Mississippi Newborn Screening

Guide for Healthcare Providers

Mississippi State Department of Health
Office of Child & Adolescent Health
Bureau of Genetic Services

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INTRODUCTION

The purpose of this newborn screening guide is to provide information and assistance to healthcare professionals participating in the newborn screening process in Mississippi. We hope that the information in this guide is helpful. Contact the Newborn Screening Program at 601-576-7619 with questions or comments regarding newborn screening.

WHAT IS NEWBORN SCREENING?

Newborn Screening is a public health service completed prior to a newborn being discharged from the delivering facility to assist in identification of certain serious or life-threatening conditions. The screening provides early detection of numerous disorders so that timely treatment can be initiated and long-term sequelae minimized. Long-term sequelae may include organ damage, developmental delay, or death if left undiagnosed and untreated.

WHO SHOULD BE SCREENED?

Sections 41-21-201 and 41-21-203 of the Mississippi Code of 1972 require all newborns born in Mississippi to be screened prior to discharge from the hospital regardless of the age of the newborn or the feeding status. Parents of newborns who object on grounds that the screening conflicts with their religious practices are exempt from screening, but they are required to sign a refusal form stating their understanding of this process.

NEWBORN SCREENING PANEL

The Mississippi State Department of Health (MSDH) contracts with an outside laboratory to perform newborn screening blood spot testing and, when indicated, second tier DNA testing for some disorders. Evaluation and confirmatory testing is performed at a tertiary care center by medical subspecialists. Page 2 lists the diseases/disorders screened for in Mississippi by the blood spot test.

POINT-OF-SERVICE SCREENING

Point-of-service tests for newborns include early hearing and critical congenital heart disease (CCHD) screening. These screens do not involve dried blood spots and are non-invasive tests to screen for hearing loss and certain cardiac defects that may require further medical intervention.

MISSISSIPPI GENETIC NEWBORN SCREENING PANEL

Core Condition	Secondary Condition
Propionic acidemia	Methylmalonic acidemia with homocystinuria
Methylmalonic acidemia (methylmalonyl-CoA mutase)	Malonic acidemia
Methylmalonic acidemia (cobalamin disorders)	Isobutyrylglycinuria
Isovaleric acidemia	2-Methylbutyrylglycinuria
3-Methylcrotonyl-CoA carboxylase deficiency	3-Methylglutaconic aciduria
3-Hydroxy-3-methyglutaric aciduria	2-Methyl-3-hydroxybutyric aciduria
Holocarboxylase synthase deficiency	Short-chain acyl-CoA dehydrogenase deficiency
ß-Ketothiolase deficiency	Medium/short-chain L-3-hydroxyacl-CoA dehydrogenase deficiency
Glutaric acidemia type I	Glutaric acidemia type II
Carnitine uptake defect/carnitine transport defect	Medium-chain ketoacyl-CoA thiolase deficiency
Medium-chain acyl-CoA dehydrogenase deficiency	2,4 Dienoyl-CoA reductase deficiency
Very long-chain acyl-CoA dehydrogenase deficiency	Carnitine palmitoyltransferase type I deficiency
Long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency	Carnitine palmitoyltransferase type II deficiency
Trifunctional protein deficiency	Carnitine acylcarnitine translocase deficiency
Argininosuccinic aciduria	Argininemia
Citrullinemia, type I	Citrullinemia, type II
Maple syrup urine disease	Hypermethioninemia
Homocystinuria	Benign hyperphenylalaninemia
Classic phenylketonuria	Biopterin defect in cofactor biosynthesis
Tyrosinemia, type I	Biopterin defect in cofactor regeneration
Primary congenital hypothyroidism	Tyrosinemia, type II
Congenital adrenal hyperplasia	Tyrosinemia, type III
S,S disease (Sickle cell anemia)	Various other hemoglobinopathies
S, βeta-thalassemia	Galactoepimerase deficiency
S,C disease	Galactokinase deficiency
Biotinidase deficiency	T-cell related lymphocyte deficiencies
Cystic fibrosis	
Classic galactosemia	
Severe Combined Immunodeficiences	

TIPS FOR COLLECTION

All birthing hospitals are required to collect a newborn screening specimen (blood spot test) on each newborn before discharge from the hospital or transfer to a facility for higher level care, regardless of age or the feeding status. In order to expedite screen results, always follow the steps below:

Scheduling:

- The ideal time for specimen collection is 24-48 hours after birth.
- If discharged prior to 24 hours of age, collect the specimen as close to discharge as possible, and collect a repeat specimen at approximately 48 hours of age.
- Critically ill infants should be screened by 24-48 hours of age.

Technique:

- Fill out the specimen card completely.
- Collect the specimen correctly (see Appendix A).
- Fill each of the circles on the filter paper with a single, free-flowing drop of blood.
- Do not layer successive drops of blood.
- Apply the blood to only one side of the filter paper.
- Make sure the blood has saturated through to the back of the paper.

Handling:

- Air dry on a flat surface for 3-4 hours.
- Do not heat, stack, or allow blood spots on the filter paper to touch other surfaces during the drying process.
- Air-dried specimens should be refrigerated until shipped.
- Send specimens daily, via the overnight courier service provided.

The Screening Laboratory has arranged courier services with UPS.

Note: The Complete View Solution (ICVS) is a secure Web-based shipping application that has been customized to accommodate the facility's unique business requirements. Please see UPS - ICVS Training User Guide for instructions.

Mississippi law requires all newborns born in Mississippi to be screened prior to discharge from the hospital regardless of the age of the newborn or the feeding status. Parents of newborns who object on the grounds that the screening conflicts with their religious practices are exempt from screening but are required to sign a refusal form stating their understanding of this process (see Appendix B). Send a copy of this form to MSDH Newborn Screening Program.

INSTRUCTIONS FOR DRIED BLOOD SPOT CARD (DBS) COMPLETION

Results may be delayed for screening specimens submitted with incomplete information. ALWAYS check the specimen card expiration date located on the far left side of the filter paper, next to the hour glass, noted by the year and date. Expired cards are not acceptable. All information must be legible.

TOP LINE

First Specimen – If it is the first newborn screening specimen (dried blood spot) collected on the infant, place an **"X"** in the blank provided.

Home Birth – If the newborn was born at home, place an **"X"** in the blank provided.

Repeat Specimen – If the test is a repeat newborn screening specimen collected on the infant, place an **"X"** in the blank provided.

Repeat Specimen Reason – Put an **"X"** by the appropriate reason for a repeat Specimen; < 24 hr, Unsatisfactory, Abnormal, Transfused, Inconclusive.

REQUIRED INFANT INFORMATION

Infant's Last/First Name – Write infant's name in order as shown: Last name, first name, previous last name. Ensure the name is spelled correctly. If the infant's first name is not available, put last name, followed by Boy/Girl. (Example: Smith, Baby Boy).

Previous Last Name – If the infant's last name is different from the name given at birth, indicate the previous/original name in the blank provided. (Smith, John; Brown, Boy).

Birth Date – Write the date of birth using numbers only in the blank provided. (Example: December 25, 2002, will appear as 12-25-02).

Time of Birth – Write the time of birth using **Military Time Only** in the blank provided. (Example: 2:30 p.m. will appear as 1430).

Date Collected – Write the date the specimen was collected using numbers only in the blank provided. (Example: December 2, 2003, will appear as 12-02-03).

Time Collected – Write the time the specimen was collected using **Military Time Only** in the blank provided. (Example: 8:00 p.m. will appear as 2000).

Birth – Write an **"X"** in the blank provided. (EXAMPLE: in the case of twins/triplets, write an **"X"** to indicate the birth order of the infant to indicate A or B). For triplets or more use #3 blank and indicate C, D, or E as necessary.

Hospital of Birth Code or Health Department Code – Write the hospital code/Health Department code in the appropriate blank provided. **Note:** If the infant is born in the same hospital in which

the specimen is collected, then both the hospital of birth code and hospital collected code will be the same. If the infant is born in one hospital, but transferred to another hospital prior to the specimen being collected, the hospital collection code will be different from that of the hospital of birth. **When the specimen is collected or repeated by the Health Department, the Health Department county code is entered into the Health Department collected blank.**

Medical Record Number – Write the infant's medical record number in the blank provided.

Transferred – If the infant has been transferred to another facility, check "yes" and write the name of the facility in the space provided.

Physician's Name – Write the name and contact information of the healthcare professional who will be providing local medical care for the infant in the blank provided. In the event of a homebirth, **provide the name of the midwife or person attending the homebirth, with contact information.**

Additional Information – Use these lines to provide the following information:

- Collection of a **Pre transfusion** specimen
- Infant is **Adopted (**give the name and address of the adoption agency, attorney or physician handling the adoption.)
- Infant discharged/ expired from the hospital prior to newborn screening collection**.**
- **Incarceration** – if mother will return to an incarceration facility, write INMATE and to whom the infant has been discharged and the contact information.

Submitter Name/Address – Write the name and address of the hospital/Health Department (submitter) in the blank provided.

Specimen Collected By – Write initials of person collecting specimen in the blank provided.

Sex – Write an **"X"** in the appropriate blank provided.

Race – Write an **"X"** in the appropriate blank provided.

Ethnicity – Write an **"X"** in the appropriate blank provided.

Transfused – **Collect specimen PRIOR to transfusion** if at all possible. Provide the **date and time of the last transfusion for each specimen collected** in the space provided (Example: December 15, 2002, at 10:00 a.m. will appear as 12-15-02/1000.)

Note:

- **Ideal collection time: 24-48 hours of age**
- **Transfusion required > 24 hours of age: Collect specimen prior to transfusion.**
- **Transfusion required < 24 hours of age: Collect specimen prior to transfusion and 2-4 days after the transfusion.**
- **Transfusion required but no specimen collected prior to transfusion: Collect specimen 2-4 days, 2-4 weeks, and > 90 days after transfusion**

Note: Anticoagulants should not be used in the collection of newborn screening specimens. In cases where it is unavoidable, only heparin should be used and its presence in the sample should be noted on the filter paper.

Gestation/Infant's Age – Write the gestational age of the infant at birth and the age of the infant at the time of collection in the blank provided.

Birth Weight – Write the infant's weight in grams, **At the Time of Birth** in the blank provided. If specimen is collected > 14 days of age, write current weight in grams.

Feeding – Write an **"X"** in the appropriate blank provided, indicating the infant's feeding status at the time of collection. More than one blank may be marked if appropriate.

Meconium Ileus – Check the blank, if appropriate, to indicate the presence of a meconium ileus.

MOTHER'S INFORMATION

Mother's Current Last/First Name – Write mother's full name, as specified at time of delivery in the blank provided (Example: Smith, Caroline). Enter the mother's maiden name in the space provided. **(Note: If the infant is ADOPTED, do not give the birth mother's information. In the case of ADOPTION, this area should reflect the name of the agency, physician, or attorney handling the adoption.)**

Mother's Date of Birth – Write mother's date of birth using numbers only in the blank provided. (Example: December 25, 1983, will appear as 12-25-83).

Address/Phone Number – Write the physical street address **(Do Not Give P.O. Box),** and/or apartment number, as well as the city, state, and zip code in the blanks provided. Write the telephone number or emergency contact number where a voicemail/message can be accepted, in the blanks provided. **(Note: In the case of ADOPTION, give the agency, physician or attorney name, address and phone number handling the adoption in the blanks provided.)**

Medicaid Number/Mother's Social Security Number – Write mother's Medicaid number, if it applies, and Social Security number in the blanks provided.

County of Residence – Write the county two - digit code where the infant resides in the space provided (Example Hinds County/25).

Note: In the event an infant is a "Drop Off Baby", assign the baby a name. The date the baby was dropped at the hospital should be used as the date of birth, and the approximate time the baby was left should be used as the time of birth, unless other information is known to the hospital. The hospital code should be entered in the space provided for the hospital of birth and hospital collected. In the box provided for Physician's Name, note the emergency medical services provider. This will allow for adequate follow-up/tracking in the event of an abnormal or positive newborn screen.

CRITICAL CONGENITAL HEART DISEASE (CCHD)

In the spaces provided, write the date/time the initial pulse ox screening was performed. If pulse ox was not performed, indicate the reason why. In the appropriate space, indicate the pulse oximetry screening outcome "passed" or a "failed" screen. Indicate if an echocardiogram was performed. DO NOT delay sending the dried blood spot specimen to the screening lab to provide CCHD screening data. Complete the "Delayed Screening Form" and mail or FAX the delayed CCHD screening data to the newborn screening program. (See Delayed CCHD screening form APPENDIX G)

HEARING SCREENING

This section of the newborn screening form should be completed by the hospital of birth prior to discharge. Check the appropriate test performed, i.e., ABR/OAE, and the appropriate result for each ear, i.e., pass/refer. If the hearing screening is not performed, leave this section blank. DO NOT delay sending the dried blood spot specimen to the screening lab to provide hearing screening data.

OFFICE MECHANICS/FILING/RETENTION:

The yellow NCR copy of the dried blood spot card should be kept by the submitting hospital of birth/county Health Department to ensure the results are received by the submitting hospital or county Health Department. Once the newborn screen result is received, the yellow copy can be destroyed. The electronic copy/hard copy of the newborn screening result should be filed in the infant's electronic medical record/paper medical record and retained according to hospital/county Health Department policy.

SPECIMEN COLLECTION

SUPPLIES FOR SPECIMEN COLLECTION

Dried Blood Spot Card (DBS card)
Soap and water
Cotton balls soaked with 70% isopropyl alcohol (also the alcohol prep, which is likely used in most facilities)
Dry cotton balls
Sterile gauze pad
Heel Warmer (if available);
Lancet

SPECIMEN COLLECTION PROCEDURE

1. Recommended site: The least hazardous sites for heel puncture are medial to a line drawn posteriorly from the middle of the big toe to the heel, or a similar line drawn on the other side extending from between the 4th and 5th toe to the heel. Refer to Blood Specimen Collection and Handling Procedure for the shaded areas demarcating preferred puncture sites (see Appendix A).

2. Thoroughly wash the puncture site with soap and water.

3. Disinfect the skin with 70% isopropyl alcohol. Vigorous rubbing during this step may help stimulate blood flow to the area. Allow time to air dry.

4. Puncture the skin in one continuous motion using the Lancet. (Avoid the use of long tip lancets which may damage the heel bone.)

5. Wipe away and discard the first drop of blood since it may be contaminated by disinfectant or tissue fluid.

6. Allow a second drop to form from a spontaneous free flow of blood.

7. Expose the filter paper end of the DBS card. Touch the filter paper to the drop of blood, as close to the center of the circle as possible. Observing from the opposite side, allow the blood spot to enlarge until the circle is completely filled. There should only be a single application of the filter paper to the blood spot for each circle. (No dabbling of filter paper to the heel; this will cause an overlay of blood, which may cause inaccurate results.)

8. Once the blood collection is completed, press a sterile gauze pad to the puncture site until the bleeding has stopped.

9. Dry the blood spots on a level, nonabsorptive surface, away from the direct sunlight, and at room temperature for at least four hours. DBS cards should not be stacked while the

filter paper specimens are exposed. After drying, rewrap the filter paper with the cover sheet to its original position to protect the specimen.

10. IMPORTANT: All information requested on the DBS card should be completed. Failure to complete critical information such as the infant's last name, the date and time of birth, and the date and time of collection will cause the specimen to be rejected by the screening laboratory and necessitate a repeat specimen collection on the infant by the originator of the specimen.

11. The DBS card should be mailed within 24 hours of collection to the screening laboratory utilizing the overnight courier service.

PROBLEMS WITH SPECIMEN COLLECTION

Dilution or contamination of the blood spot may occur due to the following errors:

- Failure to wipe off the alcohol residue
- Failure to discard the first drop of blood that forms, since it may be contaminated with alcohol or tissue fluids
- "Milking" the puncture site to stimulate bleeding, which can dilute the blood with tissue fluid
- Touching the blood spot circles before or after filling, which can contaminate the spots with perspiration and/or oil from fingertips
- Allowing blood spots to be contaminated by contact with water, feeding formulas, or antiseptics

The blood may be unevenly distributed on the filter paper due to the following errors:

- The repeated touching of the filter paper to small drops of blood in an attempt to fill a circle. This will result in areas overfilled due to layering one drop over a partially dried drop and some areas contain no blood at all.
- A mottled appearance will occur if the filter paper is not soaked through, front to back.
- Overfilling the circles causes the center of the circle to be supersaturated. This may cause inaccurate test results.
- Blood collected in capillary tubes and used to fill the circles. There is a chance of the capillary tube scratching the surface of the filter paper, which may alter the thickness in that area.

SPECIMEN COLLECTION/TRANSFERS

Optimal time for specimen collection is 24 – 48 hours after birth.

In the event of pending transfer to a facility for higher level of care, collect the newborn screen specimen prior to transfer, if possible.

If DBS specimen is not collected prior to transfer, indicate on the DBS card that the infant has been transferred; provide the name of the facility where newborn is transferred. Mail the DBS card with **no blood** on it to:

MSDH Genetics Program
P.O. Box 1700
Jackson, MS 39215-1700

SPECIMEN COLLECTION/TRANSFUSIONS

Optimal time for specimen collection is 24 – 48 hours after birth. If an infant is discharged before 24 hours of age or transferred to another facility for higher level of care, the specimen must be collected before discharge to assure that the infant is in the system. It is also recommended that the specimen is collected before a blood transfusion, if possible.

- Ideal collection time: 24 – 48 hours of age
- Transfusion required at > 24 hours of age: collect specimen prior to transfusion
- Transfusion required < 24 hours of age: collect specimen prior to transfusion and 2 – 4 days after the transfusion
- Transfusion required but no specimen collected prior to transfusion: collect specimen 2 – 4 days, 2 – 4 weeks and > 90 days after transfusion

SCREENING FOR CCHD

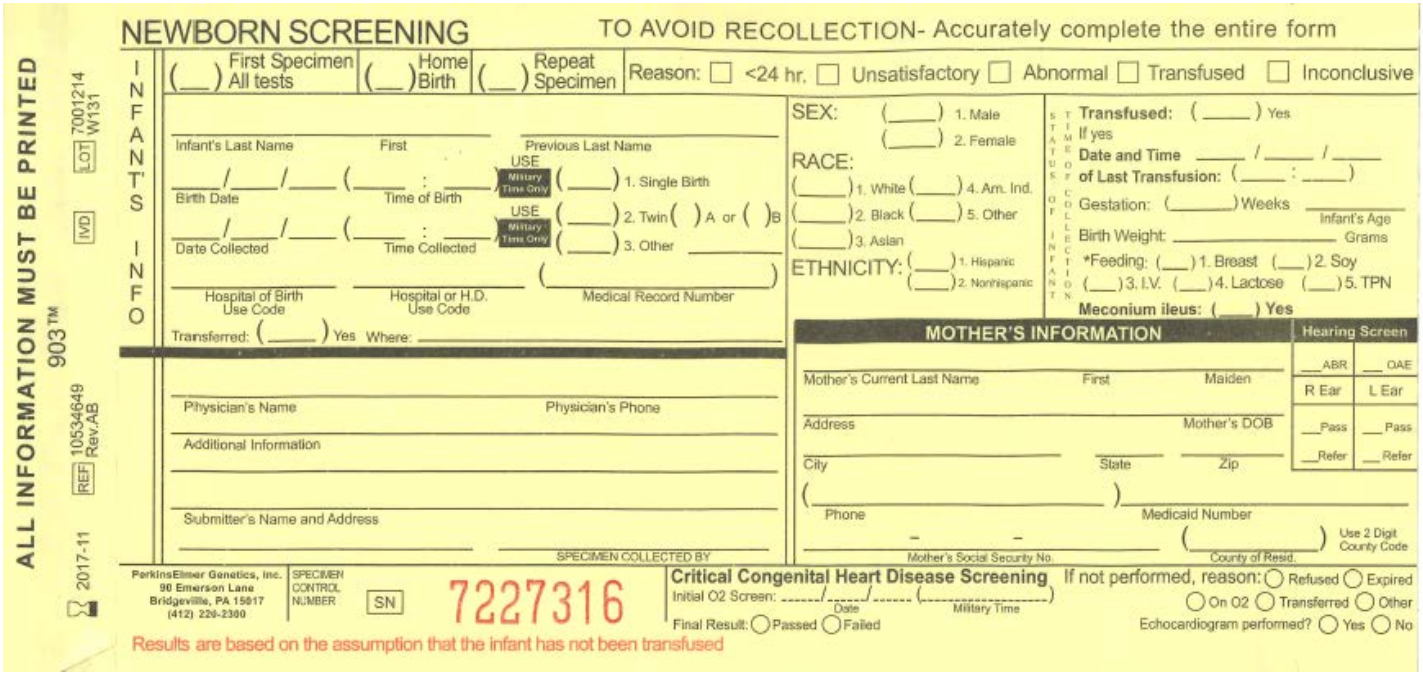
Annually, approximately 8 in every 1,000 babies are born with congenital heart disease (CHD) in the United States. CHD occurs when an infant is born with an abnormal structure of the heart, creating abnormal blood flow patterns. Critical congenital heart disease (CCHD) is a more serious group of heart defects that occurs in approximately 25% of CHD cases, and is one of the leading causes of infant death. Pulse oximetry, or "pulse ox", is a simple non-invasive screening test that detects low oxygen levels in the blood and may help to diagnose critical CHD before babies are discharged from the hospital.

CCHD screening should be performed before discharge from the nursery, after the infant turns 24 hours of age, and ideally between 24 to 48 hours. If the infant was born prematurely, screening should be performed when medically appropriate. If early discharge is planned, screening should occur as late as possible prior to discharge. Pulse ox screening should be performed while the infant is in the nursery, before discharge at a time when the infant is quiet, warm, and on room air. The pulse ox test should be performed on the right hand and one foot. In many centers a convenient time for the pulse ox screening to be completed is in conjunction with the hearing screen.

HOSPITAL HEARING SCREENING

All hospitals that deliver babies are equipped with newborn hearing screening equipment. Every baby (unless otherwise medically indicated) should have at least one hearing screen test documented on the dried blood spot card prior to submission. The first screen is completed shortly after birth when the baby is quiet. The second screen is completed prior to discharge if the baby refers in either ear on the first screen. **Both ears are to be rescreened.** If the baby refers in either ear during the first and/or second screens, an appointment is made by staff at the birthing hospital for an outpatient screen and documented in the medical record. **Both ears are to be rescreened.** In the spaces provided, check the appropriate test box and the ear screened (L or R). In the appropriate space, indicate the result "pass" or "refer".

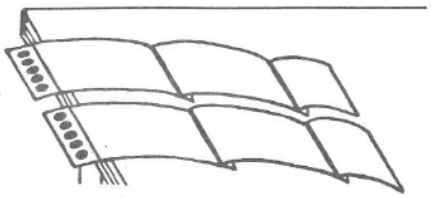
DRIED BLOOD SPOT CARD



INSTRUCTIONS

1. Hold infant's limb in a dependent position to increase blood flow.
2. Clean heel thoroughly. Wipe with alcohol and dry before puncturing.
3. Puncture heel with sterile lancet deep enough to assure free flow of blood.
4. Wipe away first drop and discard.
5. Allow a large drop of blood to form on infant's heel. Apply the back side of the filter paper directly to the puncture site where the drop of blood has formed. **The drop of blood should be large enough to approximately fill one circle.**
   DO NOT: a) Apply more than one drop of blood per circle.
   DO NOT: b) Apply blood to both front and back of filter paper.
6. Apply blood to all circles.
7. Allow blood spots to completely dry in a horizontal position at room temperature for a minimum of 4 hours (see diagram). Do not stack specimens while specimen is exposed. After drying, rewrap this cover sheet to its original position to protect specimen.
8. Send by Pre-Paid Overnight Courier within 24 hours of collection to:
9. If you have questions please call: Mississippi State Department of Health Genetic Screening Program at: (601) 576-7619

PerkinsElmer Genetics, Inc.
90 Emerson Lane
Bridgeville, PA 15017
(412) 220-2300



HOW TO ORDER DRIED BLOOD SPOT CARDS

To order Newborn Screening Pamphlets and dried blood spot cards complete a Newborn Screening Supply Form and fax to 601-576-7498 or call 601-576-7619. (See Appendix C.)

NEWBORN SCREENING RESULTS

The screening laboratory provides results to the birthing hospitals to file in the infant's medical record. Birthing hospitals can also obtain newborn screening results for specimens submitted by their facility by accessing Information@PerkinElmer.com. To request a User Access Form call PerkinElmer Genetics at 1-866-463-6436.

In addition, all results are electronically downloaded into the Mississippi State Department of Health's Newborn Screening Program system. Primary care providers can obtain a copy of the newborn screening results from the hospital of birth by faxing a written request (Request For Newborn Screening Results / Dried Blood Spot Card Form – see Appendix D) to the Newborn Screening Program (NBS) at 601-576-7498 or call the NBS Program at 601-576-7619 for questions Monday – Friday, 8 am – 5 pm.

The following information is needed to provide newborn screen results:

- Name
- Sex
- Date of birth
- Mother's name
- Hospital of birth

Parents should ask their baby's physician about the Newborn Screening results at the first well baby checkup. For more information on what parents should know about newborn screening and FAQs. (See Appendices E and F.)

UNACCEPTABLE OR INCONCLUSIVE RESULTS

The NBS Program coordinates repeat specimen collection for unacceptable or inconclusive test results. Repeat specimen collection is addressed by the hospital of birth or the Health Department in the county of residence.

IMMEDIATE FOLLOW-UP OF ABNORMAL OR PRESUMPTIVE POSITIVE RESULTS

All abnormal or presumptive positive screening results are reported to the NBS Program by the screening laboratory. If the infant is a hospital inpatient, the attending physician is notified of the results. If the infant has been discharged, the primary care physician of record is notified of the screen results. A disease-specific American College of Medical Genetics (ACMG) ACTion sheet is provided to the notified physician. The ACMG ACTion sheet provides a brief overview of the condition description, differential diagnosis, diagnostic evaluation, and course of action which can be used in conjunction with a consultation of a tertiary care specialist (www.acmg.net).

Once the primary care physician of record is notified of the screening results, the NBS Program may facilitate a referral to the tertiary care specialist as requested by the physician. Diagnosis will require an evaluation and confirmatory testing. Evaluation and treatment may require ongoing clinic visits with the appropriate clinical provider. (See Appendix I.)

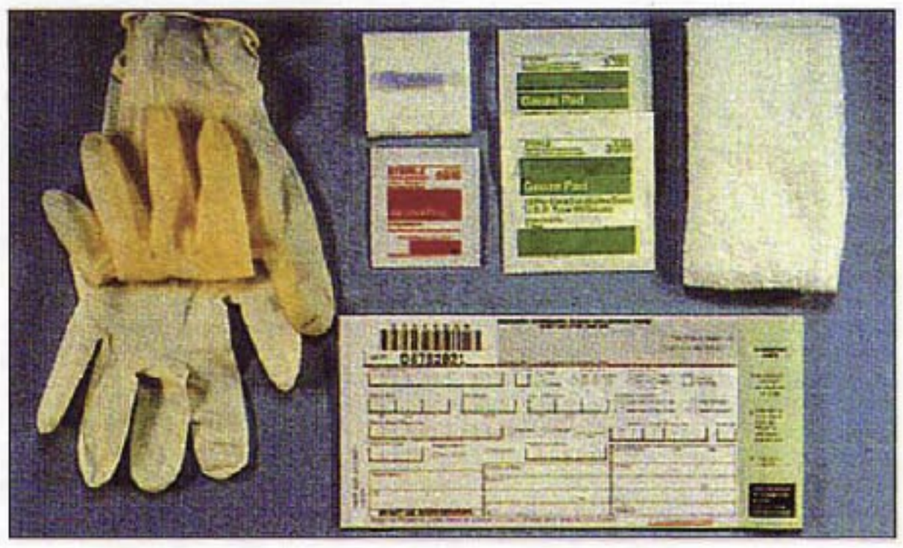
IMMEDIATE FOLLOW-UP OF ABNORMAL OR PRESUMPTIVE POSITIVE RESULT AFTER HOURS/WEEKENDS

After normal business hours, which includes weekends and holidays, abnormal or presumptive positive screening results are reported to the NBS Program staff. The NBS Program staff who is on-call immediately contacts the hospital of birth and the on-call physician. The results are given to the physician via telephone with instructions on how to access the ACMG website (www.acmg.net**)** for a disease specific ACTion sheet, and also how to contact the appropriate specialist (see Appendix I). The next business day, a copy of the newborn screening result will be faxed to the hospital of birth and the physician of record.

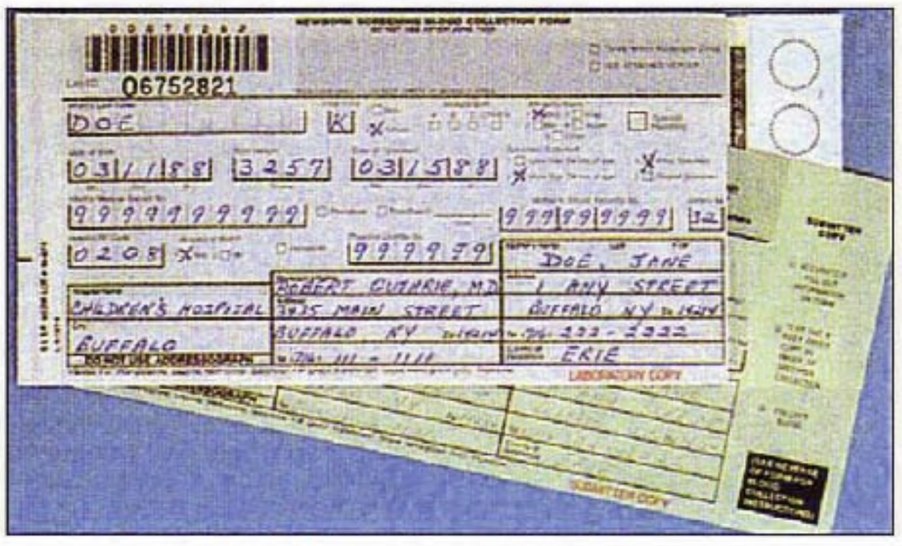
APPENDICES

Neonatal Screening

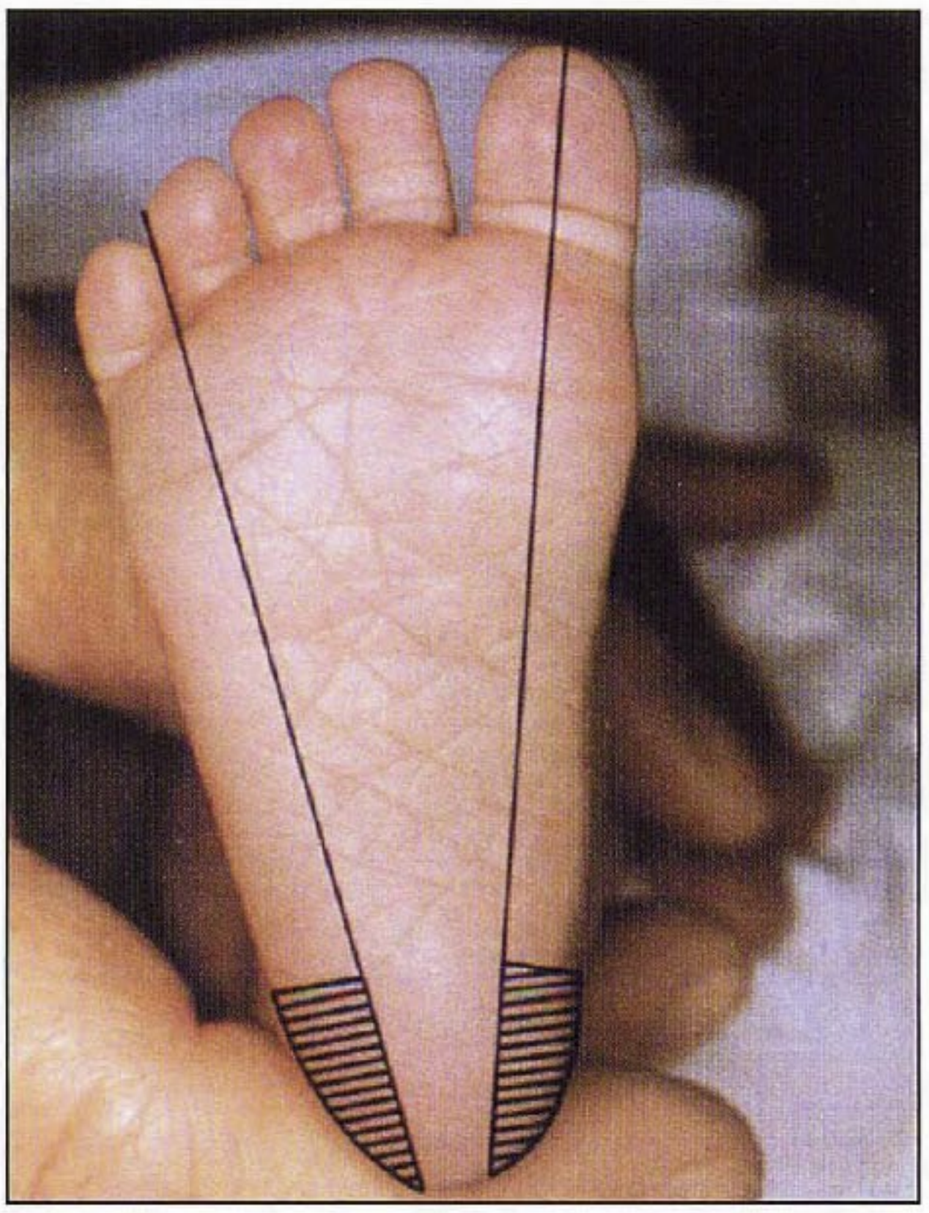
Blood Specimen Collection and Handling Procedure



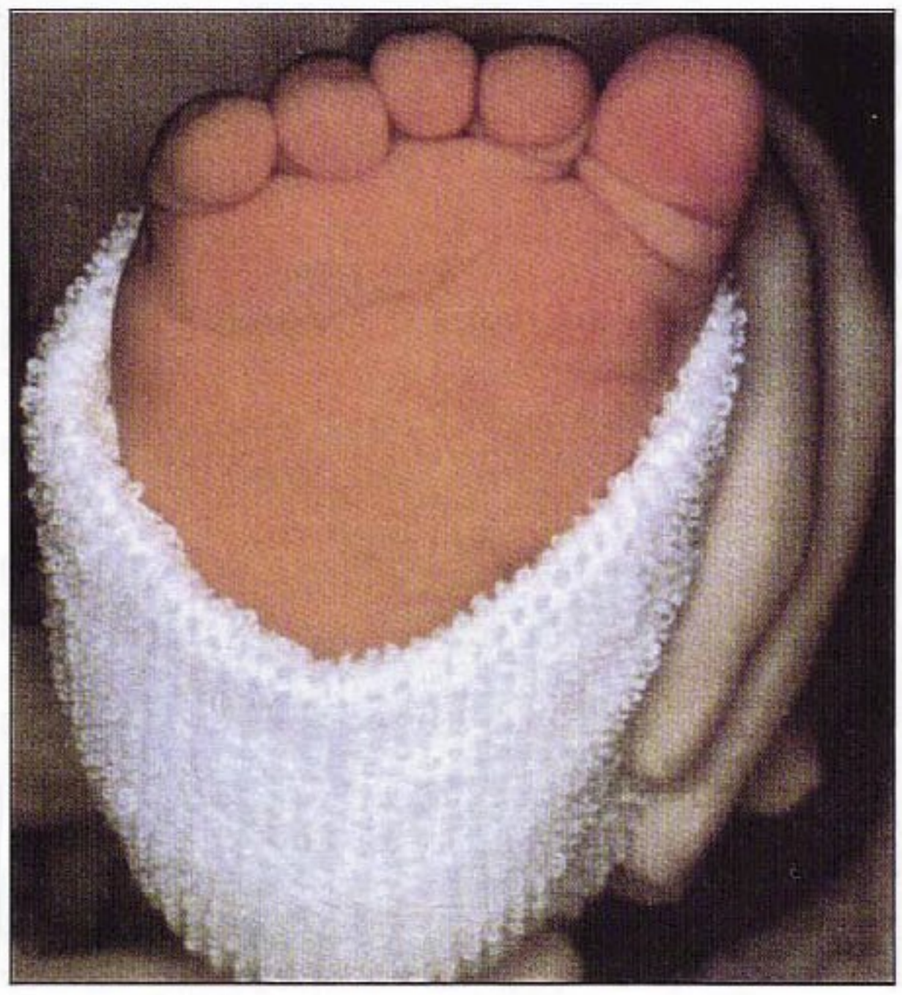
1 Necessary equipment: sterile lancet with tip approximately 2.0 mm, sterile alcohol prep, sterile gauze pads, soft cloth, blood collection form, gloves.



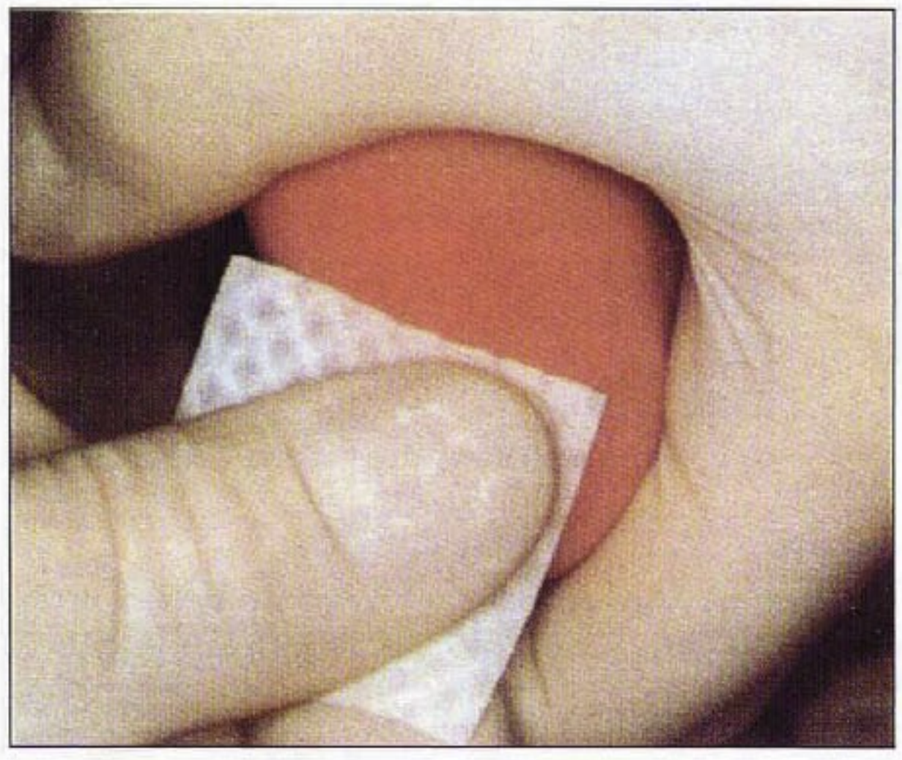
2 Complete ALL information. Do not contaminate filter paper circles by allowing the circles to come into contact with spillage or by touching before or after blood collection. Keep "SUBMITTER COPY" if applicable.



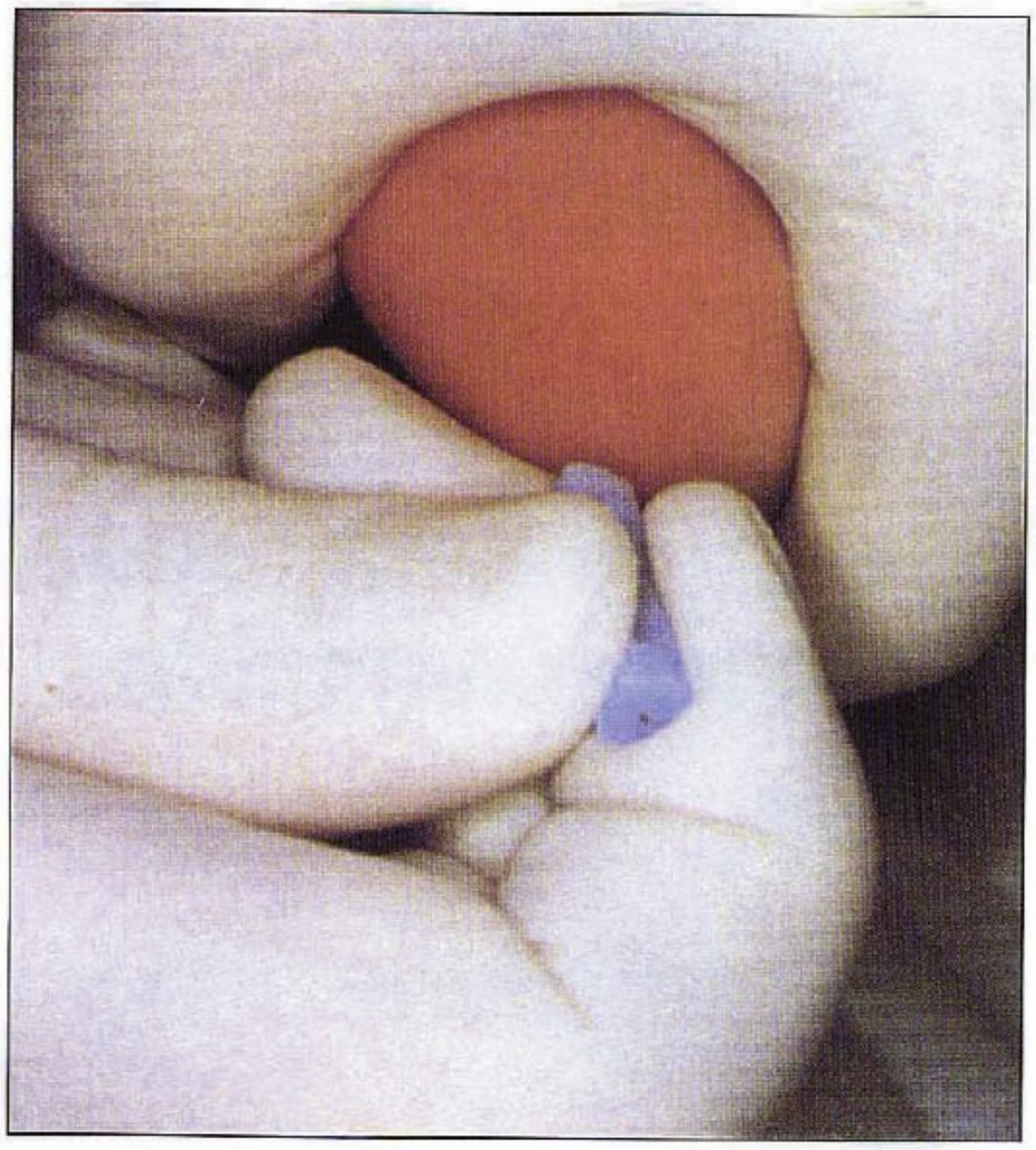
3 Hatched area (▨)indicates safe areas for puncture site.



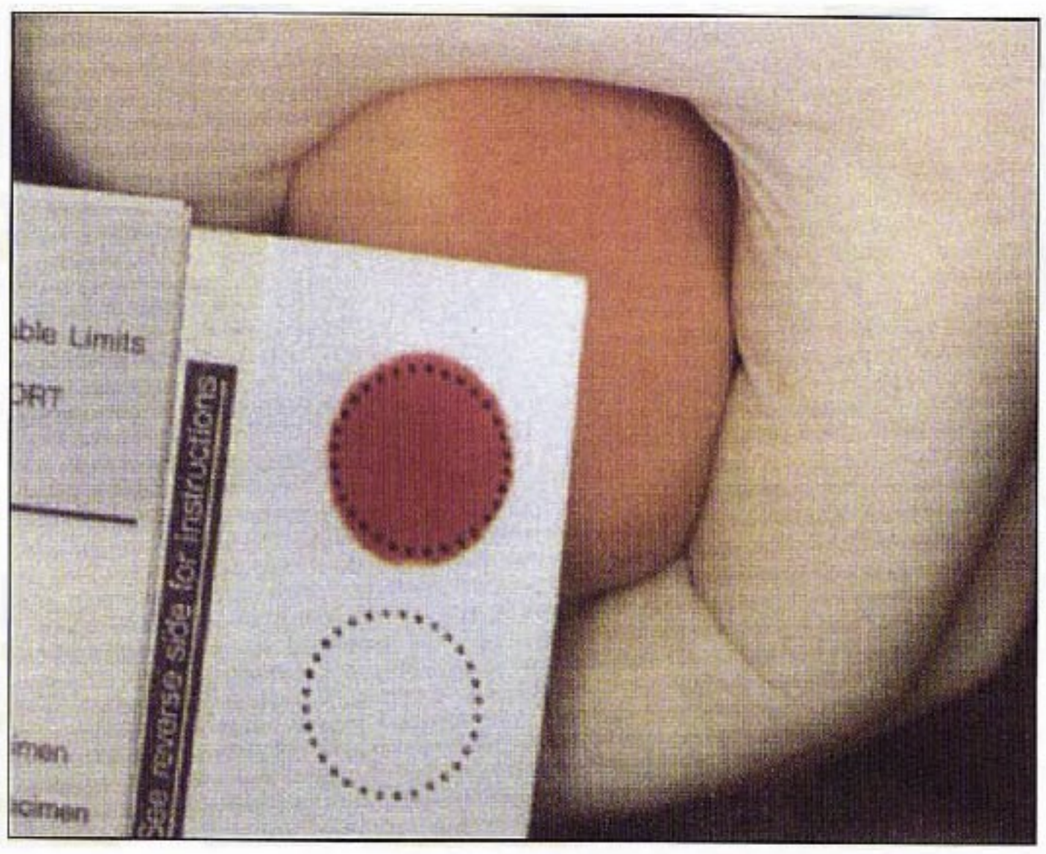
4 Warm site with soft cloth, moistened with warm water up to 41° C, for three to five minutes.



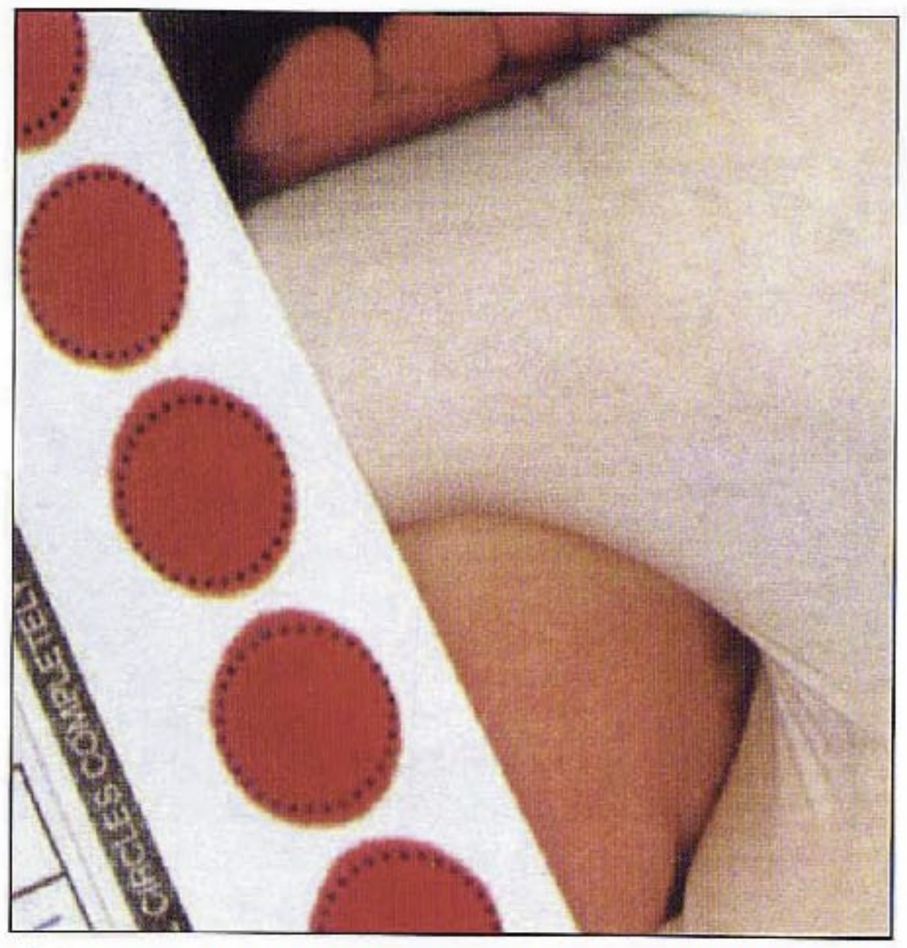
5 Cleanse site with alcohol prep. Wipe DRY with sterile gauze pad.



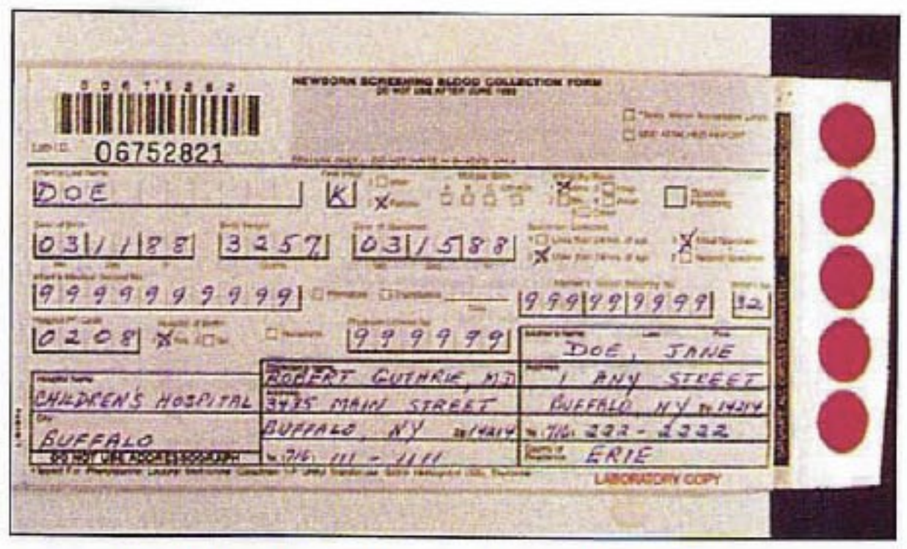
6 Puncture heel. Wipe away first blood drop with sterile gauze pad. Allow another LARGE blood drop to form.



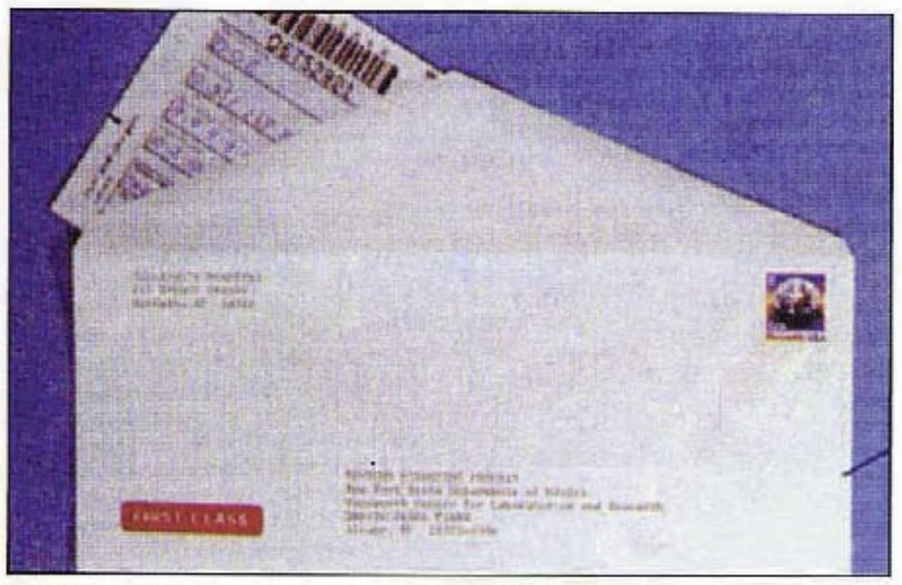
7 Lightly touch filter paper to LARGE blood drop. Allow blood to soak through and completely fill circle with SINGLE application of LARGE blood drop. (To enhance blood flow, VERY GENTLE intermittent pressure may be applied to the area surrounding the puncture site). Apply blood to one side of filter paper only.



8 Fill remaining circles in the same manner as step 7, with successive blood drops. If blood flow is diminished, repeat steps 5 through 7. Care of skin puncture site should be consistent with your institution's procedures.



9 Dry blood spots on a dry, clean, flat, non-absorbent surface for a minimum of four hours.



10 Mail completed form to testing laboratory within 24 hours of collection.

Information provided by The New York State Department of Health.

Eastern Business Forms
530 Old Sulphur Springs Road
Greenville, SC 29607
(864)288-2451
will@ebf-inc.com

Refusal of Newborn Screening

Parent(s) are allowed to refuse the newborn screening test for their infant only if their religious beliefs and practices do not allow testing. When a parent refuses the newborn screening test, staff *should*:

- Give parent a copy of the Newborn Screening pamphlet and discuss the significance of the NBS test.
- Ask for a signed statement from parent stating refusal to have baby screened for these very serious conditions (See example : Genetic Services – Newborn Screening Refusal)
- Place signed form or statement in the baby's hospital medical record, and send a copy of the signed/witnessed form to the MS Newborn Screening Program.

Mailing Address:
MSDH Genetics Program
P.O. Box 1700
Jackson, MS 39215-1700
Fax Number: 601-576-7498

APPENDIX B

Sample
Newborn Screening Refusal Form

Patient's Name _____

Date of Birth _____

Mother's Name _____

Hospital _____

Person(s) Counseled _____

Date _____

I, _____ have been counseled on the importance of Newborn
(Parent's Name)
Screening tests and have received literature on Newborn Screening. I do understand that the Mississippi state law requires every baby to be screened between 24 – 48 hours of birth. I understand that appropriate screening can allow for early detection, treatment, and follow- up. Many of these disorders may cause life-threatening medical conditions, mental retardation or even death, and many of these serious consequences can be lessened with timely medical intervention. However, due to my religious beliefs, I decline to have these tests performed on my child.

Signed _____

Witnessed _____

APPENDIX C

NEWBORN SCREENING SUPPLY FORM

Fax completed request to the Newborn Screening Program 601-576-7498

Requested by:	Date:	
ATTN:	Ship to:	
	Telephone Number:	
Description	**Quantity Requested**	**Quantity Shipped**
Newborn Screening Dried Blood Spot Cards "Filter Paper"		
Newborn Screening Pamphlet(#5198 English)		
Newborn Screening Pamphlet (#5198 Spanish)		
Screening for Critical Congenital Heart Defects		
5 Things to Know About Congenital Heart Defects		
What Does A Safe Sleep Environment Look Like (#5404 English)		
What Does A Safe Sleep Environment Look Like (#5404S Spanish)		
Baby's Safe Sleep Crib Checklist (# 5400 English)		
Newborn Hearing Screening "What to Expect" (#5272 English)		
Newborn Hearing Screening "What to Expect" (#5272S Spanish)		
Newborn/Infant Hearing Screening Report (#288)		
Infant Hearing Screening Log (#291)		
Infant Hearing Screening Log/ Data Summary (#81)		
Disk Mailers		
Newborn Hearing Screening/ Pass Card (Yellow & Green)		
Newborn Hearing Screening/ Refer Card (Yellow & Red)		
Supplier Use Only		
Filled by:	Date:	

Mail or FAX request to the below address, Attn: Genetic Services, fax to 601-576-7498 or call 601-576-7619.

570 East Woodrow Wilson ● Post Office Box 1700 ● Jackson, MS 39215-1700
601-576-8090 ● 1-866-HLTHY4U ● www.HealthyMS.com

Revised Date: February 2015

APPENDIX D

MISSISSIPPI STATE DEPARTMENT OF HEALTH

REQUEST FOR NEWBORN SCREENING RESULTS / DRIED BLOOD SPOT CARD

Phone (601) 576-7619 Fax (601) 576-7498

PLEASE PRINT LEGIBLY.
ILLEGIBLE AND INCOMPLETE REQUEST WILL BE RETURNED TO SENDER.

TYPE OF REQUEST:

☐ Newborn Screening Results ☐ Dried Blood Spot Card

REASON FOR REQUEST:

☐ Parent Request ☐ Recent Death

☐ Primary Care Provider/Pediatrician ☐ Other: _____

☐ Family History

REQUESTOR'S REQUIRED INFORMATION

Name of Facility: _____ Date: _____

Contact Person: _____ Requestor Name: _____

Facility Address: _____

Telephone ( ) _____Fax # ( ) _____

Child's Name (at birth) _____

Last First Middle Initial

Child's Date of Birth: _____ Sex: Male Female Indeterminate (Circle One)

Mother's Name: _____

Mother's Address: _____

Mother's DOB: _____

County of residence at birth: _____

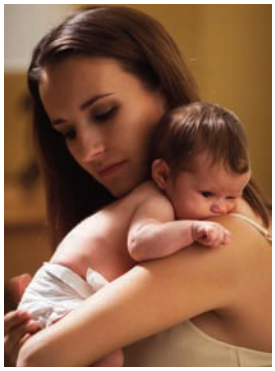
If child has a different last name than above please list below:

CONFIDENTIALITY NOTE: This information has been disclosed to you from records whose confidentiality is protected. Statutes/Regulations prohibit you from making further disclosures other than treatment, payment or health care operation, without the specific written authorization of the person to whom it pertains, or as otherwise permitted by such regulations.

Bureau of Genetic Services • 570 East Woodrow Wilson • Post Office Box 1700 • Jackson, MS 39215-1700
601-576-7619 • 1-866-HLTHY4U • www.HealthyMS.com

Equal Opportunity in Employment/Services

What Parents Should Know About Newborn Screening


All babies born in Mississippi are required by law to have a blood sample collected after birth to screen for a number of genetic disorders. Babies with genetic disorders may look healthy at birth, yet be at risk for serious health problems. Serious health problems may be prevented if these disorders are identified as soon as possible after birth. You will receive a pamphlet about newborn screening before you and your baby are discharged from the hospital.

Twenty-four to forty-eight hours after your baby's birth, a nurse or laboratory technician will draw a few drops of blood from your baby's heel. He or she will then send it to a laboratory for testing. Both you and your baby's physician will be notified if the screening indicates that further follow-up is needed, and a copy of the screening results will be sent to the hospital where your baby was born. These results will be included in your baby's medical record. Ask your baby's physician about the newborn screening results at your first "well baby" checkup.

Occasionally a second screening is required to confirm test results. If your baby requires a second screening, it should be done as soon as possible. Your local county health department will contact you if rescreening is necessary. To help with prompt notification, be sure to provide the name of your baby's doctor, your physical address and a telephone number before hospital discharge.

If you have any further questions about newborn screening, talk with your obstetrician and your baby's physician. You may also contact the Mississippi State Department of Health Newborn Screening Program at 601-576-7619 or 1-866-HLTHY4U (1-866-458-4948). You may also visit the Mississippi State Department of Health's website at www.HealthyMS.com.

Also available in Spanish

MISSISSIPPI STATE DEPARTMENT OF HEALTH

APPENDIX F
NEWBORN SCREENING IN MISSISSIPPI FREQUENTLY ASKED QUESTIONS

What is Newborn Screening?
Newborn screening refers to a dried blood spot screening test that is performed on babies before they are discharged from the hospital. The screen is done before discharge to identify babies who may be at risk for a condition with serious effects who might not otherwise be detected for several days, months, or even years. Only additional testing (diagnostic and/or confirmatory testing) can tell if the baby has a disorder.

Why should my baby have the screening?
The conditions for which babies will be screened are individually rare. However, some are very serious and can result in mental retardation, other serious medical problems, and/or death if not treated appropriately. Babies with these conditions often appear normal at birth. The newborn screen helps to identify babies who need medical treatment, such as medications or special diets.

Newborn screening may not always detect a disorder. If your baby does not seem well, talk to your baby's physician as soon as possible.

My baby looks healthy. Is this screen still necessary?
Yes! Most babies with a condition found by newborn screening show no signs of the condition right after birth. However, babies with a condition will need special medical care and the screen can usually identify these babies before they become sick.

Do I need to give permission for my baby to be screened for these conditions?
No. The state law (MS Statute 41-21-201) states that all babies born in Mississippi must be screened prior to discharge from the hospital unless your religious beliefs and practices do not allow this testing.

May I refuse the screening test?
As a parent, you may refuse newborn screening only if your religious beliefs and practices do not allow this testing. If you refuse to have the screen performed on your baby, you will be asked to sign a form (Newborn Screening Refusal) stating you refused to have your baby screened for these very serious conditions. This form will go in your baby's hospital medical record.

Is there a fee for the screening test?
A fee is billed to the hospital where your baby was born for the screen.

What conditions are currently screened for in Mississippi?
Mississippi Genetic Newborn Screening Panel

APPENDIX F

When and how is the screening test done?
The screening specimen is usually collected between 24 – 48 hours of age. A few drops of blood are taken by pricking the baby's heel and dropping blood onto a special screening card and shipped to the screening laboratory.

Will I be told the newborn screen test results?
Results are mailed to the hospital of birth. The physician of record is notified of any abnormal screen results by the Mississippi State Department of Health. It is very important that the hospital have the parent's correct last name, physical address, and a working telephone number.

Your baby's physician can request the screen result from the hospital or call the Mississippi State Department of Health's Genetic Services Program for the screen results. Ask your baby's physician about the screen results at the first well baby checkup.

If a repeat screen is requested, does it mean my baby may have a condition?
No. There are several reasons why a repeat screen may be needed. If the screen is done incorrectly and is not okay for testing, a second test (repeat) is needed. If a screen is collected too early, a repeat screen is needed. A retest for these reasons simply means another sample is needed.

If the first screen was abnormal for one of the conditions, a second test or a confirmatory test may be required. An abnormal screening result does not mean that your baby has a problem. The screening test does not say "yes" or "no" to whether your baby has a condition. It identifies a few babies who may need more specific testing. A confirmatory test may say "yes" or "no" to whether or not your child has a condition.

Can these conditions be cured?
Currently, there is no known cure for these conditions. The serious effects of these conditions can be lessened if a special diet, medical treatment, or other intervention is started early.

If my baby has one of these conditions, does it mean my future children will also be affected?
Your doctor can discuss this with you or refer you to a genetic specialist. These specialists have information about the specific condition and how it is inherited. They can help explain the potential risk for your future children.

APPENDIX G

CCHD DELAYED SCREEN REPORTING FORM

Utilize this form if the CCHD pulse oximetry screen was not completed **prior** to the submission of the newborn screening dried blood spot card.

Newborn Demographic Information:

First Name: _____ Last Name: _____

Birth Last Name: _____

DOB (mm/dd/yyyy): _____ Time of birth: _____

Sex: Male Female Indeterminate (Circle One)

Gestational age at birth (weeks): _____ Birth weight (grams): _____

Medical Record Number: _____

Mother Demographic Information:

First Name: _____ Last Name: _____

DOB (mm/dd/yyyy): _____

Address: _____

City: _____ State: _____ Zip: _____

Delayed Screening Information:

Date of initial pulse ox screening for CCHD: _____ Military Time: _____

If not performed, indicate the reason: (required field)

Refused	On O2	Other _____
Expired	Transferred	_____

Echocardiogram performed?

Yes No Unsure

Final Result of the CCHD Screen:

❑ Pass (Negative) ❑ Fail (Positive)

Person completing form: _____

Print Name

Title: _____ **Date Completed:** _____

Phone Number: _____ **Facility Name:** _____

APPENDIX H

CCHD FAILED SCREEN REPORTING FORM

Utilize this form if the CCHD pulse oximetry screen was failed.

Newborn Demographic Information:

First Name: _____ Last Name: _____

Birth Last Name: _____

DOB (mm/dd/yyyy): _____ Time of birth: _____

Sex: Male Female Indeterminate (Circle One)

Gestational age at birth (weeks): _____ Birth weight (grams): _____

Medical Record Number: _____

Mother Demographic Information:

First Name: _____ Last Name: _____

DOB (mm/dd/yyyy): _____

Address: _____

City: _____ State: _____ Zip: _____

Failed Screening Information:

Date of initial pulse ox screening for CCHD: _____ Military Time: _____

Was a prenatal ultrasound performed? (Circle one) Yes No Unsure

Screening Information	First Pulse Ox Screen Saturation Results	Second Pulse Ox Screen (if indicated) Saturation Results	Third Pulse Ox Screen (if indicated) Saturation Results
Right hand	%	%	%
Foot	%	%	%
Age (in hours)			

Was an echocardiogram performed? (Circle one) Yes No Unsure

If **yes** - date: _____ Facility Name: _____

Was the patient transferred? (Circle one) Yes No

If **yes** - Where? (Facility name): _____ Date of transfer: _____

Comment sections on back

Reason for failed screen. What is the final diagnosis that explains the failed pulse oximetry screening?

Cardiac Defects (check all that apply):

- ❑ Aortic Arch Atresia
- ❑ Aortic Arch Hypoplasia
- ❑ Coarctation of the Aorta
- ❑ Double-outlet Right Ventricle
- ❑ Ebstein Anomaly
- ❑ Hypoplastic Left Heart Syndrome
- ❑ Interrupted Aortic Arch
- ❑ Pulmonary Atresia, intact septum
- ❑ Pulmonary Stenosis
- ❑ Single Ventricle
- ❑ Tetrology of Fallot
- ❑ Total Anomalous Pulmonary Venous Return
- ❑ Transposition of the Great Arteries
- ❑ Tricuspid Atresia
- ❑ Truncus Arterious
- ❑ Ventricular Septal Defect

Other Cardiac Defect(s) – Describe: _____

Non-Cardiac – Explanation: _____

Normal evaluation after failed screen – Explanation: _____

Pending diagnosis – Explain:

_____

_____

_____

Person completing form: _____

Print Name

Title: _____ **Date Completed:**_____

Facility Name: _____

Phone Number: _____

APPENDIX I

University of Mississippi Medical Center
Jackson, Mississippi

TERTIARY CARE CENTER

Amino Acid Disorders, Organic Acid and Fatty Acid Disorders

Clinic	Staff
University Medical Center University of Mississippi Health Care Pediatric Medical Genetics 2500 North State Street Jackson, MS 39216 Emergency after hours: 601-984-1000 (Page metabolic/newborn screening physician)	Omar Ali Abdul-Rahman, MD Brian Kirmse, MD Julie Kaplan, MD Jennifer Cook, CNP Leslie Berryhill, R.D. LD Phone No. 601-984-1929 Pager No. 601-929-1521 Fax No. 601-984-1916

Endocrine Disorders

Clinic	Staff
University Medical Center University of Mississippi Health Care Pediatric Endocrine Clinic 2500 North State Street Jackson, MS 39216 Emergency after hours: 601-984-1000 (Page pediatric endocrine/newborn screening physician)	George Moll, MD Michael Torchinksy, MD Naznin Dixit, MD Simeen Pasha, MD Phone No. 601-984-5216 FAX No. 601-815-3672

APPENDIX I

University of Mississippi Medical Center
Jackson, Mississippi

TERTIARY CARE CENTER **(continued)**

Cystic Fibrosis

Clinic	**Staff**
University Medical Center University of Mississippi Health Care Pediatric Pulmonary Clinic 2500 North State Street Jackson, MS 39216 Emergency after hours: 601-984-1000 (Page pediatric pulmonary /newborn screening physician)	J. Marc Majure, M.D David Josey, M.D. Lynn Walker, M.D. Phone No. 601-984-5205 FAX No. 601-815-1050

Pediatric Allergy/Immunology

Clinic	**Staff**
University Medical Center University of Mississippi Health Center Pediatric Allergy/Immunology 2500 North State Street Jackson, MS 39216-4505 Emergency after hours: 601-984-1000 (Page pediatric allergy/immunology)	Anne Yates, MD Ray Rodriquez, MD Nina Dave, MD Phone No 601-984-5247 FAX # 601-984-2608

Pediatric Hematology/Sickle Cell Disease

Clinic	**Staff**
University Medical Center University of Mississippi Health Care Pediatric Hematology 2500 North State Street Jackson, MS 39216 Emergency after hours: 601-984-1000 (Page pediatric hematology/sickle cell/ newborn screening physician)	Gail Megason, MD Betsy Herrington, MD Cathy Gordon, MD Suvankar Majumdar, MD Amy Forsythe, CFNP Tobi Breland, CFNP Phone No. 601- 984-5220 FAX No. 601- 984-5279

* Out of state tertiary centers will be identified upon request of providers/family.

APPENDIX J
MISSISSIPPI NEWBORN SCREENING FLOW CHART

The flow chart above explains the path of a screening specimen from collection to results. This process includes birthing hospitals, the newborn screening laboratory, newborn screening staff, primary care providers, and tertiary care centers.

* Reasons for a repeat newborn screening include, but are not limited to, specimen quantity insufficient for testing, specimen appears diluted or contaminated, and specimen exhibits serum rings.