aciduria (2M3HBA) 3 Hydroxy 3 Methylglutaryl CoA Lyase Deficiency (HMG) 3 Methyl Crotonyl CoA Carboxylase Deficiency (3 MCC) 3 Methylglutaconyl CoA Hydratase Deficiency (3MGA) Methylmalonic Acidemia due to: Methylmalonyl-CoA Mutase Deficiency Deficient Synthesis of 5-Prime Deoxyadenosylcobalamin Defects in the MMAA gene Methylmalonic Acidemia with B12 defect and Homocystinuria
C5-DC + C6-OH	C5-DC + C6-OH < 0.47 $\mu\text{mol/L}$	Glutaric Acidemia Type I (GAI) 3-Hydroxyacyl CoA Dehydrogenase Deficiency (M/SCHAD)
C6-DC	C6-DC < 0.28 $\mu\text{mol/L}$	3 Hydroxy 3 Methylglutaryl CoA Lyase Deficiency (HMG)

**Newborn Screening (Continued)**

**Fatty Acid Disorders**

<b>Metabolites</b>	<b>Normal Values</b>	<b>Disorder(s) Related</b>
C0	C0 > 6.0 $\mu\text{mol/L}$	Carnitine Uptake Deficiency (CUD)
C0	C0 < 75 $\mu\text{mol/L}$	Carnitine Palmitoyl Transferase Deficiency Type I (CPT I)
C4	C4 < 1.33 $\mu\text{mol/L}$	Short Chain AcylCoA Dehydrogenase Deficiency (SCAD) Multiple AcylCoA Dehydrogenase Deficiency (MADD or GAI1)
C5	C5 < 0.62 $\mu\text{mol/L}$	Multiple AcylCoA Dehydrogenase Deficiency (MADD or GAI1)
C5:1	C5:1 < 0.08 $\mu\text{mol/L}$	Mitochondrial Acetoacetyl CoA Thiolase (Beta Ketothiolase/SKAT) Deficiency
C4-DC + C5-OH	C4-DC + C5-OH < 0.60 $\mu\text{mol/L}$	Mitochondrial Acetoacetyl CoA Thiolase (Beta Ketothiolase/SKAT) Deficiency
C5-DC + C6-OH	C5-DC + C6-OH < 0.47 $\mu\text{mol/L}$	Multiple AcylCoA Dehydrogenase Deficiency (MADD or GAI1)
C6	C6 < 0.29 $\mu\text{mol/L}$	Medium Chain AcylCoA Dehydrogenase Deficiency (MCAD)
C8	C8 < 0.41 $\mu\text{mol/L}$	Medium Chain AcylCoA Dehydrogenase Deficiency (MCAD)
C8/C10 Ratio	C8/C10 < 2.1	Medium Chain AcylCoA Dehydrogenase Deficiency (MCAD)
C10	C10 < 0.22 $\mu\text{mol/L}$	Multiple AcylCoA Dehydrogenase Deficiency (MADD or GAI1)
C10:1	C10:1 < 0.15 $\mu\text{mol/L}$	Medium Chain AcylCoA Dehydrogenase Deficiency (MCAD)
C10:2	C10:2 < 0.11 $\mu\text{mol/L}$	2,4 Dienyl CoA Reductase Deficiency
C14	C14 < 0.58 $\mu\text{mol/L}$	Very Long Chain AcylCoA Dehydrogenase Deficiency (VLCAD)
C14:1	C14:1 < 0.50 $\mu\text{mol/L}$	Very Long Chain AcylCoA Dehydrogenase Deficiency (VLCAD) Multiple AcylCoA Dehydrogenase Deficiency (MADD or GAI1)
C14-OH	C14-OH < 0.05 $\mu\text{mol/L}$	Long Chain Hydroxyl AcylCoA Dehydrogenase Deficiency (LCHAD)
C16	C16 > 0.43 $\mu\text{mol/L}$	Carnitine Palmitoyl Transferase Deficiency Type I (CPT I)
C16	C16 < 8.55 $\mu\text{mol/L}$	Multiple AcylCoA Dehydrogenase Deficiency (MADD or GAI1) Very Long Chain AcylCoA Dehydrogenase Deficiency (VLCAD) Long Chain Hydroxyl AcylCoA Dehydrogenase Deficiency (LCHAD) Trifunctional Protein Deficiency Carnitine/Acylcarnitine Translocase Deficiency (CACTD) Carnitine Palmitoyl Transferase Deficiency Type II
C16:1	C16:1 < 0.50 $\mu\text{mol/L}$	Carnitine /Acylcarnitine Translocase Deficiency(CACTD) Carnitine Palmitoyl Transferase Deficiency Type II
C16-OH	C16-OH < 0.14 $\mu\text{mol/L}$	Long Chain Hydroxyl AcylCoA Dehydrogenase Deficiency (LCHAD) Trifunctional Protein Deficiency
C18	C18 < 1.55 $\mu\text{mol/L}$	Carnitine /Acylcarnitine Translocase Deficiency(CACTD) Carnitine Palmitoyl Transferase Deficiency Type II Long Chain Hydroxyl AcylCoA Dehydrogenase Deficiency (LCHAD) Trifunctional Protein Deficiency Very Long Chain AcylCoA Dehydrogenase Deficiency (VLCAD)
C18:1	C18:1 < 2.27 $\mu\text{mol/L}$	Carnitine /Acylcarnitine Translocase Deficiency(CACTD) Carnitine Palmitoyl Transferase Deficiency Type II
C18:2	C18:2 < 0.99 $\mu\text{mol/L}$	Carnitine /Acylcarnitine Translocase Deficiency(CACTD) Carnitine Palmitoyl Transferase Deficiency Type II
C18:1-OH	C18:1-OH < 0.15 $\mu\text{mol/L}$	Long Chain Hydroxyl AcylCoA Dehydrogenase Deficiency (LCHAD) Trifunctional Protein Deficiency

## Newborn Screening (Continued)

### VII. Specimen Criteria

Newborn Screening Laboratory personnel closely examine each specimen that arrives in the lab for quality and quantity before testing begins. Two technologists look at all unsatisfactory specimens and then a final check is made by the supervisor. The collection hospital, provider and the infant's mother are notified by mail that a repeat specimen needs to be obtained. Unsatisfactory reports are usually mailed within 2 to 4 working days after the specimen is received. The specimens have to be recollected and sent to the laboratory which can be a costly delay if the infant is diagnosed with a genetic disorder. A satisfactory blood drop is one single drop of whole blood applied evenly and allowed to soak through the filter card and can be seen clearly with no white showing through on the other side. The spots must be large enough to allow NINE 1/8-inch discs to be punched out with no white areas showing.

See Chart IV - 1 COMMON CAUSES OF UNSATISFACTORY NEWBORN SCREENING SPECIMENS for unsatisfactory specimen conditions and their possible causes.

**Chart IV - 1**  
**Common Causes of Unsatisfactory Newborn Screening Specimens**

Unsatisfactory Specimen Conditions	POSSIBLE CAUSE
Nonuniform	Applying many small drops of blood to each circle. Applying blood with any type of capillary tube. Touching the blood drops when they are wet. Uneven soaking through the filter card is caused by exposure to moisture, glove powder touching the filter card area before collection or the use of hand creams or lotions. Contaminated surfaces with any of the above contaminants may also cause non-uniform absorption of the blood.
Filter Paper Expired	Specimen was collected on filter paper that was expired.
> 6 months	The infant was greater than 6 months of age at the time the specimen was collected.
< 24 hours	The specimen was collected before the infant was 24 hours of age.
> 10 days	The specimen was received in the laboratory greater than 10 days after the date of collection. This may be a delay in the U.S. Postal service or a delay in the hospital mail service. Mail specimens within 24 hours of collection.
Contaminated	Specimen was contaminated in some way with something like alcohol, water, formula, urine, or hand lotion for example.
Inaccurate Information	Information on the form was inaccurate or incorrect. This most often occurs when the date or time of collection is written on the form as occurring before the date and time of birth.
Incomplete Information	All blanks on the form were not filled out completely.
Quantity Not Sufficient (QNS)	The drops of blood are too small. This can be caused by improper use of the lancet or dropping blood from a capillary tube device.

**Newborn Screening (Continued)**

**Chart IV - 1  
 Common Causes of Unsatisfactory Newborn Screening Specimens**

Unsatisfactory Specimen Conditions	POSSIBLE CAUSE
Supersaturated	The drops of blood are too large. The drops of blood overlap or touch one another. The filter card is pressed against the puncture site. The blood is dropped in very large drops from a capillary tube.
Cells & Serum Separated	The usual cause is squeezing the heel during the specimen collection. It can also be caused by waiting too long for the drop of blood to form or by clotted blood. When applying blood with a capillary tube device, if the blood is not well mixed it may appear on the filter card as clotted blood.
Clotted Specimen	Clotted specimens are due to improper puncture, application with a capillary tube device and waiting too long for a drop of blood to form allowing the blood to clot.
Altered Card	Use of a capillary tube or syringe to apply the blood can scratch the filter card when wet, or rubbing the spot when it is still wet. It can also be caused by pressing the heel to the filter card during the collection process.
Both Sides	This is from applying blood to both sides of the filter card. It is easily recognized in the lab by holding the specimen up to a light and looking at both sides for shadows.
No Blood	The form was received but there was no blood collected on the filter paper.
Heated	Specimen appears much darker than usual and appears to have been heated. Caused by too long in transit especially during the summer. Heat and humidity can affect test results. Also caused by heating a specimen to dry it.
QNSCOM	Insufficient blood to complete testing. Quantity not sufficient for test completion.
Poly Bag	Specimen received in a sealed poly bag, plastic zip lock bag, plasticene envelope, or plastic shipping bag
Detached	Blood spot filter paper detached from information portion of the card.
Accident	Laboratory Accident

## Newborn Screening (Continued)

### VIII. Confirmation Procedures

The laboratory reports a presumptive positive result to Family Health and Wellness (FHW) as soon as it is determined, generally within 24 to 48 hours after the specimen is received. FHW notifies the provider listed on the form by telephone and fax to initiate confirmatory testing, follow-up and treatment of the infant. FHW also notifies the appropriate endocrinologist and Genetic Center or Sickle Cell Center. As soon as abnormal results are determined, FHW faxes or mails, by certified letter, presumptive positive results to the provider. Final results are mailed when all other test are completed, generally within 5 to 7 days after the specimen is received.

**Galactosemia Confirmation:** Infants who require confirmatory diagnostic testing for galactosemia should be referred immediately to the nearest genetic center for instructions regarding blood specimen collection and mailing. The FHW Follow-up Program will provide information about the nearest genetic center.

Infants with a borderline result for galactosemia are required to have a repeat filter-paper blood specimen sent as soon as possible to the TDH Laboratory.

Infants who received a transfusion before the newborn screening test need to have a repeat filter-paper blood specimen collected ten days following the last date of transfusion and within three days of lactose feedings. Repeat galactosemia screening is done again three months after the transfusion.

**Hemoglobinopathy Confirmation:** All infants either transfused or whose initial screen identified an abnormal hemoglobin trait or disease must have a hemoglobin confirmation performed by the Meharry Sickle Cell Center. All supplies, including mailers, collection devices and forms can be ordered from:

Comprehensive Sickle Cell Center  
Meharry Medical College  
Nashville, TN 37208

Telephone: (615) 327-6763

**Congenital Hypothyroidism Confirmation:** FHW will notify the provider of infants who require confirmatory diagnostic thyroid testing. FHW recommends serum thyroid testing for confirmatory testing. This may be handled by the primary care physician or an endocrinologist.

**Congenital Adrenal Hyperplasia (CAH) Confirmation:** Infants who require confirmatory diagnostic testing for CAH should be referred immediately to the nearest pediatric endocrinologist for instructions regarding blood specimen collection and follow-up.

**Biotinidase Deficiency Confirmation:** Those infants who have a deficient Biotinidase level on newborn screening require confirmatory testing for Biotinidase. These infants should be referred immediately to the nearest genetic center as recommended by the Family Health and Wellness (FHW) Follow-up Program. The genetic center will provide information regarding follow-up procedures for blood collection and mailing.

Infants who have a partial deficiency Biotinidase level are required to have follow-up. A repeat filter-paper blood specimen should be submitted to the TDH Laboratory.

## **Newborn Screening (Continued)**

**Amino Acid Disorder Confirmation:** Infants who have an elevated amino acid level(s) are required to have follow-up. The genetic center will provide information regarding follow-up procedures for blood collection and mailing.

**Organic Acid Disease Disorder Confirmation:** Infants who have elevated organic acid level(s) are required to have follow-up. The genetic center will provide information regarding follow-up procedures for blood collection and mailing.

**Fatty Acid Oxidation Disorders Confirmation:** Infants who have an elevated Fatty acid level(s) are required to have follow-up. The genetic center will provide information regarding follow-up procedures for blood collection and mailing.

**Cystic Fibrosis Confirmation:** Infants <8 days of age above the cutoff are rescreened at 2 weeks of age. The genetic center will follow-up on repeat screens. The second screen on infants collected after 14 days of age will be referred for follow-up testing if the second sample is elevated. Infants 8 days of age or greater that are elevated above the cutoff are referred to genetic centers for follow-up. Genetic centers will refer patients to Cystic Fibrosis Foundation Centers.

## **Newborn Screening (Continued)**

### **IX. Material for Parents**

The state provides pamphlets for parents to educate them about the Newborn Screening Test. These pamphlets entitled, *Your Baby and Newborn Screening* can be obtained from Family Health and Wellness (FHW) by calling (615) 532-8462 or (855) 202-1357. The pamphlet answers questions most often asked and explains briefly the disorders Tennessee screens and how the testing is done.

### **X. Infant Discharged before Specimen Was Obtained**

When the nursery or lab forgets to get a specimen on an infant before he/she leaves, we ask that you still send in a newborn screening collection form completely filled in and write, "LEFT BEFORE OBTAINED" on the form. You are responsible for notifying the parent and the physician of the need for the test. Newborn Screening will also contact the physician and make sure the infant is screened. When collection forms for these infants are not sent in, we are unaware that the infant exists.

### **XI. Refusal to Have Newborn Screening Performed**

When a parent refuses the newborn screening test, we ask that you do send in a newborn screening collection form completely filled in and write "REFUSED" on the form and we will follow up. If a parent refuses the test due to religious reasons, please fill out the NEWBORN SCREENING REFUSAL FORM (See Page IV - 30), have it notarized and fax it to Family Health and Wellness at (615) 532-8555.

### **XII. Death Notice**

If you are aware of an infant that has expired, please fax us the child's name, date of birth, mother's name and the date the infant expired. If a specimen needs to be repeated we will continue to send letters to the mother until we receive a specimen. In this case, we do NOT want to notify the parent. Family Health and Wellness' fax number is (615) 532-8555.

### **XIII. Specimen Collection for the Hemoglobin Confirmation Procedure from Meharry Sickle Cell Center**

Proper collection and handling of specimens are essential when sending samples to the Meharry Sickle Cell Center. The following steps must be adhered to so specimens are not damaged in transit and comply with Occupational Safety and Health Administration (OSHA) standards and postal regulations. If these directions are not followed, your samples may not reach Meharry. Your cooperation in this matter is appreciated.

**Supplies Needed:** Request form (Adult/Children), Microvette Tube EDTA, Plastic Biohazard Bags, Styrofoam Mailing Boxes and Packing Tape.

Each Microvette tube contains enough EDTA to anticoagulant up to 200 µl of blood.

**Note special instructions in tube package.**

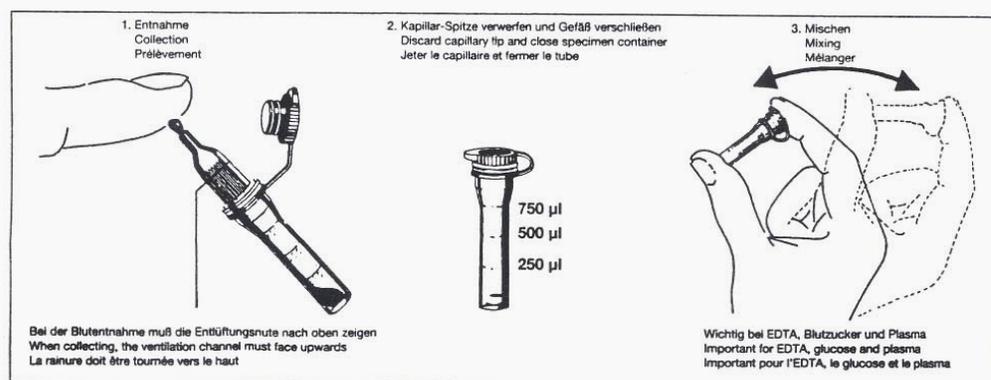
## Newborn Screening (Continued)

1. Label the Microvette tube with the patient's name (label not included).
2. Perform a heel stick using good laboratory practice and following the instructions of the lancet device manufacturer.
3. Wipe off the first drop of blood. Then touch the capillary tip of the Microvette Tube to the droplet of blood as it appears at the incision. Excessive pressure or squeezing on puncture area must be avoided. This could lead to premature coagulation in the capillary.
4. After the blood has been taken, the capillary unit is removed and properly discarded and the sample container is closed with the attached stopper.
5. Properly mix the blood with the EDTA. Hold the stoppered collection Microvette Tube between thumb and index finger and invert several times.
6. Complete the laboratory request form with all information. Include the address to send results. Fold the laboratory request form and place it in the unsealed pocket of a biohazard bag.
7. Place the sealed Microvette Tube in the ziplock pocket of the biohazard bag and place into a Styrofoam mailing box.
8. Place the Styrofoam box in the mailing sleeve and seal both ends. Please put the return address on the mailing box.
9. Mail appropriately packaged sample(s) to :

ATTN: Laboratory Supervisor  
Comprehensive Sickle Cell Center  
Meharry Medical College  
1005 D. B. Todd Boulevard  
Nashville, Tennessee 37208

Phone (615) 327-6763 Fax (615) 327-6008

## Microvette® CB 1000



**CHART IV - 2**  
**Pounds & Ounces to Grams Conversion**

**1 Kilo = 1000 Grams**

Example: To obtain grams equivalent to 5 pounds, 8 ounces, read "5" on top scale. "8" on side scale; equivalent is 2495 grams.

Pounds

oz	0	1	2	3	4	5	6	7	8	9	10	11	12	13	14
0	0	454	907	1361	1814	2268	2722	3175	3629	4082	4536	4990	5443	5897	6350
1	28	482	936	1389	1843	2296	2750	3203	3657	4111	4564	5018	5471	5925	6379
2	57	510	963	1417	1871	2325	2778	3232	3686	4139	4593	5046	5500	5953	6407
3	85	539	992	1446	1899	2353	2807	3260	3714	4167	4621	5075	5528	5982	6435
4	113	567	1021	1474	1928	2381	2835	3289	3742	4196	4649	5103	5557	6010	6464
5	142	595	1049	1503	1956	2410	2863	3317	3770	4224	4678	5131	5585	6038	6495
6	170	624	1077	1531	1984	2438	2892	3345	3799	4252	4706	5160	5613	6067	6520
7	198	652	1106	1559	2013	2466	2920	3374	3827	4281	4734	5188	5642	6095	6549
8	227	680	1134	1588	2041	2495	2948	3402	3856	4309	4763	5216	5670	6123	6577
9	255	709	1162	1616	2070	2523	2977	3430	3884	4337	4791	5245	5698	6152	6605
10	383	737	1191	1644	2098	2551	3005	3459	3912	4366	4819	5273	5727	6180	6634
11	312	765	1219	1673	2126	2580	3033	3487	3941	4394	4848	5301	5755	6209	6662
12	340	793	1247	1701	2155	2608	3062	3515	3969	4423	4876	5330	5783	6237	6690
13	369	822	1276	1729	2183	2637	3090	3544	3997	4451	4904	5358	5812	6265	6719
14	397	850	1304	1758	2211	2665	3118	3572	4026	4479	4933	5386	5840	6294	6747
15	425	879	1332	1786	2240	2693	3147	3600	4054	4508	4961	5415	5868	6322	6776

**Note:** 1 pound = 453.59237 grams  
 1 ounce = 28.349523 grams  
 1000 grams = 1 kilogram  
 Gram equivalents have been rounded to whole number by adding one when the first decimal place is 5 or greater.

## Newborn Screening (Continued)

### Chart IV – 3

#### Military Time Conversion Chart

CIVILIAN TIME	MILITARY TIME
1:00 AM	0100
2:00 AM	0200
3:00 AM	0300
4:00 AM	0400
5:00 AM	0500
6:00 AM	0600
7:00 AM	0700
8:00 AM	0800
9:00 AM	0900
10:00 AM	1000
11:00 AM	1100
12:00 a.m. noon	1200
1:00 PM	1300
2:00 PM	1400
3:00 PM	1500
4:00 PM	1600
5:00 PM	1700
6:00 PM	1800
7:00 PM	1900
8:00 PM	2000
9:00 PM	2100
10:00 PM	2200
11:00 PM	2300
12:00 p.m. midnight	2400

An infant born at 12:05 AM or 5 minutes after midnight would be written as 0005 military time.

**Newborn Screening (Continued)**

**Chart IV – 4**

**TENNESSEE COUNTY CODE LIST**

COUNTY CODE	COUNTY NAME	COUNTY CODE	COUNTY NAME
01	ANDERSON	49	LAUDERDALE
02	BEDFORD	50	LAWRENCE
03	BENTON	51	LEWIS
04	BLEDSON	52	LINCOLN
05	BLOUNT	53	LOUDON
06	BRADLEY	54	MCMINN
07	CAMPBELL	55	MCNAIRY
08	CANNON	56	MACON
09	CARROLL	57	MADISON
10	CARTER	58	MARION
11	CHEATHAM	59	MARSHALL
12	CHESTER	60	MAURY
13	CLAIBORNE	61	MEIGS
14	CLAY	62	MONROE
15	COCKE	63	MONTGOMERY
16	COFFEE	64	MOORE
17	CROCKETT	65	MORGAN
18	CUMBERLAND	66	OBION
19	DAVIDSON	67	OVERTON
20	DECATUR	68	PERRY
21	DEKALB	69	PICKETT
22	DICKSON	70	POLK
23	DYER	71	PUTNAM
24	FAYETTE	72	RHEA
25	FENTRESS	73	ROAN
26	FRANKLIN	74	ROBERTSON
27	GIBSON	75	RUTHERFORD
28	GILES	76	SCOTT
29	GRAINGER	77	SEQUATCHIE
30	GREENE	78	SEVIER
31	GRUNDY	79	SHELBY
32	HAMBLEN	80	SMITH
33	HAMILTON	81	STEWART
34	HANCOCK	82	SULLIVAN
35	HARDEMAN	83	SUMNER
36	HARDIN	84	TIPTON
37	HAWKINS	85	TROUSDALE
38	HAYWOOD	86	UNICOI
39	HENDERSON	87	UNION
40	HENRY	88	VAN BUREN
41	HICKMAN	89	WARREN
42	HOUSTON	90	WASHINGTON
43	HUMPHREYS	91	WAYNE
44	JACKSON	92	WEAKLY
45	JEFFERSON	93	WHITE
46	JOHNSON	94	WILLIAMSON
47	KNOX	95	WILSON
48	LAKE	96	OUT OF STATE

**Newborn Screening (Continued)**

**Patient's Name** \_\_\_\_\_  
**Mother's Name** \_\_\_\_\_  
**Date of Birth** \_\_\_\_\_  
**Hospital of Birth** \_\_\_\_\_

**NEWBORN SCREENING REFUSAL**

I, \_\_\_\_\_ have been informed of the need for newborn screening which includes testing for phenylketonuria (PKU), congenital hypothyroidism, congenital adrenal hyperplasia, galactosemia, hemoglobinopathies and other metabolic/genetic defects that would result in mental retardation or physical dysfunction as determined by the health department.

I have been informed that state law requires this testing and that violation of the law is a misdemeanor.

Nonetheless, I refuse these tests at this time for my newborn baby \_\_\_\_\_ because such test conflicts with my religious tenets and practices.

Under penalty of perjury, I affirm such refusal because of a conflict with my religious tenets and practices.

IN WITNESS WHEREOF, the undersigned has signed this form this \_\_\_\_\_ day of \_\_\_\_\_, 2\_\_\_\_.

\_\_\_\_\_  
Signature

STATE OF TENNESSEE

COUNTY OF \_\_\_\_\_

On this \_\_\_\_\_ day of \_\_\_\_\_, 2\_\_\_\_, before me personally appeared \_\_\_\_\_, to me known (or proved to me on the basis of satisfactory evidence) to be the person described in and who executed the foregoing instrument and acknowledged that she executed the same as her own free act and deed.

\_\_\_\_\_  
Notary Public

My Commission Expires:

\_\_\_\_\_  
REFUSAL

### REQUEST FOR RELEASE OF RECORDS

Baby's Last Name \_\_\_\_\_

Baby's First Name \_\_\_\_\_

Date of Birth \_\_\_\_\_

Mother's Last Name \_\_\_\_\_

Mother's First Name \_\_\_\_\_

Mother's Social Security Number \_\_\_\_\_

Hospital of Birth or Collection \_\_\_\_\_

Any other previous last names used by the mother or the baby:

\_\_\_\_\_

Name of Contact person in provider's office: \_\_\_\_\_

Direct Phone number of Contact Person: \_(\_\_\_\_\_)\_\_\_\_\_

In the event we have trouble locating the baby listed above we will attempt to contact the provider for additional information.

I, \_\_\_\_\_ the legal parent or guardian of the baby listed above do hereby give consent for the State of Tennessee Newborn Screening Laboratory to release results to the Physician or Group Listed here \_\_\_\_\_.

\_\_\_\_\_  
Parent's Signature

\_\_\_\_\_  
Witness's Signature