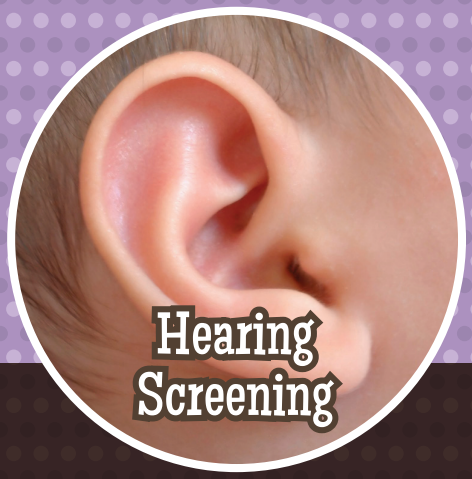
Alabama Newborn Screening Program

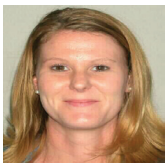


Newborn Screening Reference Manual for Medical Providers 2015

Alabama Newborn Screening Follow-up Program
Bureau of Family Health Services
201 Monroe Street
RSA Tower - Suite 1350
P.O. Box 303017
Montgomery, AL 36130-3017

Alabama Newborn Screening Laboratory
Bureau of Clinical Laboratories
8140 AUM Drive
P.O. Box 244018
Montgomery, AL 36124-4018

Cindy Ashley, BSN, RN, C
Newborn Screening Director
Phone: 334-206-2971
Fax: 334-206-3791



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Newborn Hearing Screening Coordinator
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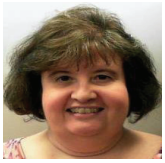
Fax: 334-206-3791



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Newborn Screening Laboratory Supervisor
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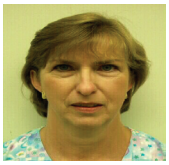
Fax: 334-260-3439



Lynn Green, BS, MT (ASCP)
Newborn Screening Laboratory Supervisor
Phone: 334-260-3476
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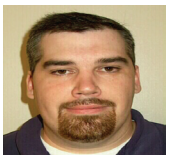

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Table of Contents

Alabama Newborn Screening Program

Section 1 - Program Overview

Alabama Newborn Screening Program

The goal of the Alabama Newborn Screening Program is to ensure state laws, rules and regulations mandating newborn screening are carried out in order to identify specific genetic disorders early and provide appropriate follow-up care.

The Alabama Newborn Screening Program is a comprehensive and coordinated system that provides education, screening, follow-up, diagnosis, evaluation, and management of disorders typically not apparent at birth. Newborn screening is mandated by Statutory Authority Code of Alabama 1975, Section 22-20-3. The screening allows treatment to be initiated within the first few weeks of life preventing some of the complications associated with genetic and endocrine disorders. Early diagnosis may reduce morbidity, premature death, intellectual disability, and other developmental disabilities. The Alabama newborn screening panel includes 31* of 32 disorders recommended by the U. S. Department of Health and Human Services Secretary's Discretionary Advisory Committee on Heritable Disorders in Newborns and Children (DACHDNC).

The Alabama Newborn Hearing Screening Program collaborates with the National Center for Hearing Assessment and Management (NCHAM) to ensure that all infants and toddlers with hearing loss are identified as early as possible and provided with timely and appropriate audiological, educational, and medical intervention. In addition, the Program collaborates with Children's Rehabilitation Service (CRS) to ensure infants receive second tier follow-up screening and diagnostic confirmation of hearing loss by three months of age, and the Alabama Early Intervention System (AEIS) to ensure infants are enrolled in early intervention before six months of age.

The Newborn Screening (NBS) Laboratory performs tests that aid in the diagnosis of 29 genetic disorders. In addition, screening is performed for over 15 secondary disorders, bringing the total to more than 45 disorders. Each year, the Alabama NBS Program identifies approximately 150-200 infants with a metabolic, endocrine, hematological, or other congenital disorder that may not be apparent at birth. All newborns identified with a disorder have access to a diagnostic evaluation through medical specialists throughout the state. These consultants work closely with the NBS laboratory, follow-up staff, and the primary care provider in determining needs, such as additional testing, medication, diet, and developing a treatment plan when necessary.

*SCID screening to be added in 2015

HIPAA

The Alabama Newborn Screening Program contacts health providers daily, and sometimes providers are unsure if information may be shared due to HIPAA regulations. Please read the notice provided by the ADPH General Counsel regarding HIPAA guidelines related to newborn screening.

STATE OF ALABAMA DEPARTMENT OF
PUBLIC HEALTH

Donald E. Williamson, MD
State Health Officer

January 18, 2012

Dear Alabama Newborn Screening Providers:

Subject: HIPAA and Newborn Screening Information

In light of HIPAA, concerns have been raised regarding sharing information with the Alabama Department of Public Health regarding newborn screenings. Exchange of information regarding newborn screenings is permissible under HIPAA because HIPAA allows the disclosure of protected health information without patient authorization if the disclosure is required by law or if the disclosure is required for public health activities. Disclosures regarding newborn screenings fall into both of these categories.

Specifically, the HIPAA regulations state that they do not pre-empt laws "for the conduct of public health surveillance, investigation, or intervention." 45 CFR 160.203(a)(2)(c). The regulations further provide that disclosures can be made without patient consent if the disclosure is required by law or if the disclosure is required for public health activities such as "preventing and controlling disease, injury, or disability" and "the conduct of public health surveillance, public health investigation, and public health interventions." 45 CFR 164.512(a) and (b).

State law requires that health care providers report all results of the newborns tested to the Alabama Department of Public Health. Ala. Admin. Code 420-10-1-.04(2). Therefore, providers must continue reporting newborn screening results to the Alabama Department of Public Health pursuant to state law and in compliance with HIPAA.

The U.S. Department of Health and Human Services (HHS), who promulgated the HIPAA regulations, and the Centers for Disease Control (CDC) emphasized the public health exception to HIPAA in guidance issued on April 11, 2003. The guidance states that covered entities may disclose protected health information to public health entities, without patient authorization, for the conduct of public health surveillance, investigations, or interventions, as well as for the purpose of preventing or controlling diseases. Additionally, the HHS Office of Civil Rights guidance issued on July 6, 2001 states that covered entities may rely on the judgment of a public health entity when requesting a disclosure as to the minimum amount of information that is needed by Public Health.

The RSA Tower • 201 Monroe Street • Montgomery, AL 36104
P.O. Box 303017 • Montgomery, AL 36130-3017

Alabama NBS Panel of Disorders

There are thirty-one disorders currently part of the Alabama Newborn Screening Panel of Primary Disorders and over forty total disorders including secondary conditions. Please see appendix for a brief description and timeline of each disorder.

Primary Conditions:

1. 3-Hydroxy-3-methylglutaric aciduria (HMG)
2. 3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC)
3. Argininosuccinic Acidemia (ASA)
4. Beta-ketothiolase Deficiency (βKD)
5. Biotinidase Deficiency (BIOT)
6. Carnitine Uptake Defect (CUD)
7. Citrullinemia type I (CIT)
8. Classic Galactosemia (GALT)
9. Congenital Adrenal Hyperplasia (CAH)
10. Congenital Hypothyroidism (CH)
11. Critical Congenital Heart Disease (CCHD)
12. Cystic Fibrosis (IRT/DNA)
13. Glutaric Acidemia type I (GA1)
14. Hearing Loss (HEAR)
15. Hemoglobin S/beta-thalassemia (Hb S/β Th)
16. Hemoglobin SC Disease (HbS/C)
17. Hemoglobin SS Disease (HbSS)
18. Homocystinuria (HCY)
19. Isovaleric Acidemia (IVA)
20. Long-chain L-3-OH Acyl-CoA Dehydrogenase Deficiency (LCHAD)
21. Maple Syrup Urine Disease (MSUD)
22. Medium-chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
23. Methylmalonic Acidemia (Cbl A, B)
24. Methylmalonic Acidemia due to mutase deficiency (MUT)
25. Multiple Carboxylase Deficiency (MCD)
26. Phenylketonuria (PKU)
27. Propionic Acidemia (PROP)
28. Trifunctional Protein Deficiency (TFP)
29. Tyrosinemia type I (TYR I)
30. Very Long-chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)
31. Severe Combined Immunodeficiency (SCID) to be added 2015

Secondary Conditions:

1. Carnitine acylcarnitine translocase deficiency
2. Carnitine palmitoyltransferase II (CPTII)
3. Glutaric acidemia type II (GA-II)
4. 2-Methylbutyrylglycinuria (2-MBG)
5. 2-Methyl-3-hydroxybutyric aciduria (2M3HBA)
6. 3-Methylglutaconic aciduria (3MGA)
7. Methylmalmonic acidemia (CBL-CD)
8. Defects of biopterin cofactor biosynthesis
9. Defects of biopterin cofactor regeneration
10. Citrullinemia type II
11. Benign Hyperphenylalaninemia (H-Phe)
12. Hypermethioninemia (MET)
13. Tyrosinemia type II
14. Tyrosinemia type III
15. Variant Hgb

Medical Provider Responsibilities

Medical Providers in Alabama are notified of abnormal newborn screening results by:

- Mail as long as the provider is identified on the specimen collection form.
- Immediate phone call and mail for potential positives and/or abnormal results that are outside of set cutoff values.
- Healthcare providers are encouraged to use Alabama's Secure Remote Viewer (SRV) to access newborn screening results (see page 11-12).

The role and responsibilities of primary healthcare providers and birthing hospitals in newborn screening follow-up may include:

1. Ensuring that newborn screening occurs and that results are known. Do not assume the newborn screen is normal if not notified of an abnormal result as some of the following may occur:
    - Provider information may be missing or inaccurate on form.
    - Specimens may be lost in the mail.
    - Hospitals may fail to collect a newborn screen prior to hospital discharge or transfer.

2. Contacting families about out-of-range or invalid screening test results in a knowledgeable and sensitive fashion**.**

3. Facilitating repeat or confirmatory testing and appropriate subspecialty care and reporting the results of confirmatory tests and the diagnosis to the Alabama Newborn Screening Program.

4. Collecting a repeat newborn screen as soon as possible if the first test is unsatisfactory.

5. Ensuring that the recommended hearing screening method is used for rescreening of infants who fail an initial screen (see page 36).

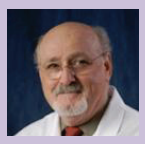
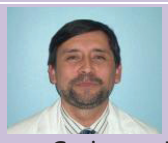
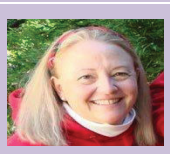
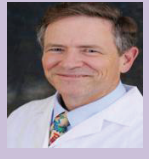
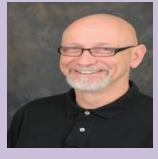
6. Obtaining a signed statement for parent refusal of newborn screening (see page 15).

7. Collecting a second newborn screen at 2-6 weeks of age (4 weeks optimal) on full-term infants and 4-12 weeks for low birth weight infants (less than 2000 grams) since hypothalamic immaturity could obscure meaningful TSH elevations.

8. Ensuring that provider contact information stays current with the state lab so that collection forms and test reports can be provided appropriately (see page 33).

    Reference: Clinical and Laboratory Standards Institute (CLSI). *Newborn Screening Follow-up; Approved Guideline.* CLSI document I/LA27-A.

Alabama NBS Medical Consultants

Disorders:	Specialists:
Endocrine – CH/CAH  Gail Mick, M.D. COA  Anne Marie Kaulfers, M.D. USA	1. USA Medical Center, Endocrinology Samar Bhowmick, M.D. **251-405-5147** Anne Marie Kaulfers,M.D. Christina Hair, R.N. **251-434-3723** 2. Children's of Alabama, Endocrinology Gail Mick, M.D. **205-638-9107** Leslie Pitts, CRNP **205-996-9166**
Hemoglobinopathies - Sickle cell disease, trait conditions and other hemoglobinopathies  Felicia Wilson, M.D. USA  Thomas Howard, M.D. COA	1. USA Sickle Cell Center Felicia Wilson, M.D. **251-405-5147(3)** Andretta McCovey, R.N. **251-405-5121** 2. Children's of Alabama, Pediatric Hematology Thomas Howard, M.D. **205-638-9285** Sharon Carlton, R.N. **205-558-2390** 3. St. Jude Clinic at Huntsville Hospital Carolyn Russo, M.D. **256-265-5833** Heidi Simpson, CRNP **256-265-5833**
Cystic Fibrosis  Hector Gutierrez, M.D. COA	1. Children's of Alabama CF Care Center Hector Gutierrez, M.D. **205-638-9583** Staci Self, LGSW **205-638-5494**
Metabolic Amino Acid Disorders Fatty Acid Disorders Organic Acid Disorders  S. Lane Rutledge, M.D. UAB	1. UAB Genetics S. Lane Rutledge, M.D. **205-975-6867** Maria Descartes, M.D. **205-934-1154** Alicia Roberts, R.D **205-996-6983**
Critical Congenital Heart Disease (CCHD)  Wally F. Carlo, M.D. UAB	1. UAB Pediatric Cardiology **205-934-3460 (direct)** **1-800-UAB-MIST (paging)** 2. Pediatric Specialists of Montgomery **334-612-2111 (direct and paging)** 3. Cardiology Associates of Mobile **251-434-9177 (direct and paging)** 4. Diagnositc & Medical Clinic (Mobile) **251-435-1200**
Severe Combined Immunodeficiency (SCID)  T. Prescott Atkinson, M.D. COA  Frederick Goldman, M.D. UAB	1. Children's of Alabama T. Prescott Atkinson, M.D. Allery, Asthma, Immunology **205-638-9586** 2. UAB – Lowder Pediatric Blood and Marrow Transplantation Program* ***infants to be referred for bone marrow transplantation by immunologist if indicated**

Secure Remote Viewer Instructions

Secure Remote Viewer (SRV) is a web-based system that allows healthcare providers access to newborn screening results. The system allows users to search, view, and print results immediately from their computer.

SRV REGISTRATION

The Secure Remote Viewer (SRV) requires registration with the Alabama Newborn Screening Program. Physicians may register with the system by completing the registration form found on the next page and faxing it to (334) 260-3439. We will verify that you are currently in the State Newborn Screening Laboratory system to be eligible to gain access to SRV.

Each physician is required to provide their state license number, National Provider Identifier (NPI), and an email address. On the registration form you will also be asked to provide two options for the account's user name. Once registration is complete, the registrant will receive their username and password via the email account provided. The email will not include the link to the SRV website for security purposes. You will need to log into the link below to access the SRV once you receive your username and password.

Authorized users will be able to find and view the most recent newborn screening results for each patient after providing the required minimum search criteria.

The following is a listing of requirements in order to utilize the SRV application:

- ✓ Web Browser: compatible with Mozilla Firefox v3.0 and higher; Microsoft Internet Explorer v7.0-v9.0
- ✓ Operating Systems: Windows 2000, Windows 7, XP, and Vista
- ✓ Pop-up Blocker: must be turned off in the browser settings or a website exception added in settings to ensure authentication and for the lab report (PDF) pop-up's to appear.
- ✓ PDF Viewer: must have to view lab report
- ✓ Cookies: browser must be set to enable cookies or a website exception added for the SRV web address (see user guide)

SRV INSTRUCTIONS

1. You will receive an email from donotreply_srv@adph.state.al.us with your username and password (check SPAM or Junk Mail if you have not received it within 2 days of submitting your request).

2. To access SRV, go to: https://neosrv.adph.state.al.us

3. Log in using the username and password provided.

4. You will be prompted to reset your password.

5. Once you are at the Home Screen select the icon ***Access Result Reports***.

6. An infant's test results can be found by entering the last name, date of birth, and hospital of birth in addition to any one of the following: mother's first or last name, infant's first or last name, mother's social security number, or form number (6 digit number on filter form followed by last 2 digits of birth year).

7. Once the search criterion has been entered select the ***Perform Search*** button at the bottom of the page.

8. If the minimum criterion has not been entered "Invalid Search Criteria" will display.

9. If the system is unable to find an infant, "No Records Found" will be displayed.

10. If there are results matching the search criteria, they will be displayed along with the specimen's status (pending or reported).

11. Once the infant's results are located the user will check the box next to the name and select ***View Report***. The user has the ability to print or save the results. More than one box may be checked if the infant has multiple reports.

Secure Remote Viewer Registration

Please complete this form if you would like access to the Secure Remote Viewer (SRV), which provides newborn screening results via the web. In order to gain access to SRV you must currently be registered to receive results via mail with the State Health Laboratory.

PLEASE PRINT

Name of Physician _____
(first name) (last name)

Name of Facility _____

Mailing Address _____

_____

_____

Area Code/Telephone Number _____

E-Mail Address _____
REQUIRED: The registrant will receive an invitation via email.

User name: option 1:_____ option 2: _____

Physician's State License # _____NPI# _____

Signature of Physician _____

Please fax or mail to:

Alabama Department of Public Health
Bureau of Clinical Laboratories
P.O. Box 244018
8140 AUM Drive
Montgomery, Alabama 36124-4018
Fax: 334-260-3439

If you have any questions please call 334-260-3476.

Disclaimer: You must agree that you are a healthcare professional providing care for those infants whose records you will view and agree to keep confidential all information made available to you before gaining access to the SRV system. Any unauthorized access, use, and/or disclosure of information may result in loss of access privileges and may be subject to penalties, fines, and criminal charges in accordance to the Health Insurance Portability and Accountability Act of 1996 (HIPAA), Public Law 104-91.

Alabama Voice Response System

Notice:

The Alabama Voice Response System will no longer be supported. You must register for Secure Remote Viewer to access newborn screening lab reports. **See page 11-12.**

Newborn Screening Education Material

Newborn screening material may be ordered directly online at the following site http://adph.org/newbornscreening/Default.asp?id=2164 or by completing the information below and faxing to 334-206-3791.

Newborn Screening materials are reserved for Alabama Newborn Screening Providers. There is a limit to the number of material that can be ordered at one time.

Please use a separate order form for each item ordered

Hospital/Practice Name:_____

Mailing Address:_____

City/Zip Code:_____

Telephone:_____ Contact Person:_____

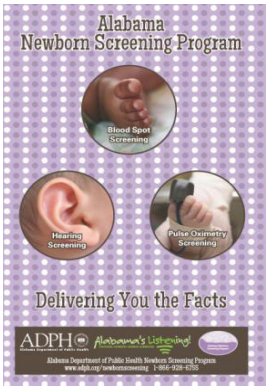
Education Material Number:

ADPH-FHS-_____

Quantity Requested (FHS-533 comes in packets of 50 and FHS-537 comes in packets of 100):

_____ packets (limit of 10 packets)

FHS-533



Description: Booklet that includes bloodspot, hearing, and pulse ox screening information. Replaces all other brochures. Spanish version not available at this time.

FHS-537 (5x8 card)

Description: Single 5x8 card for expecting parents. Includes four statements parents need to know about newborn screening. A Spanish version is also available.

Alabama Newborn Screening Program

Newborn Screening Refusal Form

The American Academy of Pediatrics and the Alabama Department of Public Health strongly recommend Newborn Screening for all infants.

Child's Name:_____ Date of Birth:_____

Name of Delivery Hospital: _____

Parent/Legal Guardian: _____

My child's medical provider _____ has advised me that my child

(named above) should participate in the newborn screening program.

☐ As the parent or legal guardian of my child (named above), I choose to decline participation in my state's newborn screening program, on the grounds that such tests conflict with my religious tenets and/or practices (as allowed by the Code of Alabama 1975, 22-20-3).

I have been provided information about newborn screening in my state and the importance of early identification of the disorders. I have had the opportunity to discuss these with my child's medical provider, who has answered my questions regarding the recommended screening. I understand the following:

- The purpose and need for newborn screening to include bloodspot screening, hearing screening, and pulse oximetry screening.
- The risks and benefits of newborn screening.
- **If my child does not participate in newborn screening, the consequences of a late diagnosis may include delayed development, mental retardation, or death.**
- My child's medical provider, the Alabama Department of Public Health, and the American Academy of Pediatrics strongly recommend that all newborns be screened for certain disorders.
- If my child has one of my state's screened conditions, failure to participate in newborn screening may endanger the health or life of my child.

Nevertheless, I have decided at this time to decline participation in the newborn screening program for my child as indicated by checking the box above.

I acknowledge that I have read this document or it has been read to me in its entirety, and I fully understand it.

Parent/Legal Guardian Signature_____ Date _____

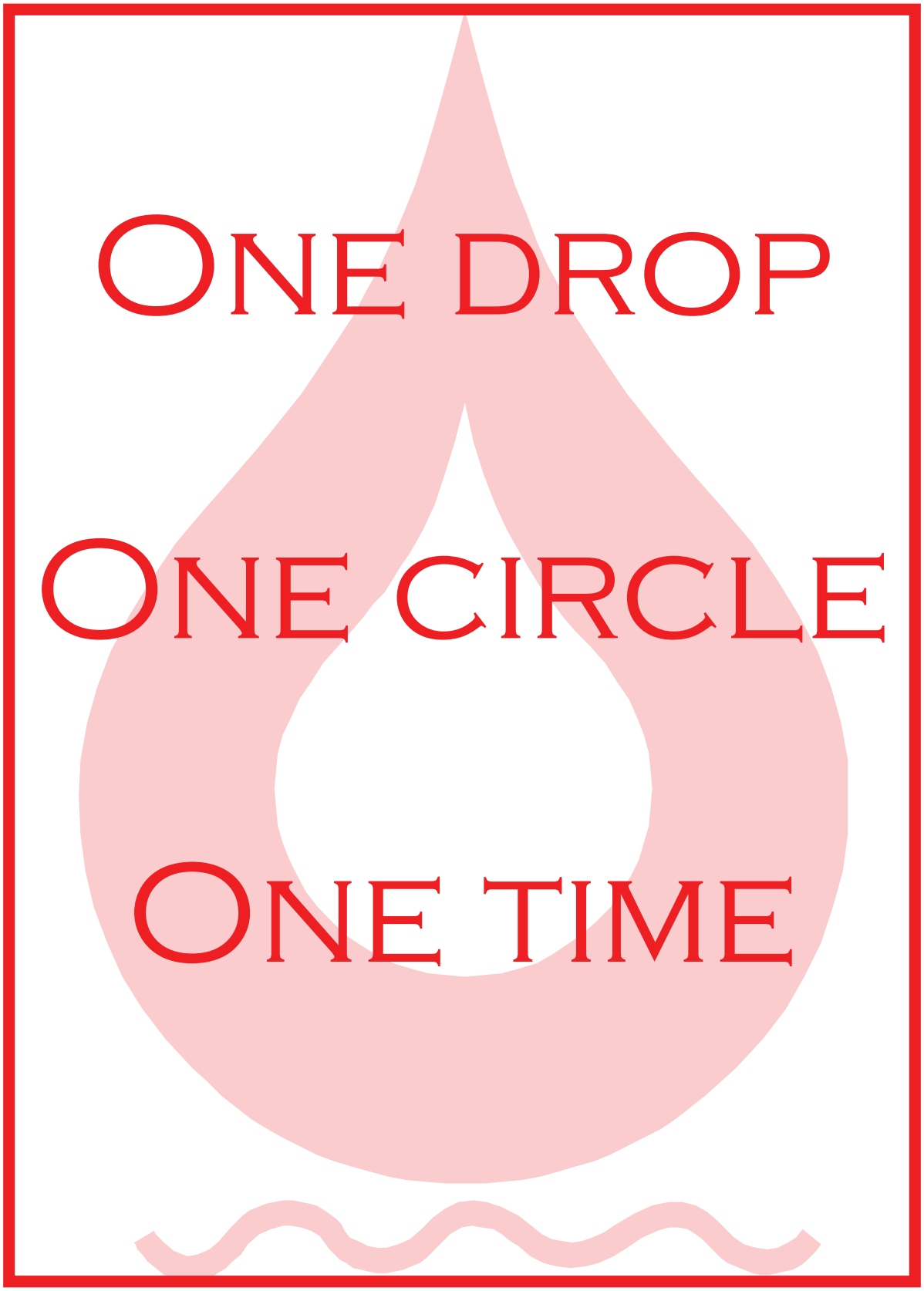
Witness _____ Date_____

I have had the opportunity to discuss my decision not to participate in my state's newborn screening program and still decline the recommended participation.

Alabama Birthing Hospitals

Birthing Hospital	Birth # 2013	Birthing Hospital	Birth # 2013
Huntsville Hospital	4687	Walker Baptist Medical Center	755
UAB Hospital	3963	Crestwood Medical Center	715
Brookwood Medical Center	3943	Dekalb Regional Medical Center	694
St. Vincent's Hospital	3500	Vaughn Regional Medical Center	686
Baptist Medical Center East	3463	Cullman Regional Medical Center	666
USA Children's & Women's Hospital	2787	Marshall Medical Center South	660
DCH Regional Medical Center	1876	Springhill Medical Center	630
Providence Hospital	1793	South Baldwin Regional Medical Center	573
East Alabama Medical Center	1741	Trinity Medical Center	573
Southeast Alabama Medical Center	1451	Madison Hospital	539
Northport Medical Center	1382	UAB Medical West	530
Northeast AL Regional Medical Center	1344	Athens Limestone Hospital	482
Jackson Hospital	1316	Andalusia Regional Hospital	467
Flowers Hospital	1215	Coosa Valley Medical Center	460
Thomas Hospital	1162	Russell Medical Center	455
Gadsden Regional Medical Center	1114	George H. Lanier Memorial Hospital	398
Shelby Baptist Medical Center	1114	Marshall Medical Center North	359
Mobile Infirmary Medical Center	1068	Citizens Baptist Medical Center	313
Helen Keller Memorial Hospital	1060	Highlands Medical Center	312
St. Vincent's East	1044	D.W. McMillan Memorial Hospital	278
Eliza Coffee Memorial Hospital	927	Princeton Baptist Medical Center	255
Decatur Morgan Hospital	917	North Baldwin Infirmary	239
Medical Center Enterprise	840	Grove Hill Memorial Hospital	200
Baptist Medical Center South	739	Monroe County Hospital	192

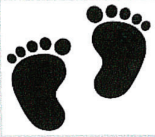
Section 2 - Specimen Collection


One drop
One circle
One time

2014 Notice of New Filter Forms

ATTENTION

NEWBORN SCREENING PROVIDERS



Effective November 1, 2014 the new NBS collection forms (CL-89) should be used for collecting the NBS specimen.

Please discontinue use of all old "A" forms (orange) and "B" forms (purple) after their expiration date of 10-31-2014. All specimens that are collected on these filter paper forms after 10-31-2014 will not be satisfactory for testing. Discard all old forms.

- The new **First test (A) forms** are shaded in **BLUE.** Please use these forms to collect all initial screening specimens - expiration date **05-2017**.
- The new **Second test (B) forms** are shaded in **PINK**. Please use these forms to collect all second test, follow-up and requested repeat specimens - expiration date **05-2017**.
- An **"INSURANCE INFORMATION"** form is included in both the A and B forms. These forms should remain intact for submission with the NBS specimen. The form should be filled out completely as instructed on the back of the insurance form.
- **PHYSICIAN INFORMATION:** The new forms have three fields for physician information. **The information in all 3 fields must be provided**.
  - ORDERING PHYSICIAN - The physician that is ordering the NBS panel
  - NPI# - The National Provider ID for the **ordering physician**
  - REFERRAL PHYSICIAN - The physician that will provide care for the newborn. This physician will be contacted for follow up on an abnormal NBS report.
- **PULSE OXIMETRY SCREENING** - Results provided by the hospital performing the test.

DO NOT WRITE ON OR AFFIX LABELS TO THE "LABORATORY USE ONLY" AREA OF THE NBS COLLECTION FORM.

NOTE: INFORMATION ON THE COLLECTION FORM WILL NOT BE CHANGED BY THE LABORATORY AFTER IT HAS BEEN RECEIVED FOR TESTING PURPOSES. IT IS IMPERATIVE THAT THE FORM BE COMPLETED ENTIRELY AND ACCURATELY. TEST RESULTS WILL BE EVALUATED WITH THE INFORMATION THAT IS ON THE FORM AT THE RECEIPT OF THE SPECIMEN IN THE LABORATORY.

We would like to thank everyone for your diligence in proper collection and completion of collection forms. Please call Danita Rollin, Deannie Morris, Lynn Green, or Derek Kennedy with any questions concerning Newborn Screening at the NBS Laboratory at 334-260-3400.

Newborn Screening Collection Guidelines

Section 22-20-3 (as amended in 1987) of the Code of Alabama states that all infants must be administered a reliable test for PKU, Cystic Fibrosis, Hypothyroidism, CAH, Galactosemia, Abnormal Hemoglobins, Biotinidase Deficiency, Amino Acid Disorders, Fatty Acid Disorders, and Organic Acid Disorders and that the testing be performed by the Public Health Laboratory.

Timing of Screening:	
FIRST TEST ("A" FORM) – ***This specimen is tested for Hypothyroidism, CAH, Cystic Fibrosis, Galactosemia, Hemoglobinopathies, Biotinidase Deficiency, Amino Acid Disorders, Fatty Acid Disorders, and Organic Acid Disorders.***	
Full Term Infants	A newborn screening test should be collected when the infant is 24-48 hours of age. If the infant is discharged prior to 24 hours of age, a specimen MUST be obtained before discharge, and the parent or guardian must be informed of the importance of obtaining a repeat test before one week of age.
Home Births	The Newborn Screening Statute applies to all infants born in Alabama. The birthing attendant is responsible for collecting the newborn screening test. It is recommended that the test be collected at 24-48 hours of age.
Extended Hospital Stay (low birth weight/sick infants)	It is recommended that a specimen be collected upon admission to the NICU if the infant is expected to receive TPN or transfusions unless the infant is so unstable that it cannot be done safely. The Alabama Newborn Screening Sick Infant Blood Collection Guidelines follows.
Transitioning Infants	Infants admitted to NICU for short term observation but are not receiving TPN or transfusions should have a specimen collected according to the Full Term Infant Protocol.
Dying Infants	If an infant is likely to die, it is appropriate to collect a newborn screening specimen. While dying infants may have abnormal results as a response to organ failure, the specimen may also provide a diagnosis of an early onset screening disorder.
Older Infants	The American Academy of Pediatrics recommends that physicians know the screening status of all children in their care. While older infants may enter the practice without evidence of a newborn screen, the Alabama Department of Public Health's Newborn Screening Program has established standards and cutoffs for newborns and infants and therefore **cannot accept specimens on children older than 12 months of age**.
Special Considerations:	
Transfused Infants	A specimen should be collected prior to transfusion regardless of age or treatments unless the infant is so unstable it cannot be done safely. If the specimen is not collected prior to transfusion, collect a specimen greater than 72 hours post transfusion. Another specimen should be collected at 3-4 months post transfusion for Hemoglobinopathies, Biotinidase Deficiency, and Galactosemia. If a Galactosemia condition is suspected and the specimen was not collected prior to transfusion, place the infant on a galactose-free diet until a definitive diagnosis can be made.
Transferred Infants	The transferring facility must collect a specimen prior to transfer regardless of age or treatments unless the baby is so unstable that it cannot be done safely. If the specimen cannot be obtained prior to transfer, the transferring facility must ensure that the next facility is aware of the need for collection of the newborn screening specimen.

Newborn Screening Collection Guidelines

Parent Refusal	Parents may refuse newborn screening only for religious reasons. Parents who refuse under this condition should sign a statement that is placed in the infant's medical record. **A newborn screening collection form should be filled out completely with a statement as to the refusal and mailed to the State Laboratory**.

SECOND TEST ("B" FORM) – *This specimen is tested for Hypothyroidism, CAH, Cystic Fibrosis, Galactosemia, Biotinidase Deficiency, Amino Acid Disorders, Fatty Acid Disorders, and Organic Acid Disorders.*

Note: This specimen is not routinely tested for Hemoglobinopathies. If no valid test has been done for this disorder, please see instructions below for collection of requested repeat specimens, "Requested Repeat."

1. A second newborn screening specimen should be collected at 2-6 weeks of age (4 weeks optimal) on all full term infants with a normal first test screen.
2. If the first test specimen was collected when the infant was greater than one week of age but less than two weeks of age, the second test specimen should be collected at 4-6 weeks of age.
3. If the first test specimen was collected after two weeks of age, a second ("B") specimen need NOT be collected.

Requested Repeat ("B" form)

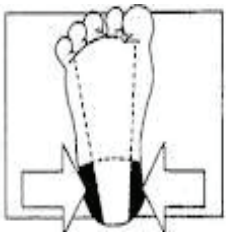
1. A repeat specimen may be requested by the State Laboratory when the results are abnormal or questionable. The specimen should be collected in the time frame indicated by the report. The "Retest-Prior Abnormal" box must be marked on the collection form.
2. If the first test is unsatisfactory for testing, a repeat test should be collected as soon as possible. The "Retest-Prior Unsat" box must be marked on the collection form.

Collection of Filter Paper Bloodspot Specimen

Materials needed for Blood Collection:

1. Gloves
2. 70% isopropyl alcohol pads
3. Dry sterile gauze pads
4. Sterile sticking device with a point not greater than 2.4 mm in depth (the most effective method is the use of scalpel bladed lancets)
5. Newborn Screening filter paper collection form (CL-89) with protective envelope

Bleeding Procedure:



1. The preferred puncture site is indicated by the shaded areas on the heel. The least hazardous sites for heel puncture are medial to a line drawn posterior from the middle of the big toe to the heel or lateral to a similar line drawn on the other side extending from between the 4th and 5th toe to the heel.
2. Warm the infant's foot if necessary using warm water, towel, or chemical pack. Heat sources should not exceed 42°C and should not be left in contact with the skin for a prolonged period.
3. Disinfect the skin with alcohol pads and allow to air dry. Vigorous rubbing during this step stimulates blood flow to the area.
4. Puncture the skin in one continuous motion using a sterile sticking device with a tip <2.4mm. **THE USE OF LONGER TIPS MAY DAMAGE THE HEEL BONE**.
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 of blood to form by the spontaneous free flow of blood.

Newborn Screening Collection Guidelines

Collecting the Blood Spots:

1. Before collecting the blood, fold back the protective flap to expose the filter paper. Do not touch or handle the filter paper before or after applying the blood.
2. Lightly touch the filter paper against a large drop of blood and allow a sufficient quantity of blood to soak through to completely fill the circle. Apply blood to one side of the filter paper only, allowing full saturation of each circle. Either side of the filter paper may be chosen. Fill all circles. Do not layer successive small drops of blood to the same circle. Avoid touching or smearing the blood spots.
3. If blood flow is diminished, repeat the bleeding procedure with sterile equipment.
4. Once all the circles have been filled, press a sterile gauze pad to the puncture site and hold the infant's foot above the level of the heart until bleeding has stopped.
5. Dry the blood spots on a level, non-absorptive surface away from direct sunlight and at room temperature for at least 4 hours.
6. After blood spots are completely dry, replace the protective flap over the specimen and place form in the protective envelope (do not use plastic) and mail to the State Laboratory within 24 hours.

Guidelines and Possible Sources of Error:

Improperly prepared blood spots and failure to complete the information form accurately constitute a major problem for the testing laboratory. Good blood spot preparation will assure prompt and accurate testing.

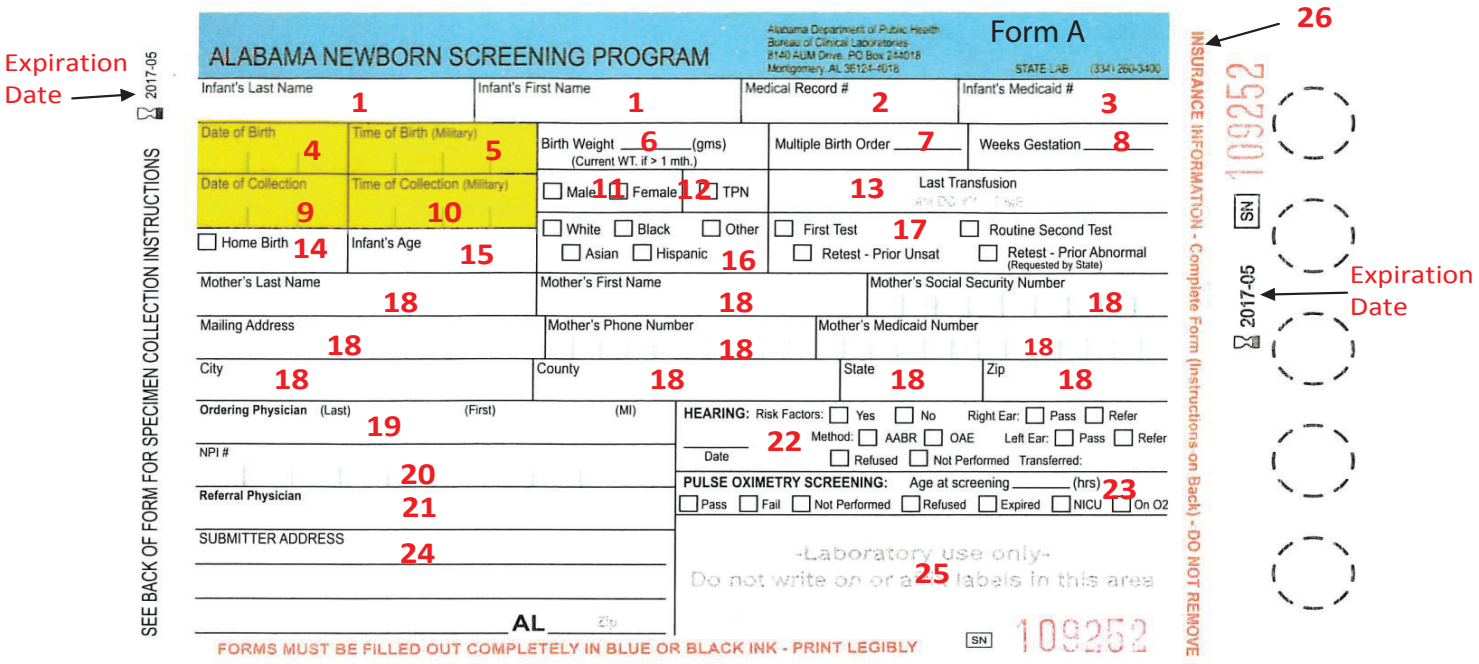
The following guidelines may help eliminate unsatisfactory specimens or erroneous test results.

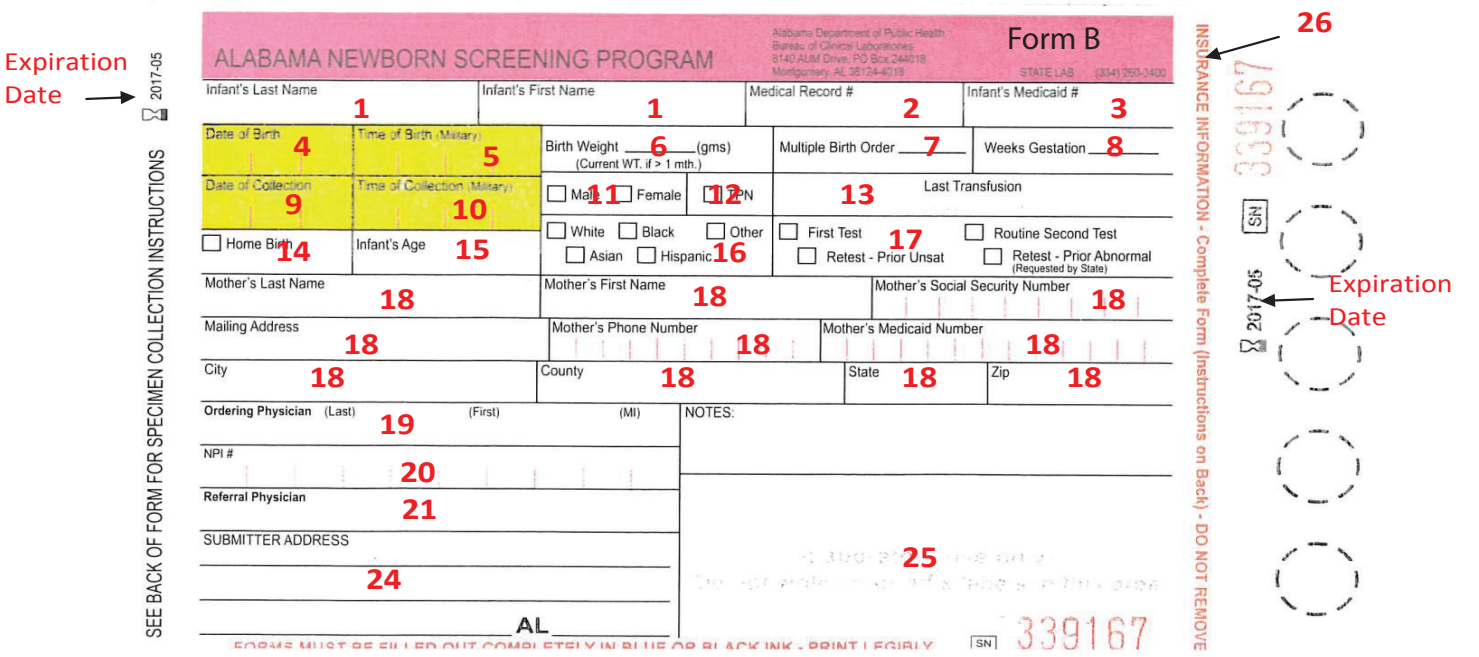
1. Do not touch any part of the filter paper circles before, during, or after collection.
2. Collect the specimen on the proper Newborn Screening collection form.
3. Complete all demographic data. This information is vital for interpretation of newborn screening results and for identification and location of infants for follow-up of abnormal test results.
    a) Always note any transfusion of red blood cells.
    b) Mark TPN feeding if TPN is being administered at time of collection.
    c) NPI # should be provided for the Ordering Physician (physician ordering the NBS screen).
4. Wipe away the first drop of blood to remove tissue fluids and alcohol. Do not "milk" the puncture site.
5. Do not expose the specimen to heat or humidity at any time. Do not dry on heater, in microwave, with a hair dryer, or in the sunlight. Do not place in plastic bags, leave in hot mailbox, or hot car; proteins and enzymes will be destroyed.
6. Ensure that the specimen is properly dried before replacing the protective flap and before placing in the protective envelope.
7. Dry specimens in a horizontal position. Hanging wet specimens will cause heavier red cells to migrate to the end of the circle causing an uneven saturation.
8. Do not superimpose blood drops on top of each other.
9. Apply blood to only one side of the filter paper.
10. Collecting blood samples after feeding promotes better blood flow.
11. Do not allow specimens to come in contact with water, feeding formulas, antiseptics, urine, etc.

Newborn Screening Collection Guidelines

Completing the Demographic Data – A & B FORMS

Always complete the specimen collection form using a black or blue ball point pen and print legibly to ensure that the patient is identified properly.





These forms are examples and may not be current. **These forms will expire 5-2017**.

Newborn Screening Collection Guidelines

1	**Name field** – enter the patient's last name and first name (if applicable).	2	**Medical Record field** – enter the patient's medical record number. This number is for the submitting facility to identify the patient when the report is received.
3	**Medicaid field** – enter the infant's Medicaid number if applicable.	4	**Birth date field** – enter the birth date in the format MM/DD/YY (required field).
5	**Time of Birth field** – enter in military format, failure to use military format may result in erroneous test results since many lab tests are based on the age of the infant at the time of collection.	6	**Birth Weight field** – enter the infant's birth weight in **grams**. If the infant is more than one month of age, enter the current weight. The laboratory sets standards and cutoffs for some tests using weight. Indicating the weight helps to ensure accurate test results and eliminate the need for unnecessary repeat specimens.
7	**Multiple Birth Order field –** complete **only** if there is a multiple birth. Enter the birth order as A, B, C, etc.	8	**Gestational Age field** – enter the gestational age as number of completed weeks.
9	**Date of Collection** – enter the date of collection in the format MM/DD/YY (required field).	10	**Time of Collection** – enter the time of collection in military format (required field)
11	**Sex field –** check appropriate box	12	**TPN field –** If infant is receiving TPN feeding at time of collection, check the box
13	**Last Transfusion field** – Complete this box with the date and time of the infant's last transfusion of **red blood cells**. Date should be entered as MM/DD/YY and time in military format .The date and time of transfusion are important for the laboratory to determine whether the the results are valid. Failure to indicate transfusions can result in an infant with a NBS disorder being missed due to the presence of donor cells in the specimen.	14	**Home birth field** – check the home birth box if the infant was born outside of the birthing facility with a birthing attendant present.
15	**Infant's Age field** – enter the infant's age at the time of specimen collection.	16	**Race field** – mark the appropriate box for the infant's race.
17	**Type of Tests field** - mark the "First Test" box if the specimen is the first one collected on this infant. Mark the "Routine Second Test" box if the specimen is the routine second test specimen collected on this infant. If a prior test on this infant was reported as unsatisfactory, mark the "Retest-Prior Unsat" box. If a prior test on this infant was abnormal and the State Laboratory requested a repeat sample, mark the "Retest-Prior Abnormal" box.	18	**Mother's Information fields** – enter the mother's information in the appropriate fields. ***Mother's social security number should be entered accurately.*** This will allow the submitting facility to access test results more readily and ensures that infants needing immediate follow-up can be located quickly.
19	**Ordering Physician field** – enter the **full name** of the physician who has ordered the NBS tests. ***This information is required to be provided and complete.***	20	**NPI field** - enter the National Provider Identification **9 digit** number for the **ordering physician**. ***This information is required to be provided and complete.***
21	**Referral Physician field** – enter the **full name** of the physician who will be caring for the infant. This physician will be contacted if the infant has a potential NBS disorder and his/her name will be listed as the physician on the NBS laboratory report. (This physician may be the same as the ordering physician – but should be entered in this field as instructed)	22	**Hearing field** – On the "A" form enter the date that the hearing screen was performed in the format MM/DD/YY and check the appropriate boxes to indicate the results of the hearing screen and the testing method used.
23	**Pulse Oximetry Screening field** – On the "A" form enter the age, in hours, of the infant when the screening was performed. Check appropriate "Pass" or "Fail" box. Check appropriate "Not Performed", "Refused", "Expired", "NICU", and/or "On O2" as it applies	24	**Submitter field** – enter the name and address of the facility submitting the specimen. Do not use abbreviations as there are facilities with similar names. An address label may be attached in this area as long as it does not obscure other fields or hang off of the edge. ***This information is required to be complete and accurate.***
25	**Lab use field** - Do not write or place labels in this area. This space is used by the laboratory to attach a unique identification number to the specimen for use in the laboratory.	26	**INSURANCE FORM: - Insurance information MUST be entered completely and accurately. This sheet should not be removed from the NBS form.**

Sick Infant Blood Collection Guidelines

February 26, 2015 (replaces previous guidelines dated 5/28/2013)

The following newborn screening protocol for premature or sick infants has been developed by a task force of professional medical providers and consultants and has been approved by the Alabama Newborn Screening Advisory Committee. These recommendations are in keeping with the recommendations of the Clinical Laboratory Standards Institute (CLSI) as well as the standards required by the Alabama Department of Public Health Laboratory.

Premature or Sick Infants (5 day or less NICU stay):

- Verify that these infants have at least one screen done at > 24 hours of age before discharge.
- Collect the recommended outpatient newborn screen between 2-6 weeks of age as per the Alabama Department of Public Health Bureau of Clinical Laboratories Newborn Screening Blood Collection Guidelines.

Premature or Sick Infants (5 days or longer NICU stay):

- The **first** newborn screening specimen is to be collected when the infant is admitted to the NICU (before transfusions, TPN, or antibiotics).
- The **second** newborn screening specimen is to be collected between **48-72 hours of age** (at least 72 hours post transfusion). If on TPN at time of second screen then collect a repeat screen 2 days after TPN is discontinued or upon discharge from NICU, whichever comes first.
- The **third** newborn screening specimen is to be collected at **28 days of age** or at discharge, whichever comes first, in very low birth-weight infants (less than 2000 grams) because hypothalamic immaturity could obscure meaningful TSH elevations.

Collect the first sample on a First Test Form (A Form). Collect any subsequent samples on a Second Test Form (B Form).

Transferred Infants – the receiving hospital, on admission, should collect a specimen on a Second Test Form and mark the "First Test" box.

Note: If results from the first or second newborn screens place infant at high suspicion for a condition, appropriate confirmatory or diagnostic tests should be done as with any other infant, being alert to the effects that treatments and the infant's condition may have on the screening test results.

Hemoglobinopathy Repeats

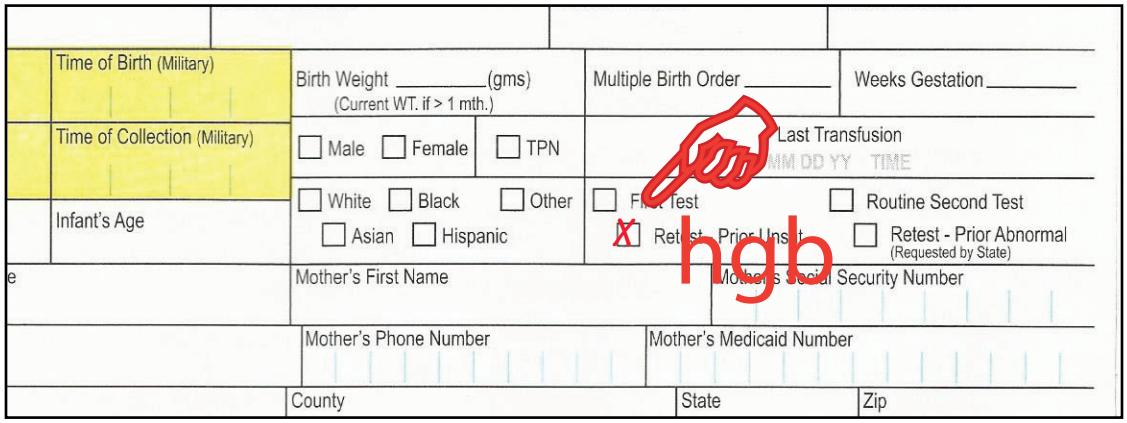
Please verify initial Hemoglobinopathy results before requesting a repeat newborn screen.

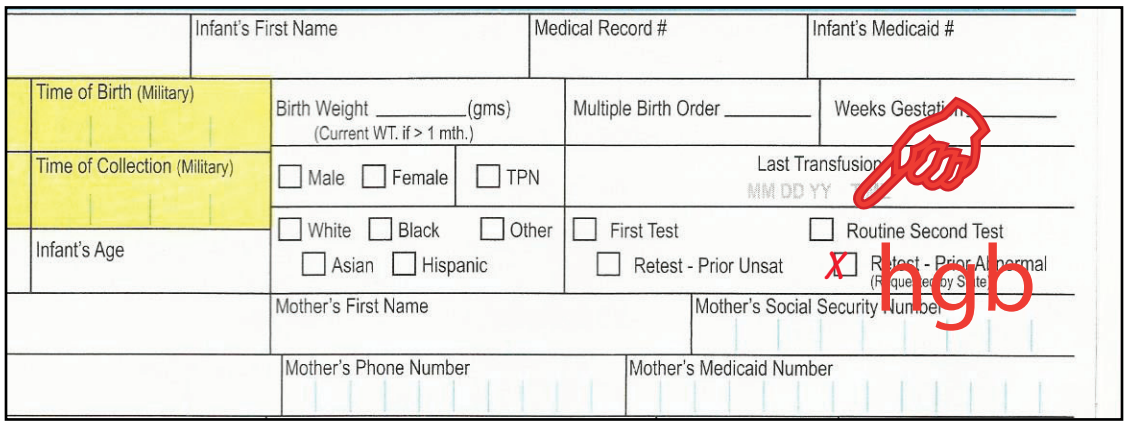
1. If a repeat Hgb is needed, please complete the "B" filter form by marking either *Retest Prior Abnormal (Requested by State)* or *Retest Prior Unsat* and write "hgb" on form as shown in the example below.

2. **Hemoglobinopathy testing will not be routinely performed on the "repeat" or "second test" specimen unless a repeat test is requested by the Alabama Newborn Screening Laboratory.**

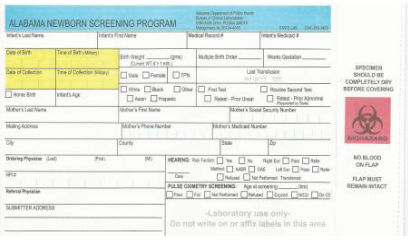
3. If you are aware that an infant was born outside of a hospital setting for any reason, please ensure that the infant receives an initial newborn screen and newborn hearing screen immediately. Please mark the box indicating "first test." Please use the audio form on page 41 for submitting newborn hearing results.

4. If an infant was born outside of the state of Alabama, the physician should request the newborn screening results, including hemoglobinopathy, from the state in which the infant was born.

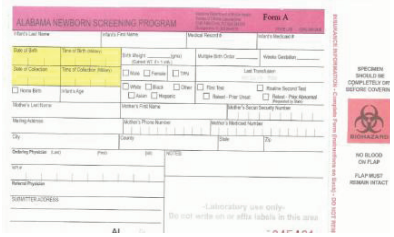




Newborn Screening Specimen Collection Tips



Newborn screens can have a dramatic impact on the welfare of the infant and the family. It is important to understand the significance of screening both from a medical outcome and a legal liability standpoint.



1. Storage of the filter paper both pre-use and post-use can be very important. If the paper is stored in a dry, hot environment such as an unventilated warehouse it will affect the performance of the paper. Always try to store paper at room temperature and room humidity. Post-use storage should be in keeping with NBS lab guidance (©ID Biological Systems Report).

2. The type of lancet used can have a definite effect on the specimen collected. The "switch blade" type lancet achieves better blood flow than the puncture type. This could make a difference in your blood collection (©ID Biological Systems Report).

3. Only allow <u>well trained</u> individuals to collect newborn screening blood in order to reduce unsatisfactory specimens (Alabama NBS Coordinator Conference 2009).

4. Track the performance of these collectors and re-train or substitute as necessary if unsatisfactory or invalid results occur (Alabama NBS Coordinator Conference 2009).

5. **<u>Perform a quality control inspection</u>** of all specimens before mailing them to the state lab. At a minimum check for the following:
   - Complete and correct demographic information. **Any corrections should be legible and initialed**.
   - Complete and correct newborn hearing test information.
   - Record the name of the person that collected the sample.
   - Inspect the blood spots for specimen quality and quantity before mailing.
   - Allow specimens to dry first and then review a second time prior to mailing. A specimen may appear uniform when wet but when dry may reveal uneven saturation (dark spots).
   - Confirm results are received on each specimen submitted.

If you believe you are having issues with specimen collection, please contact the NBS Nurse Educator at 334-206-5729 or the NBS State Health Laboratory at 334-260-3400. You may also refer to the Clinical and Laboratory Standards Institute® (CLSI) Screening Collection Manual (copies provided to all birthing centers).

***Remember**: Collection technique will not improve overnight. It takes practice to become proficient with newborn screening specimen collection.*

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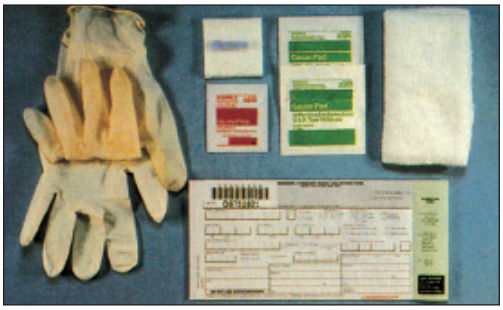
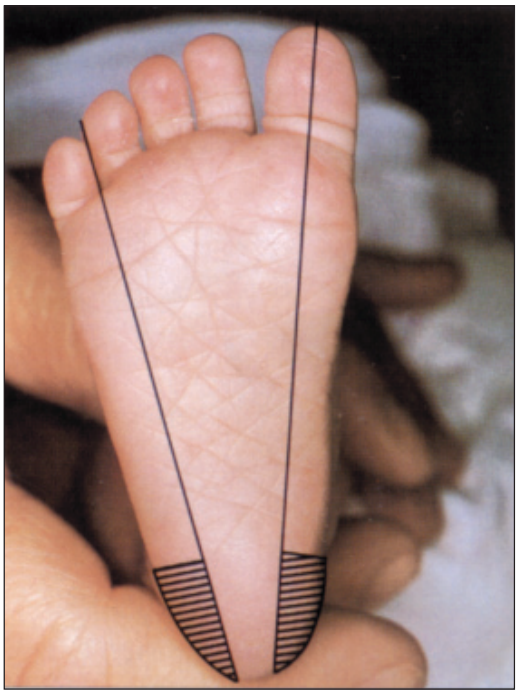
Neonatal Screening

Blood Specimen Collection and Handling Procedure

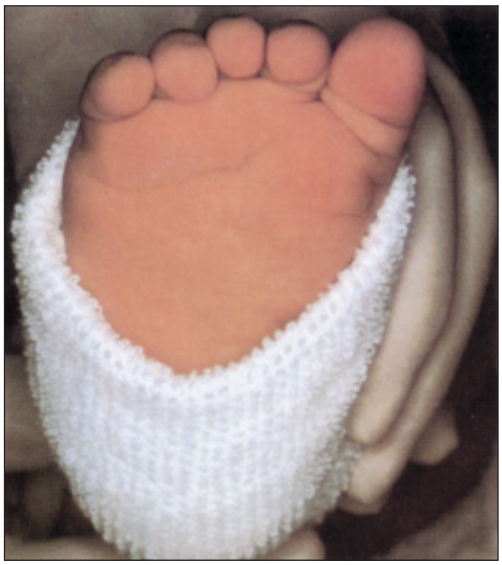


1 Necessary equipment: sterile lancet with tip approximately 2.0 mm, sterile alcohol prep, sterile gauze pads, soft cloth, blood collection form, gloves.

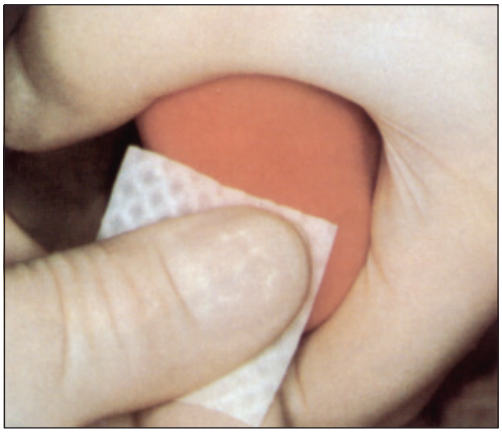
2 Complete ALL information. Do not contaminate filter paper circles by allowing the circles to come into contact with spillage or by touching before or after blood collection. Keep "SUBMITTER COPY" if applicable.



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

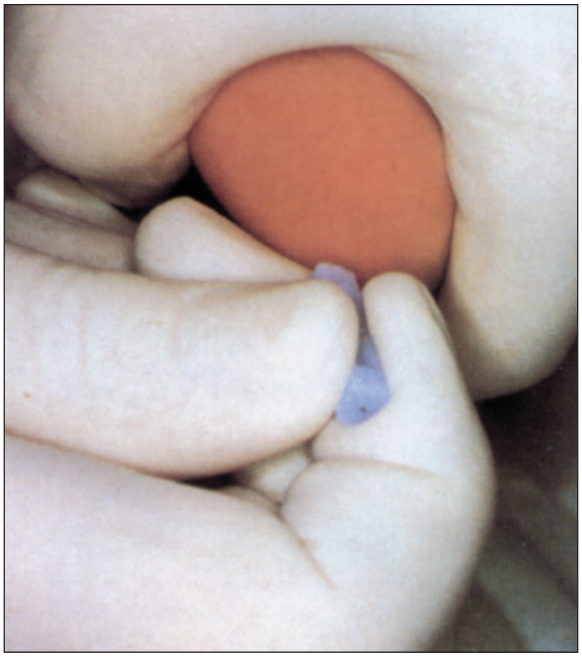


4 Warm site with soft cloth, moistened with warm water up to 41° C, for three to five minutes.

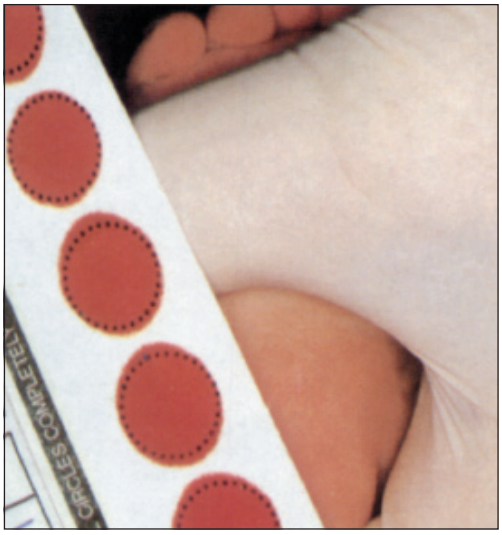


5 Cleanse site with alcohol prep. Wipe DRY with sterile gauze pad.

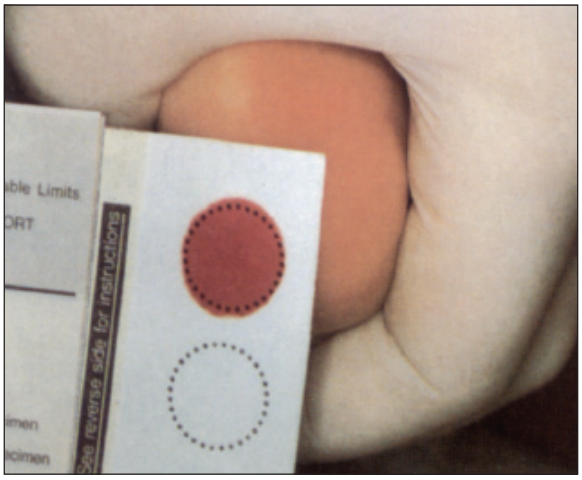
Over



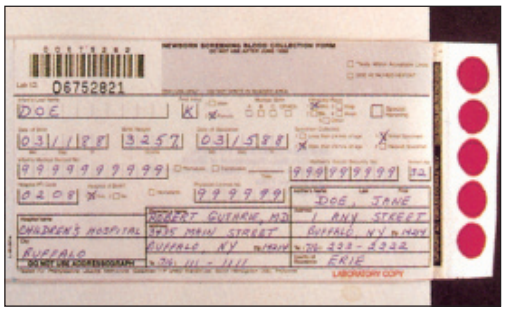
6 Puncture heel. Wipe away first blood drop with sterile gauze pad. Allow another LARGE blood drop to form.



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.



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.



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.

North America – Whatman Inc. • Tel: 1-800-WHATMAN • Tel: 1-973-245-8300 • Fax: 1-973-245-8329 • E-mail: info@whatman.com
Europe – Whatman International Ltd • Tel: +44 (0) 1622 676670 • Fax: +44 (0) 1622 691425 • E-mail: information@whatman.com
Japan – Whatman Japan KK • Tel: +81 (0) 3 5215 1240 • Fax: +81 (0) 3 5215 1245 • E-mail: japaninfo@whatman.com
Asia Pacific – Whatman Asia Pacific Pte Ltd • Tel: +65 6534 0138 • Fax: +65 6534 2166 • E-mail: wap@whatman.com

Simple Spot Check

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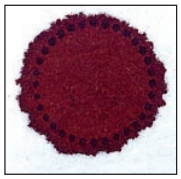
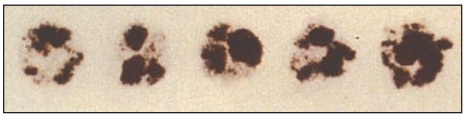
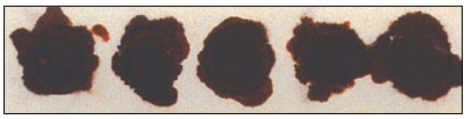
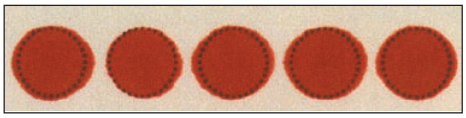
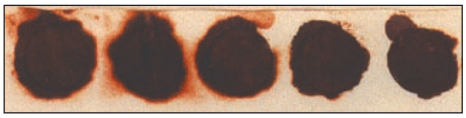
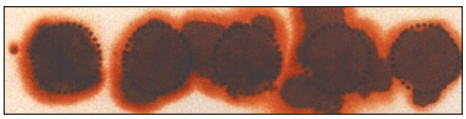
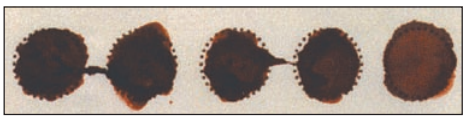
Valid specimen:



Allow a sufficient quantity of blood to soak through to completely fill the preprinted circle on the filter paper. Fill all required circles with blood. Do not layer successive drops of blood or apply blood more than once in the same collection circle. Avoid touching or smearing spots.

Invalid specimen:	Possible causes:
 **1. Specimen quantity insufficient for testing.**	• Removing filter paper before blood has completely filled circle or before blood has soaked through to second side. • Applying blood to filter paper with a capillary tube. • Allowing filter paper to come into contact with gloved or ungloved hands or substances such as hand lotion or powder, either before or after blood specimen collection.
 **2. Specimen appears scratched or abraded.**	• Applying blood with a capillary tube or other device.
 **3. Specimen not dry before mailing.**	• Mailing specimen before drying for a minimum of four hours.
 **4. Specimen appears supersaturated.**	• Applying excess blood to filter paper, usually with a device. • Applying blood to both sides of filter paper.
 **5. Specimen appears diluted, discolored or contaminated.**	• Squeezing or "milking" of area surrounding the puncture site. • Allowing filter paper to come into contact with gloved or ungloved hands or substances such as alcohol, formula, antiseptic solutions, water, hand lotion or powder, etc., either before or after blood specimen collection. • Exposing blood spots to direct heat.
 **6. Specimen exhibits serum rings.**	• Not wiping alcohol from puncture site before making skin puncture. • Allowing filter paper to come into contact with alcohol, hand lotion, etc. • Squeezing area surrounding puncture site excessively. • Drying specimen improperly. • Applying blood to filter paper with a capillary tube.
 **7. Specimen appears clotted or layered.**	• Touching the same circle on filter paper to blood drop several times. • Filling circle on both sides of filter paper.
 **8. No blood.**	• Failure to obtain blood specimen.

North America – Whatman Inc. • Tel: 1-800-WHATMAN • Tel: 1-973-245-8300 • Fax: 1-973-245-8329 • E-mail: info@whatman.com
Europe – Whatman International Ltd • Tel: +44 (0) 1622 676670 • Fax: +44 (0) 1622 677011 • E-mail: information@whatman.com
Japan – Whatman Japan KK • Tel: +81 (0) 3 5215 1240 • Fax: +81 (0) 3 5215 1245 • E-mail: japaninfo@whatman.com
Asia Pacific – Whatman Asia Pacific Pte Ltd • Tel: +65 6534 0138 • Fax: +65 6534 2166 • E-mail: wap@whatman.com

Information provided by The New York State Department of Health.

BUREAU OF CLINICAL LABORATORIES
SHARON P. MASSINGALE, PH.D., HCLD(ABB) ♦ DIRECTOR

Alabama Newborn Screening Program
Reorder Form

In order to assure that you have an adequate supply of newborn screening materials available, complete this form and mail or fax it to the State Health Laboratory at the address below when your stock has reached a **2-4 week** supply.

ALABAMA DEPARTMENT OF PUBLIC HEALTH
Bureau of Clinical Laboratories
Newborn Screening Division
8140 AUM Drive, Zip 36117-7001
P.O. Box 244018, Zip 36124-4018
Montgomery, AL

FAX (334) 260-3439

Name of Hospital or Doctor: _____

Street/Shipping Address **ONLY No P.O. Box:** _____

City, State, and Zip Code: _____

Telephone Number: _____

Signature and Title: _____

Number of "A" (first test) Newborn Screening Kits Requested: _____

*Note "A" forms are sent to Hospitals and Birthing Centers only.

Number of "B" (second test) Newborn Screening Forms Requested: _____

Please indicate the number of newborn infants that you screen per month: _____

_____

NOTE: All orders will be shipped within 5 working days of receipt. Please plan your orders accordingly. We cannot make emergency shipments.

STATE OF ALABAMA
DEPARTMENT OF PUBLIC HEALTH
DONALD E. WILLIAMSON, M.D. • STATE HEALTH OFFICER

BUREAU OF CLINICAL LABORATORIES
SHARON P. MASSINGALE, PH.D., HCLD(ABB) • DIRECTOR

MEMORANDUM

TO: Health Care Providers

FROM: Newborn Screening Division
Bureau of Clinical Laboratories

SUBJECT: Newborn Screening Provider Update

In order to provide more efficient service in providing newborn screening forms we are updating our provider list. It would be of a great assistance to us if you would fill out the following information and return it as soon as possible to:

ALABAMA DEPARTMENT OF PUBLIC HEALTH
Bureau of Clinical Laboratories
Newborn Screening Division
P.O. Box 244018
Montgomery, AL 36124-4018
FAX (334) 260-3439

Thank you for your prompt attention to this matter.

Group or Name of Practice: _____

Street/Shipping Address **ONLY** No P.O. Box: _____

City, State, and Zip Code: _____

Telephone Number: _____

Approximate Number of Specimens per Month: _____

NAMES OF **ALL** PHYSICIANS THAT SEND NEWBORN SCREENING SPECIMENS: (Please include NPI#)

_____ NPI#

_____ NPI#

_____ NPI#

_____ NPI#

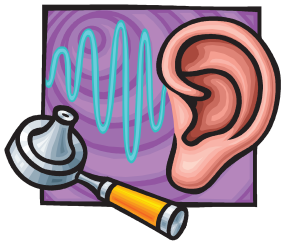
_____ NPI#

_____ NPI#

Section 3 - Newborn Hearing Screening

Alabama's Listening!
UNIVERSAL NEWBORN HEARING SCREENING

Newborn Hearing Screening FAQ



What is hearing loss?

There are two main types of hearing loss:
1. Conductive hearing loss – occurs when sound cannot enter into the inner ear. This may be caused by wax buildup, fluid in the ear, or structural abnormalities. It can usually be corrected with medical or surgical intervention.

2. Sensorineural hearing loss – occurs when there is damage to the inner ear. This may be caused by diseases, birth injury, toxic drugs, viruses, or genetic syndromes.

In addition, there are various degrees of hearing loss. They include:

- mild hearing loss
- moderate hearing loss
- severe hearing loss
- profound hearing loss

It is important to note that milder hearing losses or hearing losses that affect only one ear may not be apparent.

Why should a baby's hearing be screened?

The first two years of a baby's life are critical for learning speech and language. Thus, it is important to diagnose hearing problems early because a hearing loss could affect a baby's speech and language development. In addition, early detection makes talking, learning, and adjusting to hearing devices easier.

How is the hearing screen performed?

There are two types of screening methods that may be used. Both tests are very safe, take only minutes to perform, and are non-invasive. Most babies sleep through the hearing screening.

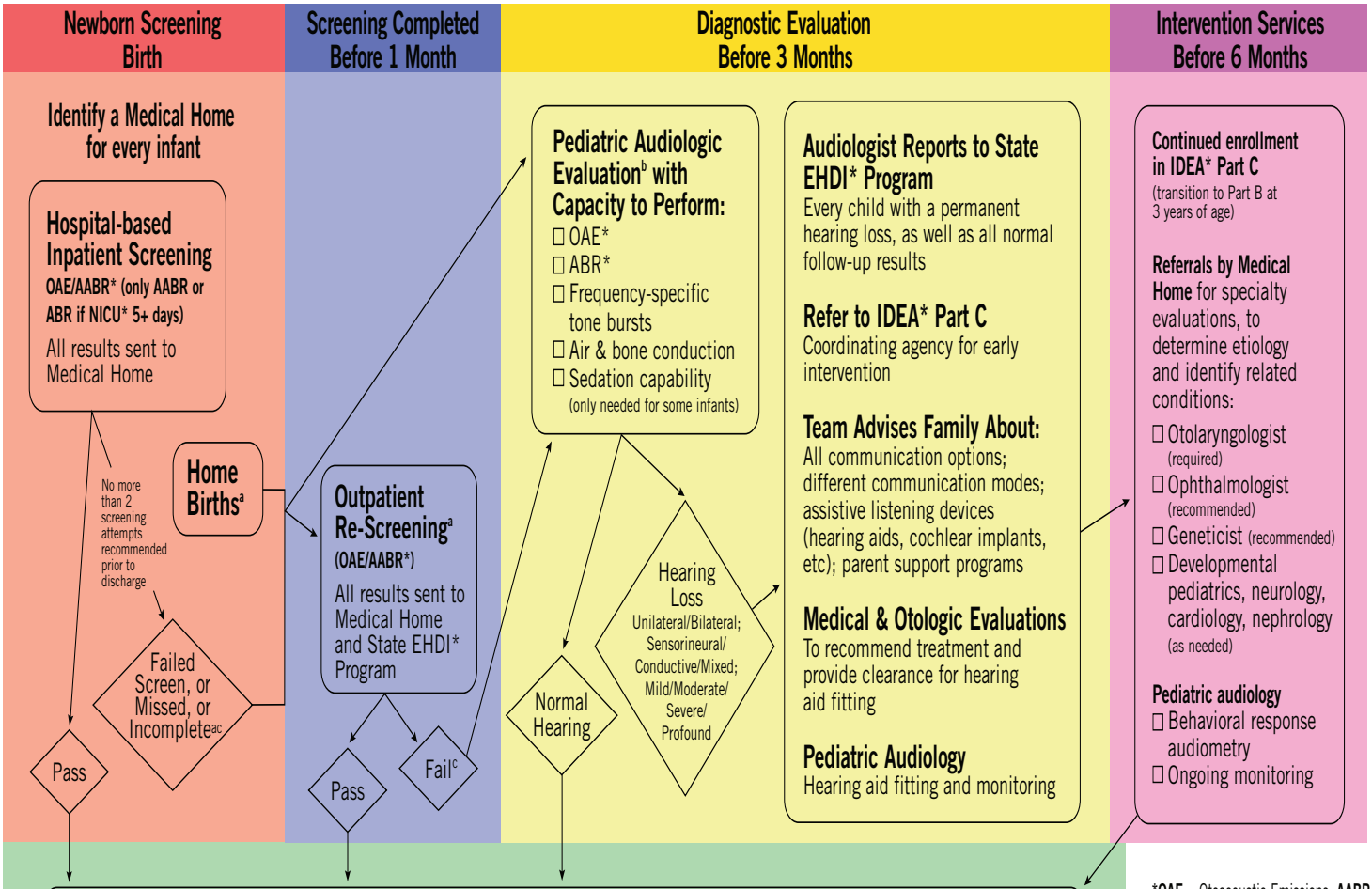
1. Auditory Brainstem Response (ABR) – determines the infant's ability to hear soft sounds normally by inserting miniature earphones and attaching electrodes to measure brain-wave responses to the sound. **This diagnostic testing is recommended by the Joint Committee on Infant Hearing (JCIH) for high risk newborns admitted to the NICU greater than five days and should be completed as a second test method if an infant is initially tested with ABR**.

2. Otoacoustic emissions (OAE) – measures inner ear function by inserting a miniature microphone in the ear canal via a soft probe tip and measuring tones from the ear by sending responses to a special computer.

What if a baby does not pass the hearing screen?

If a baby does not pass the first hearing screen then an attempt should be made to repeat the screen before a baby goes home, or it may be scheduled after going home. **The JCIH recommends that all testing be completed by three months of age, and infants with hearing loss be enrolled in appropriate intervention services as early as possible, but no later than six months of age**.

Early Hearing Detection and Intervention (EHDI) Guidelines for Pediatric Medical Home Providers



Ongoing Care of All Infants[d]; Coordinated by the Medical Home Provider

- **Provide parents with information about hearing, speech, and language milestones**
- **Identify and aggressively treat middle ear disease**
- **Provide vision screening (and referral when indicated) as recommended in the AAP "Bright Futures Guidelines, 3rd Ed."**
- **Provide ongoing developmental screening (and referral when indicated) per the AAP "Bright Futures Guidelines, 3rd Ed."**
- **Refer promptly for audiology evaluation when there is any parental concern‡ regarding hearing, speech, or language development**
- **Refer for audiology evaluation (at least once before age 30 months) infants who have any risk indicators for later-onset hearing loss:**
  - Family history of permanent childhood hearing loss‡
  - Neonatal intensive care unit stay of more than 5 days duration, or any of the following (regardless of length of stay): ECMO‡, mechanically-assisted ventilation, ototoxic medications or loop diuretics, exchange transfusion for hyperbiliruinemia
  - In utero infections such as cytomegalovirus‡, herpes, rubella, syphilis, and toxoplasmosis
  - Postnatal infections associated with hearing loss‡, including bacterial and viral meningitis
  - Craniofacial anomalies, particularly those that involve the pinna, ear canal, ear tags, ear pits, and temporal bone anomalies
  - Findings suggestive of a syndrome associated with hearing loss (Waardenburg, Alport, Jervell and Lange-Nielsen, Pendred)
  - Syndromes associated with progressive or delayed-onset hearing loss‡ (neurofibromatosis, osteopetrosis, Usher Syndrome)
  - Neurodegenerative disorders‡ (such as Hunter Syndrome) or sensory motor neuropathies (such as Friedreich's ataxia and Charcot Marie Tooth disease)
  - Head trauma, especially basal skull/temporal bone fracture that requires hospitalization
  - Chemotherapy‡

‡Denotes risk indicators of greater concern. Earlier and/or more frequent referral should be considered.

***OAE** = Otoacoustic Emissions, **AABR** = Automated Auditory Brainstem Response, **ABR** = Auditory Brainstem Response, **EHDI** = Early Hearing Detection and Intervention, **IDEA** = Individuals with Disabilities Education Act, **NICU** = Newborn Intensive Care Unit, **AAP** = American Academy of Pediatrics

Notes:

(a) In screening programs that do not provide Outpatient Screening, infants will be referred directly from Inpatient Screening to Pediatric Audiologic Evaluation. Likewise, infants at higher risk for hearing loss (or loss to follow-up) also may be referred directly to Pediatric Audiology.

(b) Part C of IDEA* may provide diagnostic audiologic evaluation services as part of Child Find activities.

(c) Even infants who fail screening in only one ear should be referred for further testing of both ears

(d) Includes infants whose parents refused initial or follow-up hearing screening.

February 2010 - American Academy of Pediatrics Task Force for Improving Newborn Hearing Screening, Diagnosis and Intervention (www.medicalhomeinfo.org)

ALABAMA NEWBORN HEARING RE-SCREENING RESULTS
PHONE 334.206.2944 FAX 334.206.3791
USE THIS SIDE FOR FOLLOW-UP RE-SCREEN
BOTH EARS MUST BE TESTED

Alabama's Listening!
UNIVERSAL NEWBORN HEARING SCREENING

NEWBORN'S NAME	DATE OF BIRTH
HOSPITAL OF BIRTH	MEDICAL ID #
MOTHER'S NAME	PHONE
ADDRESS	
PRIMARY CARE PHYSICIAN	PHONE
ADDRESS	

BIRTH	HEARING SCREEN PERFORMED AT BIRTH FACILITY	Inpatient Screen Date_____ Right Ear: ☐Pass ☐ Refer ☐ Not Tested Left Ear: ☐ Pass ☐ Refer ☐Not Tested Method: ☐ AABR ☐ OAE ☐ TEOAE ☐DPOAE	Infants who fail initial OAE screen can have an OAE or AABR re-screen. Infants who fail initial AABR screen must have an AABR re-screen.
BEFORE 1 MONTH	REPEAT SCREENING RESULTS Inpatient ☐ Outpatient ☐	DATE SCREENED: Right Ear: ☐Pass ☐ Refer ☐ Not Tested Left Ear: ☐ Pass ☐ Refer ☐Not Tested Method: ☐ AABR ☐ OAE ☐ TEOAE ☐DPOAE	RISK FACTORS FOR DELAYED HEARING LOSS ☐ NICU >48 hrs ☐ Received ototoxic medications ☐ Transfused ☐ Other_____ _____ If any present refer for audiology evaluation at least once prior to 30 months of age.

TEST SITE NAME	PHONE	FAX
ADDRESS		

COMMENTS/FOLLOW-UP
_____
_____
_____

The completed form should be returned as soon as the hearing re-screen/initial diagnostic audiological evaluation is completed, but no later than 4 weeks from the date of the referral. Fax to the Newborn Hearing Screening Program at 334-206-3791 .

NBS.Hearing Re-Screen.2015

Alabama Newborn Screening Program

ALABAMA NEWBORN HEARING RE-SCREENING RESULTS
PHONE 334.206.2944 FAX 334.206.3791
USE THIS SIDE FOR DIAGNOSTIC TESTING ONLY
BOTH EARS MUST BE TESTED

Alabama's Listening!
UNIVERSAL NEWBORN HEARING SCREENING

NEWBORN'S NAME	DATE OF BIRTH
HOSPITAL OF BIRTH	MEDICAL ID #
MOTHER'S NAME	PHONE
ADDRESS	

TEST SITE

Name	Phone	Fax
Address		

Before 3 Months	**Pediatric Diagnostic Audiology Evaluation**	**DIAGNOSTIC TEST DATE**_____ **METHOD:** ☐ABR ☐AABR ☐OAE ☐ TEOAE ☐DPOAE ☐ Normal Hearing ☐ Hearing Loss Confirmed (Please Complete Section Below)
Before 6 Months	**Enrollment in Early Intervention**	Date of Referral to EI_____ Enrollment Date_____ Medical Referral: ☐Otolaryngologist ☐Geneticist ☐Ophthalmologist ☐Pediatrician ☐ Other (specify)_____ Additional Audiology Services_____

		dB HL	SEVERITY/TYPE	Sensorineural	Conductive	Mixed	Unknown	Auditory Neuropathy
___UNILATERAL LOSS	RIGHT EAR	16 to 25	Slight					
		26 to 40	Mild					
		41 to 55	Moderate					
		56 to 70	Moderately Severe					
		71 to 90	Severe					
		91+	Profound					
			Unknown Severity					
___BILATERAL LOSS	LEFT EAR	dB HL	SEVERITY/TYPE	Sensorineural	Conductive	Mixed	Unknown	Auditory Neuropathy
		16 to 25	Slight					
		26 to 40	Mild					
		41 to 55	Moderate					
		56 to 70	Moderately Severe					
		71 to 90	Severe					
		91 +	Profound					
			Unknown Severity					

COMMENTS/FOLLOW UP

_____

_____

_____

The completed form should be returned as soon as the hearing re-screen/initial diagnostic audiological evaluation is completed, but no later than 4 weeks from the date of the referral. Fax to the Newborn Hearing Screening Program at 334-206-3791.

CRS Newborn Hearing Assessment Clinics

CRS is part of the Alabama Department of Rehabilitation Services (ADRS). There are multiple sites across the state offering the preferred follow-up screening method of AABR. Parents may choose the site that is most convenient for them. Please see the list below to obtain an appointment for the CRS Newborn Hearing Assessment Clinic. This clinic is for newborns who fail the initial hearing screen. For other hearing providers in Alabama that offer this method of hearing screening or OAE hearing screening, please see our website at www.adph.org/newbornscreening.

Calhoun County – Anniston CRS 1910 Coleman Road Anniston, AL 36207 Phone: 256-240-8801 or 1-800-289-9533 Counties: Calhoun, Cherokee, Clay, Cleburne, St. Clair, Talladega	**Jefferson County - Homewood CRS** 234 Goodwin Crest Drive Birmingham, AL 35209 Phone: 205-290-4550 or 1-888-430-7423 Counties: Cullman, Jefferson, Shelby, Walker
Covington County – Andalusia CRS 1082 Village Square Drive, Suite 2 Andalusia, AL 36420 Phone: 334-222-5558 or 1-800-723-8064 Counties: Butler, Conecuh, Covington, Crenshaw	**Madison County – Huntsville CRS** 3000 Johnson Road Huntsville, AL 35805 Phone: 256-650-1701 or 1-800-283-8140 Counties: Jackson, Limestone, Madison, Marshall, Morgan
Dallas County – Selma CRS 2906 Citizens Parkway Selma, AL 36701 Phone: 334-872-8422 or 1-800-967-6876 Counties: Dallas, Marengo, Perry, Wilcox	**Montgomery County – Montgomery CRS** 602 South Lawrence Street Montgomery, AL 36104 Phone: 334-293-7500 or 1-800-568-9034 Counties: Autauga, Bullock, Chilton, Coosa, Elmore, Lowndes, Montgomery, Pike
Etowah County – Gadsden CRS 1100 George Wallace Drive Gadsden, AL 35903 Phone: 256-547-8653 or 1-800-289-1353 Counties: Blount, Dekalb, Etowah	**Mobile County – Mobile CRS** 1610 Center Street, Suite A Mobile, AL 36604 Phone: 251-432-4560 or 1-800-879-8163 Counties: Baldwin, Escambia, Mobile
Houston County – Dothan CRS 795 Ross Clark Circle NE, Suite 3 Dothan, AL 36303 Phone: 334-699-6600 or 1-800-677-9123 Counties: Barbour, Coffee, Dale, Geneva, Henry, Houston	**Tuscaloosa County – Tuscaloosa CRS** 1110 Dr. Edward Hillard Drive P.O. Drawer 2817 Tuscaloosa, AL 35403 Phone: 205-759-1279 or 1-800-723-0490 Counties: Bibb, Fayette, Greene, Hale, Lamar, Pickens, Sumter, Tuscaloosa

Alabama Newborn Hearing Provider Directory

Autauga County

Center for Advanced Therapy
635 McQueen Smith Road, Suite D • Prattville, AL 36067
Phone: 334-358-6501
Medicaid Accepted: Unknown, call to verify
Services Provided: OAE screening for infants

Children's Rehab Services (CRS) - Montgomery
602 S. Lawrence Street • Montgomery, AL 36104
Phone: 334-293-7118 • www.rehab.alabama.gov
Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Baldwin County

Alabama Hearing and Balance Associates,Inc.
113 Fern Avenue • Foley, Alabama 36535
Phone: 251-970-3277
www.hearingandbalance.net
Medicaid Accepted: Yes
Services Provided: OAE and diagnostic audiology services for infants

Children's Rehab Services (CRS) - Mobile
1610 Center Street, Suite A • Mobile, AL 36604
Phone: 251-432-4560 or 1-800-568-9034
www.rehab.alabama.gov
Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Coastal Hearing and Balance LLC, d/b/a Ascent Audiology and Hearing
105 Lottie Lane, Suite A • Fairhope, Alabama 36532
Phone: 251-990-0535
www.ascentaudiologyfairhope.com
Medicaid Accepted: Yes
Services Provided: OAE screening for infants and Early Intervention Services for infants and children with hearing loss to include hearing aid amplification and wireless systems.

Baldwin County

Eastern Shore Ear, Nose, & Throat (ENT)
188 Hospital Drive • Fairhope, Alabama 36532
Phone: 251-928-0300
Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants

Naro Audiology & Hearing Solution - Bay Minette
157 A N Hoyle Avenue • Bay Minette, Alabama 36535
Phone: 251-937-8731
Medicaid Accepted: Yes
Services Provided: OAE and AABR screening for infants

Barbour County

Children's Rehab Services (CRS) - Houtson
795 Ross Clark Circle NE, Suite 3 • Dothan, AL 36303
Phone: 334-699-6600 or 1-800-677-9123
www.rehab.alabama.gov
Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Bibb County

Children's Rehab Services (CRS) - Tuscaloosa
1110 Dr. Edward Hillard Drive • P.O. Box 2817
Tuscaloosa, AL 35403
Phone: 205-759-1279 or 1-800-723-0490
www.rehab.alabama.gov
Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Alabama Newborn Hearing Provider Directory

Blount County

Children's Rehab Services (CRS) - Etowah
1100 George Wallace Drive • Gadsden, AL 35903
Phone: 256-547-8653 or 1-800-289-1353
www.rehab.alabama.gov

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Bullock County

Children's Rehab Services (CRS) - Montgomery
602 S. Lawrence Street • Montgomery, AL 36104
Phone: 334-293-7118 • www.rehab.alabama.gov

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Butler County

Children's Rehab Services (CRS) - Covington
1082 Village Square Drive, Suite 2 • Andalusia, AL 36420
Phone: 334-222-5558 • 1-800-723-8064
www.rehab.alabama.gov

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Calhoun County

Children's Rehab Services (CRS) - Anniston
1910 Coleman Road • Anniston, AL 35207
Phone: 256-240-8801 or 1-800-289-9533
www.rehab.alabama.gov

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification, wireless systems, speech therapy, care coordination and treatment planning.

ENT/Facial Plastic Surgery
901 Leighton Avenue, Suite 506 • Anniston, AL 36207
Phone: 256-238-0200

Medicaid Accepted: Yes
Services Provided: OAE and diagnostic audiology services for infants.

Cherokee County

Children's Rehab Services (CRS) - Anniston
1910 Coleman Road • Anniston, AL 35207
Phone: 256-240-8801 or 1-800-289-9533
www.rehab.alabama.gov

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification, wireless systems, speech therapy, care coordination and treatment planning.

Chilton County

Children's Rehab Services (CRS) - Montgomery
602 S. Lawrence Street • Montgomery, AL 36104
Phone: 334-293-7118 • www.rehab.alabama.gov

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Alabama Newborn Hearing Provider Directory

Clay County

Children's Rehab Services (CRS) - Anniston
1910 Coleman Road • Anniston, AL 35207
Phone: 256-240-8801 or 1-800-289-9533
www.rehab.alabama.gov

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification, wireless systems, speech therapy, care coordination and treatment planning.

Cleburne County

Children's Rehab Services (CRS) - Anniston
1910 Coleman Road • Anniston, AL 35207
Phone: 256-240-8801 or 1-800-289-9533
www.rehab.alabama.gov

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification, wireless systems, speech therapy, care coordination and treatment planning.

Coffee County

Children's Rehab Services (CRS) - Houtson
795 Ross Clark Circle NE, Suite 3 • Dothan, AL 36303
Phone: 334-699-6600 or 1-800-677-9123
www.rehab.alabama.gov

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Coffee County (cont.)

Southeastern ENT
107 E. Watts Street • Enterprise, AL 36331
Phone: 334-393-6837

Medicaid Accepted: Yes
Services Provided: OAE and AABR screening for infants

Conecuh County

Children's Rehab Services (CRS) - Covington
1082 Village Square Drive, Suite 2 • Andalusia, AL 36420
Phone: 334-222-5558 • 1-800-723-8064
www.rehab.alabama.gov

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Coosa County

Children's Rehab Services (CRS) - Montgomery
602 S. Lawrence Street • Montgomery, AL 36104
Phone: 334-293-7118 • www.rehab.alabama.gov

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Covington County

Children's Rehab Services (CRS) - Covington
1082 Village Square Drive, Suite 2 • Andalusia, AL 36420
Phone: 334-222-5558 • 1-800-723-8064

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Alabama Newborn Hearing Provider Directory

Crenshaw County

Children's Rehab Services (CRS) - Covington
1082 Village Square Drive, Suite 2 • Andalusia, AL 36420
Phone: 334-222-5558 • 1-800-723-8064
www.rehab.alabama.gov
Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Cullman County

Children's Rehab Services (CRS) - Jefferson
234 Goodwin Crest Drive • Homewood, AL 35209
Phone: 205-290-4550 • 1-888-430-7423
www.rehab.alabama.gov
Medicaid Accepted: Yes
Services Provided: OAE, AABR, and ABR diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification, care coordination, information, and referrals.

ENT Associates of Alabama, P.C. - Cullman
1948 Alabama Highway 157, Suite 410 • Cullman, AL 35058
Phone: 256-737-0368
www.entalabama.com
Medicaid Accepted: Yes
Services Provided: OAE screening for infants

Johnson Hearing Services, P.C.
1900 Main Avenue SW, Suite 1 • Cullman, AL 35055
Phone: 256-841-0930
www.johnsonhearingservices.com
Medicaid Accepted: Yes
Services Provided: OAE and AABR screening for infants. Early Intervention Service for infants and children with hearing loss to include hearing aid amplification.

Dale County

Children's Rehab Services (CRS) - Houtson
795 Ross Clark Circle NE, Suite 3 • Dothan, AL 36303
Phone: 334-699-6600 or 1-800-677-9123
www.rehab.alabama.gov
Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Lyster Army Health Clinic/Audiology Clinic
301 Andrews Avenue • Fort Rucker, AL 36362
Phone: 334-255-7185
www.rucker.amedd.army.mi
Medicaid Accepted: Yes
Services Provided: OAE screening for infants

Dallas County

Children's Rehab Services (CRS) - Dallas
2906 Citizens Parkway • Selma, AL 36701
Phone: 334-872-8422 or 1-800-967-6876
www.rehab.alabama.gov
Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

ENT Consultants
731 Dallas Avenue • Selma, AL 36701
Phone: 334-872-4778
www.rehab.alabama.gov
Medicaid Accepted: Yes
Services Provided: OAE screening for infants

Alabama Newborn Hearing Provider Directory

DeKalb County

Children's Rehab Services (CRS) - Etowah
1100 George Wallace Drive • Gadsden, AL 35903
Phone: 256-547-8653 or 1-800-289-1353
www.rehab.alabama.gov

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

DeKalb ENT
423 Medical Center Drive, SW • Fort Payne, AL 35968
Phone: 256-844-8144

Medicaid Accepted: Unknown, call to verify
Services Provided: OAE screening for infants

Elmore County

Children's Rehab Services (CRS) - Montgomery
602 S. Lawrence Street • Montgomery, AL 36104
Phone: 334-293-7118 • www.rehab.alabama.gov

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Escambia County

Children's Rehab Services (CRS) - Mobile
1610 Center Street, Suite A • Mobile, AL 36604
Phone: 251-432-4560 or 1-800-568-9034
www.rehab.alabama.gov

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Escambia County

Naro Audiology & Hearing Solution - Brewton
1305 McMillan Avenue • Brewton, AL 36426
Phone: 251-867-7711

Medicaid Accepted: Yes
Services Provided: OAE and AABR screening for infants

Etowah County

Children's Rehab Services (CRS) - Etowah
1100 George Wallace Drive • Gadsden, AL 35903
Phone: 256-547-8653 or 1-800-289-1353

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Northeast Alabama Audiology Clinic
417 South 4th Street • Gadsden, AL 35901
Phone: 256-543-8899

Medicaid Accepted: Unknown, call to verify
Services Provided: OAE screening for infants during Thursday clinic

Fayette County

Children's Rehab Services (CRS) - Tuscaloosa
1110 Dr. Edward Hillard Drive • P.O. Box 2817
Tuscaloosa, AL 35403
Phone: 205-759-1279 or 1-800-723-0490
www.rehab.alabama.gov

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Alabama Newborn Hearing Provider Directory

Geneva County

Children's Rehab Services (CRS) - Houtson
795 Ross Clark Circle NE, Suite 3 • Dothan, AL 36303
Phone: 334-699-6600 or 1-800-677-9123
www.rehab.alabama.gov
Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Greene County

Children's Rehab Services (CRS) - Tuscaloosa
1110 Dr. Edward Hillard Drive • P.O. Box 2817
Tuscaloosa, AL 35403
Phone: 205-759-1279 or 1-800-723-0490
www.rehab.alabama.gov
Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Hale County

Children's Rehab Services (CRS) - Tuscaloosa
1110 Dr. Edward Hillard Drive • P.O. Box 2817
Tuscaloosa, AL 35403
Phone: 205-759-1279 or 1-800-723-0490
www.rehab.alabama.gov
Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Henry County

Children's Rehab Services (CRS) - Houtson
795 Ross Clark Circle NE, Suite 3 • Dothan, AL 36303
Phone: 334-699-6600 or 1-800-677-9123
www.rehab.alabama.gov
Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Houston County

Children's Rehab Services (CRS) - Houtson
795 Ross Clark Circle NE, Suite 3 • Dothan, AL 36303
Phone: 334-699-6600 or 1-800-677-9123
www.rehab.alabama.gov
Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Hearing Associates of Dothan, LLC
1891 Honeysuckle Road • Dothan, AL 36305
Phone: 334-702-4327
www.hearingassociatesdothan.com
Medicaid Accepted: Yes
Services Provided: OAE screening for infants and Early Intervention Services for infants and children with hearing loss to include hearing aid amplification and wireless systems

Jackson County

Children's Rehab Services (CRS) - Madison
3000 Johnson Road • Huntsville, AL 35805
Phone: 256-650-1701 or 1-800-283-8140
www.rehab.alabama.gov
Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification, care coordination, information and referrals.

Alabama Newborn Hearing Provider Directory

Jackson County (cont.)

Scottsboro ENT
406 B Taylor Street • Scottsboro, AL 35768
Phone: 256-574-6100

Medicaid Accepted: Yes
Services Provided: Diagnostic ABR only

Jefferson County

Angela P. Naccari, M.S., CCC-A, LSLS Cert. AVT
4624 Summit Circle • Hoover, AL 35226
Phone: 205-542-2112

Medicaid Accepted: No
Services Provided: Early Intervention Services for infants and children with hearing loss to include auditory verbal therapy and auditory oral therapy

Birmingham Ear, Nose, and Throat Group, P.C.
2700 10th Avenue S., Suite 501 • Birmingham, AL 35205
Phone: 205-212-3310

Medicaid Accepted: Yes
Services Provided: OAE and ABR diagnostic audiology services for infants

Birmingham Hearing and Balance Center
2700 10th Avenue S., Suite 502 • Birmingham, AL 35205
Phone: 205-933-2951

Medicaid Accepted: No
Services Provided: OAE screening for infants and Early Intervention Services for infants and children with hearing loss to include hearing aid amplification, cochlear implant assessment and management

Birmingham Speech and Hearing Associates
4 Office Park Circle, Suite 301 • Birmingham, AL 35223
Phone: 205-871-3878
www.birminghamspeechandhearing.com

Medicaid Accepted: Yes
Services Provided: Early Intervention Services for infants and children with hearing loss to include hearing aid amplification, wireless systems, speech therapy, and services for hearing impaired in local school systems

Jefferson County (cont.)

Children's Hospital of Alabama
Children's Main Location:
1600 7th Avenue South • Birmingham, AL 35233
Phone: 205-638-9149 or 205-638-7500
Children's HEAR Center:
1208 3rd Avenue South • Birmingham, AL 35233
Phone: 205-638-9149 or 205-638-7500
Children's South:
1940 Elmer J. Bissell Road • Birmingham, AL 35243
Phone: 205-638-9149 or 205-638-7500
www.childrensal.org/HEARCenterHome

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and ABR diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification, cochlear implant assessment and management, wireless systems, speech therapy, and auditory verbal therapy.

Children's Rehab Services (CRS) - Jefferson
234 Goodwin Crest Drive • Homewood, AL 35209
Phone: 205-290-4550 • 1-888-430-7423
www.rehab.alabama.gov

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and ABR diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification, care coordination, information, and referrals.

Civitan Sparks Clinic
1720 2nd Avenue South, Suite 507, CH19
Birmingham, AL 35205
Phone: 205-934-5471
www.uab.edu/civitansparks

Medicaid Accepted: Yes
Services Provided: OAE screening and diagnostic audiology services for infants, and Early Intervention Services for infants and children with hearing loss to include hearing aid amplification

Alabama Newborn Hearing Provider Directory

Jefferson County (cont.)

ENT Associates of Alabama, P.C. - Gardendale
932 Sharit Avenue, Suite 101 • Gardendale, AL 35071
Phone: 205-631-8116 • www.entalabama.com

Medicaid Accepted: Yes
Services Provided: OAE screening for infants

ENT Associates of Alabama, P.C. - St. Vincent's
833 St. Vincent's Drive, Suite 402 • Birmingham, AL 35205
Phone: 205-933-9236 • www.entalabama.com

Medicaid Accepted: Yes
Services Provided: OAE screening for infants

ENT for KIDS Alabama
2807 Greystone Commercial Blvd, Suite 42
Birmingham, AL 35242
Phone: 205-874-9436 • FAX: 205-874-9438

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and ABR diagnostic testing for infants.

UAB Hearing Clinic
1717 6th Avenue South, RO 44 SRC
Birmingham, AL 35233
Phone: 205-934-4816

Medicaid Accepted: Yes
Services Provided: OAE and AABR screening for infant. AABR is done at UAB Hospital. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification and wireless systems.

Lamar County

Children's Rehab Services (CRS) - Tuscaloosa
1110 Dr. Edward Hillard Drive • P.O. Box 2817
Tuscaloosa, AL 35403
Phone: 205-759-1279 or 1-800-723-0490
www.rehab.alabama.gov

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Lauderdale County

Shoals Hearing Clinic, P.C.
2055 Seminary Street • Florence, AL 35630
Phone: 256-740-8383
www.drgresham.com

Medicaid Accepted: Yes
Services Provided: OAE and AABR screening for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification and wireless systems.

Lee County

All For Children
2290 Moores Mill Road, Suite 400 • Auburn, AL 36830
Phone: 334-209-2009
www.all-forchildren.com

Medicaid Accepted: Yes
Services Provided: OAE screening and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification, cochlear implant assessment/management, wireless systems, speech therapy, and auditory-verbal therapy.

Alabama Newborn Hearing Provider Directory

Lee County (cont.)

Auburn University Speech and Hearing Clinic
1199 Haley Center • Auburn, AL 36849
Phone: 334-844-9600
www.cla.auburn.edu/speechandhearingclinic.edu

Medicaid Accepted: Yes
Services Provided: OAE, ABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification and speech therapy.

Lowndes County

Children's Rehab Services (CRS) - Montgomery
602 S. Lawrence Street • Montgomery, AL 36104
Phone: 334-293-7500 • 1-800-568-9034
www.rehab.alabama.gov

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Limestone County

Children's Rehab Services (CRS) - Madison County
3000 Johnson Road • Huntsville, AL 35805
Phone: 256-650-1701 or 1-800-283-8140
www.rehab.alabama.gov

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification, care coordination, information and referrals.

Valley ENT Associates, P.C.
1005 West Market Street, Suite 8 • Athens, AL 35611
Phone: 256-233-1650
www.valleyentassociates.com

Medicaid Accepted: Unknown, call to verify
Services Provided: OAE screening for infants

Macon County

Auburn University Speech and Hearing Clinic
1199 Haley Center • Auburn, AL 36849
Phone: 334-844-9600
www.cla.auburn.edu/speechandhearingclinic.edu

Medicaid Accepted: Yes
Services Provided: OAE, ABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification and speech therapy.

Madison County

Children's Rehab Services (CRS) - Madison County
3000 Johnson Road • Huntsville, AL 35805
Phone: 256-650-1701 or 1-800-283-8140
www.rehab.alabama.gov

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification, care coordination, information and referrals.

Alabama Newborn Hearing Provider Directory

Madison County (cont.)

Hearing and Speech Clinic
303 Williams Avenue • Huntsville, AL 35801
Phone: 256-536-7405
www.hearingandspeechclinic.com

Medicaid Accepted: Yes
Services Provided: OAE and AABR screening for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification, cochlear implant assessment/management, wireless systems, speech therapy and auditory verbal therapy.

Huntsville Hospital Pediatric Therapy and Audiology
101 Sivley Road • Huntsville, AL 36305
Phone: 256-265-7952
www.huntsvillehospital.org

Medicaid Accepted: Yes
Services Provided: OAE and AABR screening for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification, cochlear implant assessment/management, wireless systems, speech therapy and auditory verbal therapy.

North Alabama ENT Associates, P.C.
927 Franklin Street, Suite 100 • Huntsville, AL 35801
Phone: 256-536-9300
www.NALENT.com

Medicaid Accepted: Yes
Services Provided: OAE screening for infants

Marengo County

Children's Rehab Services (CRS) - Dallas
2906 Citizens Parkway • Selma, AL 36701
Phone: 334-872-8422 or 1-800-967-6876
www.rehab.alabama.gov

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Marshall County

Children's Rehab Services (CRS) - Madison County
3000 Johnson Road • Huntsville, AL 35805
Phone: 256-650-1701 or 1-800-283-8140
www.rehab.alabama.gov

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification, care coordination, information and referrals.

Masdon ENT & Facial Plastic Surgery
602 Corley Avenue • Boaz, AL 35957
Phone: 256-571-8450
www.masdonent.com

Medicaid Accepted: Yes
Services Provided: OAE screening for infants. Early intervention services for infants and children with hearing loss to include hearing aid amplification.

Mobile County

Children's Rehab Services (CRS) - Mobile
1610 Center Street, Suite A • Mobile, AL 36604
Phone: 251-432-4560 or 1-800-568-9034
www.rehab.alabama.gov

Medicaid Accepted: Yes
Services Provided: OAE and diagnostic audiology services for infants

USA Hearing and Speech Clinic
1119 HAHN, 5721 USA Drive South • Mobile, AL 36688
Phone: 251-445-9378 • FAX: 251-445-9377
www.southalabama.edu

Medicaid Accepted: Yes
Services Provided: OAE screening, electrophysiological testing (ABR, AARB, ECoG, ENG), hearing aid evaluations and device dispensing, assessment of cochlear implants, speech and language evaluations, and intervention services.

Alabama Newborn Hearing Provider Directory

Montgomery County

Auburn University at Montgomery Speech and Hearing Clinic
7177 Halcyon Summit Drive • Montgomery, AL 36117
Phone: 334-244-3408

Medicaid Accepted: Yes
Services Provided: OAE screening for infants. ABR diagnostic audiology services for infants.

Central Alabama Ear, Nose, and Throat
2163 Normadie Drive • Montgomery, AL 36111
Phone: 334-284-1870

Medicaid Accepted: Yes
Services Provided: ABR diagnostic testing for infants. Diagnostic audiology services for infants.

Children's Rehab Services (CRS) - Montgomery
602 S. Lawrence Street • Montgomery, AL 36104
Phone: 334-293-7118 • www.rehab.alabama.gov

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Doctor's Hearing Clinic - A Hearing Life Company
7025 Halcyon Park Drive, Suite A • Montgomery, AL 36117
Phone: 334-396-1635 • www.doctorshearingclinic.com

Medicaid Accepted: No
Services Provided: Early Intervention Services for infants and children with hearing loss to include hearing aid amplification, cochlear implant assessment/management, wireless systems, and aural rehabilitation.

Morgan County

Children's Rehab Services (CRS) - Madison County
3000 Johnson Road • Huntsville, AL 35805
Phone: 256-650-1701 or 1-800-283-8140
www.rehab.alabama.gov

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification, care coordination, information and referrals.

Decatur ENT
1218 13th Avenue SE • Decatur, AL 35601
Phone: 256-355-6200
www.decaturent.com

Medicaid Accepted: Unknown, call to verify
Services Provided: OAE and AABR screening for infants.

Perry County

Children's Rehab Services (CRS) - Dallas
2906 Citizens Parkway • Selma, AL 36701
Phone: 334-872-8422 or 1-800-967-6876
www.rehab.alabama.gov

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Alabama Newborn Hearing Provider Directory

Pickens County

Children's Rehab Services (CRS) - Tuscaloosa
1110 Dr. Edward Hillard Drive • P.O. Box 2817
Tuscaloosa, AL 35403
Phone: 205-759-1279 or 1-800-723-0490
www.rehab.alabama.gov
Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Pike County

Children's Rehab Services (CRS) - Montgomery
602 S. Lawrence Street • Montgomery, AL 36104
Phone: 334-293-7118 • www.rehab.alabama.gov
Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Troy ENT
1320 Highway 231 South, Suite 3 • Troy, AL 36081
Phone: 334-807-8448
Medicaid Accepted: Yes
Services Provided: OAE screening for infants.

Russell County

Auburn University Speech and Hearing Clinic
1199 Haley Center • Auburn, AL 36849
Phone: 334-844-9600
www.cla.auburn.edu/speechandhearingclinic.edu
Medicaid Accepted: Yes
Services Provided: OAE, ABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification and speech therapy.

Southern Head and Neck Surgery
3368 Highway 280 G-15 • Alexander City, AL 35101
Phone: 256-329-1114
www.southernheadandnecksurgery.net
Medicaid Accepted: Yes
Services Provided: OAE and AABR screening for infants.

St. Clair County

Children's Rehab Services (CRS) - Anniston
1910 Coleman Road • Anniston, AL 35207
Phone: 256-240-8801 or 1-800-289-9533
www.rehab.alabama.gov
Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification, wireless systems, speech therapy, care coordination and treatment planning.

Shelby County

Children's Rehab Services (CRS) - Jefferson
234 Goodwin Crest Drive • Homewood, AL 35209
Phone: 205-290-4550 • 1-888-430-7423
www.rehab.alabama.gov
Medicaid Accepted: Yes
Services Provided: OAE, AABR, and ABR diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification, care coordination, information, and referrals.

Alabama Newborn Hearing Provider Directory

Shelby County (cont.)

ENT Associates of Alabama, P.C. - Alabaster
644 2nd Street NE, Suite 107 • Alabaster, AL 35007
Phone: 205-933-9236
www.entalabama.com
Medicaid Accepted: Yes
Services Provided: OAE screening for infants.

Shelby Ear, Nose & Throat
1228 Highway 31, 1st Street N • Alabaster, AL 35007
Phone: 205-621-8900
Medicaid Accepted: Yes
Services Provided: OAE screening for infants.

Sumter County

Children's Rehab Services (CRS) - Tuscaloosa
1110 Dr. Edward Hillard Drive • P.O. Box 2817
Tuscaloosa, AL 35403
Phone: 205-759-1279 or 1-800-723-0490
www.rehab.alabama.gov
Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Talladega County

Alabama Institute for Deaf and Blind
Health and Clinic Services/Dowling Building
Talladega, AL 35160
Phone: 256-761-3245
www.AIDB.org
Medicaid Accepted: Yes
Services Provided: OAE screening and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification, wireless systems, speech therapy, occupational therapy, and physical therapy.

Talladega County (cont.)

Children's Rehab Services (CRS) - Anniston
1910 Coleman Road • Anniston, AL 35207
Phone: 256-240-8801 or 1-800-289-9533
www.rehab.alabama.gov
Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification, wireless systems, speech therapy, care coordination and treatment planning.

Tuscaloosa County

Alabama Ear, Nose & Throat Specialists
302 Merchants Walk, Suite 100 • Tuscaloosa, AL 35406
Phone: 205-523-9300
www.alabamaentspecialists.com
Medicaid Accepted: Unknown, call to verify
Services Provided: Call to verify

Children's Rehab Services (CRS) - Tuscaloosa
1110 Dr. Edward Hillard Drive • P.O. Box 2817
Tuscaloosa, AL 35403
Phone: 205-759-1279 or 1-800-723-0490
www.rehab.alabama.gov
Medicaid Accepted: Yes
Services Provided: OAE, AABR, and diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification.

Ear, Nose & Throat Associates of Tuscaloosa
1224 McFarland Blvd NE, Suite A • Tuscaloosa, AL 35406
Phone: 205-333-3330
www.entassociatestuscaloosa.com
Medicaid Accepted: Yes
Services Provided: OAE screening for infants

Alabama Newborn Hearing Provider Directory

Tuscaloosa County (cont.)

Tuscaloosa Ear, Nose & Throat Center
1300 McFarland Blvd NE, Ste 150 • Tuscaloosa, AL 35406
Phone: 205-758-9041
www.tuscaloosaent.com

Medicaid Accepted: Yes
Services Provided: OAE screening for infants

University of Alabama Speech and Hearing Center
700 University Blvd E, Room 145 • Tuscaloosa, AL 35487
Phone: 205-348-7131
www.cd.ua.edu

Medicaid Accepted: Yes
Services Provided: OAE and AABR screening for infants. Early intervention service for infants and children with hearing loss to include hearing aid amplification, cochlear implant assessement/management, wireless systems, and speech therapy.

Walker County

Children's Rehab Services (CRS) - Jefferson
234 Goodwin Crest Drive • Homewood, AL 35209
Phone: 205-290-4550 • 1-888-430-7423
www.rehab.alabama.gov

Medicaid Accepted: Yes
Services Provided: OAE, AABR, and ABR diagnostic audiology services for infants. Early Intervention Services for infants and children with hearing loss to include hearing aid amplification, care coordination, information, and referrals.

ENT Associates of Alabama, P.C. - Jasper
3400 Highway 78 E., Ste 205 • Jasper, AL 35501
Phone: 251-970-3277
www.hearingandbalance.net

Medicaid Accepted: Yes
Services Provided: OAE screening for infants

Alabama's Listening! UNIVERSAL NEWBORN HEARING SCREENING

Alabama Department of Public Health
Newborn Hearing Screening Program

Outpatient Newborn Hearing Provider Information

Facility Information

Facility Name: _____

Address: _____
Street Address *Suite #*

_____
City *State* *ZIP Code*

Phone: _____ Website: _____

Practitioners: _____

_____

Questionnaire

Does this site accept Medicaid as a Payor Source? ☐ Yes ☐ No

Does this site provide outpatient hearing screening for newborns and infants? ☐ Yes ☐ No

Does this site provide Otoacoustic Emissions (OAE)? ☐ Yes ☐ No

Does this site provide Automated Auditory Brainstem Response (AABR) testing? ☐ Yes ☐ No

Does this site provide diagnostic services for newborns and infants as recommended by the Joint Committee on Infant Hearing (JCIH) 2007 Position Statement? ☐ Yes ☐ No

Does this site provide Early Intervention services for infants and children with hearing loss? ☐ Yes ☐ No
If yes, please indicate intervention services available:

☐ hearing aid amplification
☐ cochlear implant assessment/management
☐ wireless systems
☐ speech therapy
☐ other:_____

Please return this completed questionnaire via mail, email or fax to:

Alabama Department of Public Health
Newborn Hearing Screening Program
Attn: Rovetta Hanna, BSN, RN
201 Monroe Street, Suite 1350
Montgomery, Alabama 36130-3017
Fax: 334-206-3791
Email: rovetta.hanna@adph.state.al.us

Section 4 - Resources

Public Health Areas Map

PHA 1
Karen Landers, M.D., Area Health Officer
Ronnie Moore, Area Administrator
Box 929, Tuscumbia, AL 35674
(256) 383-1231

PHA 2
Judy Smith, Area Administrator
Box 1628, Decatur, AL 35602
(256) 340-2113

PHA 3
Albert T. White, Jr., M.D., Area Health Officer
Tammy Yager, Area Administrator
Box 70190, Tuscaloosa, AL 35407
(205) 554-4500

PHA 4
Mark E. Wilson, M.D., Area Health Officer
Box 2648, Birmingham, AL 35202
(205) 930-1500

PHA 5
Mary Gomillion, Area Administrator
Box 8306, Gadsden, AL 35902
(256) 547-6311

PHA 6
Mary Gomillion, Area Administrator
3400 McClellan Blvd., Anniston AL 36201
(256) 237-1896

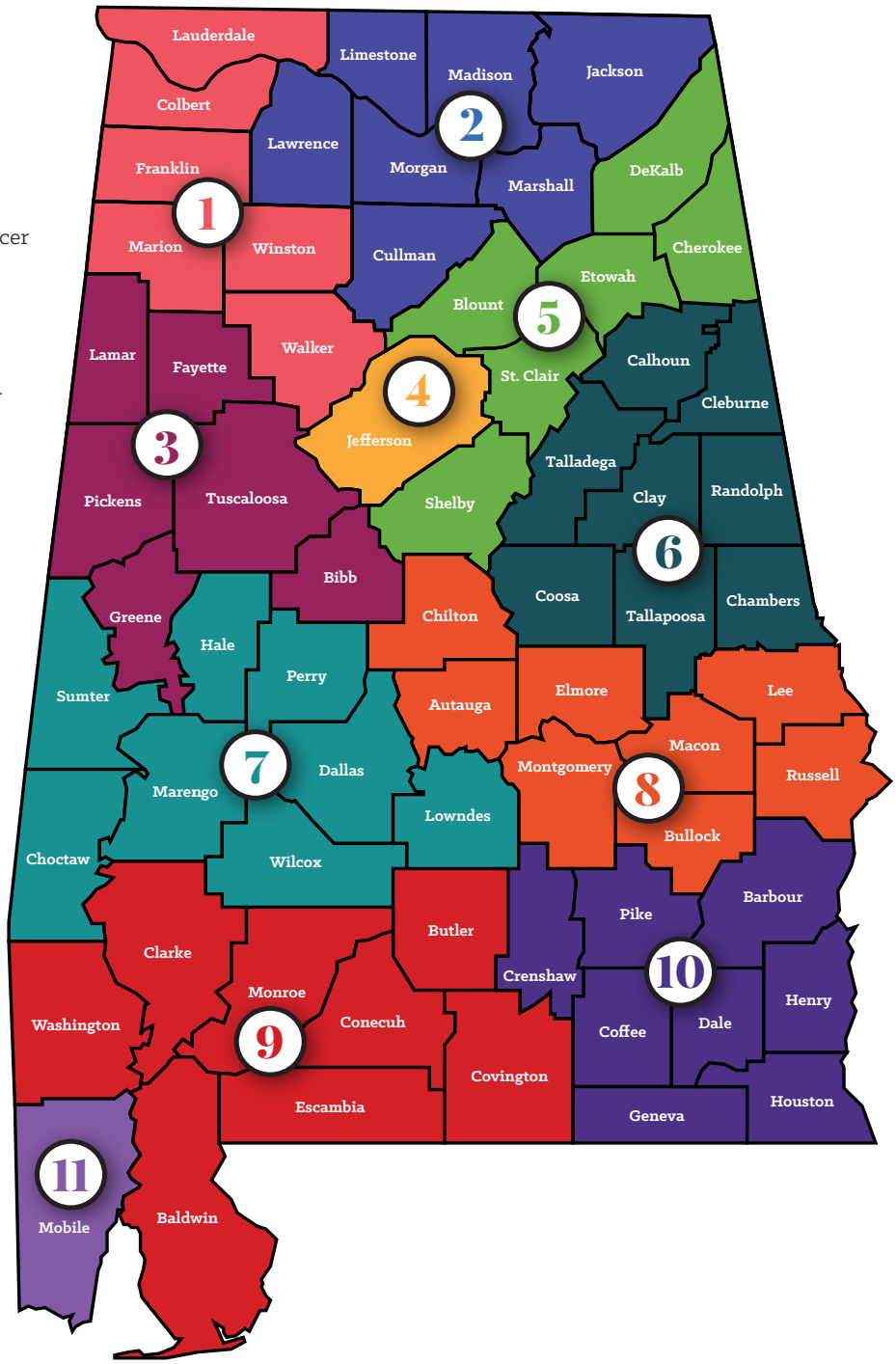
PHA 7
Jackie Holliday, Area Administrator
303 Industrial Dr., Linden, AL 36748
(334) 295-1000

PHA 8
James Martin, Area Administrator
2500 Fairlane Dr., Bldg. 2, Ste. 200,
Montgomery, AL 36116
(334) 277-8464

PHA 9
Ricky Elliott, Area Administrator
312 Courthouse Square, Ste. 31,
Bay Minette, AL 36507
(251) 937-5859

PHA 10
James Martin, Acting Area Administrator
Drawer 2087, Dothan, AL 36301
(334) 792-9070

PHA 11
Bernard H. Eichold, II, M.D.
Area Health Officer
Box 2867, Mobile, AL 36652
(251) 690-8827



Alabama County Health Departments

PHA 1	Colbert County	256-383-1231	PHA 7	Choctaw County	205-459-4026
	Franklin County	256-332-2700		Dallas County	334-874-2550
	Lauderdale County	256-764-7453		Hale County	334-624-3018
	Marion County	205-921-3118		Lowndes County	334-548-2564
	Walker County	205-221-9775		Marengo County	334-295-4205
	Winston County	205-489-2101		Perry County	334-683-6153
				Sumter County	205-652-7972
				Wilcox County	334-682-4515
PHA 2	Cullman County	256-734-1030	PHA 8	Autauga County	334-361-3743
	Jackson County	256-259-4161		Bullock County	334-738-3030
	Lawrence County	256-974-1141		Chilton County	205-755-1287
	Limestone County	256-232-3200		Elmore County	334-567-1171
	Madison County	256-539-3711		Lee County	334-745-5765
	Marshall County	256-582-3174		Macon County	334-727-1800
	Morgan County	256-353-7021		Montgomery County	334-293-6400
				Russell County	334-297-0251
PHA 3	Bibb County	205-926-9702	PHA 9	Baldwin County	251-947-1910
	Fayette County	205-932-5260		Butler County	334-382-3154
	Greene County	205-372-9361		Clarke County	251-275-3772
	Lamar County	205-695-9195		Conecuh County	251-578-1952
	Pickens County	205-367-8157		Covington County	334-222-1175
	Tuscaloosa County	205-562-6900		Escambia County	251-867-5765
				Monroe County	251-575-3109
				Washington County	251-847-2245
PHA 4	Jefferson County:		PHA 10	Barbour County	334-687-4808
	Bessemer Health Center	205-497-9300		Coffee County	334-347-9574
	Central Health Center	205-933-9110		Crenshaw County	334-335-2471
	Eastern Health Center	205-591-5180		Dale County	334-774-5146
	Morris Health Center	205-933-4242		Geneva County	334-684-2256
	West End Health Center	205-715-6121		Henry County	334-585-2660
	Western Health Center	205-788-3321		Houston County	334-678-2800
				Pike County	334-566-2860
PHA 5	Blount County	205-274-2120	PHA 11	Mobile County:	
	Cherokee County	256-927-3132		Citronelle Clinic	251-866-9126
	Dekalb County	256-845-1931		Eight Mile Clinic	251-456-1399
	Etowah County	256-547-6311		Keeler, Main Site	251-690-8158
	St. Clair County	205-338-3357		Newburn Clinic	251-405-4524
	Shelby County	205-664-2470		North Mobile Health Center	251-829-9884
				Semmes Clinic	251-690-8935
				Southwest Mobile Health Center	251-666-7413
				Teen Center	251-694-5038
				Women's Center	251-445-0582
PHA 6	Calhoun County	256-237-1896	Not all health departments perform newborn screens. Some have EPSDT MOUs with local physicians to perform screens for them.		
	Chambers County	334-756-0758			
	Clay County	256-396-6421			
	Cleburne County	256-463-2296			
	Coosa County	256-377-4364			
	Randolph County	334-863-8983			
	Talladega County	256-362-2593			
	Tallapoosa County	256-329-0531			

Child Find...

Alabama's Early Intervention System for Infants and Toddlers With Disabilities

Child Find is the process used in Alabama for *identifying* all children who may be eligible for services and *referring* them to Alabama's Early Intervention System. It is an important step that provides families with the guidance and support they need to make it on their own behalf.

There are three steps in the Child Find process:

1. Identification - Children who may be in need of special help are identified by parents or by individuals within the community. These individuals, agencies, or organizations may include:

parents	developmental disabilities programs	outpatient clinics
well-baby clinics	prenatal/postnatal facilities	public health facilities
hospital follow-up clinics	child day care centers	Medicaid programs
physicians	home child day care programs	hospitals
pediatricians' offices	Head Start programs	social service agencies
community health services	local educational agencies	other healthcare providers

Children are identified when *parents or other family members* express concern about their child's development. Children are also identified when a *service provider* suspects that there is delay in a child's development and discusses this concern with the parents. Any infant or toddler age birth to 3 years with a delay of 25 percent or more in any of the major areas of development - cognitive, physical, communication, social, emotional, or adaptive development - who lives in Alabama is eligible to receive supports and appropriate services through the state's early intervention system. Children may be identified if a child has a *diagnosed physical or mental condition* that may contribute to a developmental delay.

Once potentially eligible infants and toddlers have been identified as having a suspected or diagnosed delay, the service provider or family may make a referral to Child Find. Families need to be made aware when a service provider is making the referral.

2. Making a Referral - making a referral to Alabama's Early Intervention System is as simple as making a phone call to **Early Intervention Child Find at 1-800-543-3098** (voice/TDD). Fax-back referral forms are also available for use by doctor's offices, social workers, hospitals, etc.

If parents ask a service provider to make the referring phone call, the referral must be made no more than *two working days* after the child has been identified. Information needed to make a referral includes the child's name, sex, ethnic origin, birth date, and Social Security number, if available. Additional information needed to process the referral includes the name of the parents or guardian, language spoken by the family, areas of development that are of concern to the parents and professional, the name of the child's primary care physician, and whether the family has agreed to the referral to AEIS.

3. Processing of a Referral - When a call is received by Child Find, the child's name and other identifying information will be entered into the data base for follow-up by Alabama's Early Intervention System. The referral will be passed on to the local contact (known as the District Early Intervention Coordinator or DEIC) within the child's community. The coordinator will contact the child's family within a two-week period to discuss Alabama's Early Intervention System and explain the evaluation process. The child and family's progress through Alabama's Early intervention System will be monitored under the lead agency, the Alabama Department of Rehabilitation Services.

Any individual who works with young children and their families is in a unique position to help identify, at an early stage, those infants and toddlers who may need intervention. It is important that any child under the age of 3 that may have a delay in development be referred to Alabama's Early Intervention System as quickly as possible. The evaluation and assessment process for the state's early intervention system for infants and toddlers with disabilities, and their families, is free to the family. Families are also not required to pay for appropriate services for their eligible child.

If you work with young children and families, you can help them in the following ways:

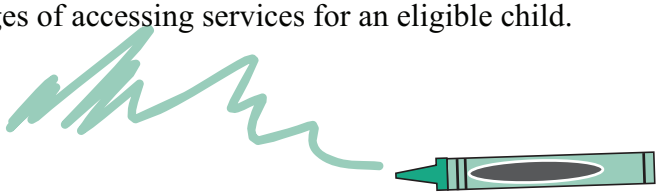
- Display information about Alabama's Early Intervention System in offices, libraries, faith-based facilities and clinics. Free materials are available by simply calling **Early Intervention Child Find at 1-800-543-3098** and making a request for free AEIS materials.

- Help families monitor their children's development by helping them to understand developmental milestones.

- Act on any concerns that you have or that are expressed by parents by discussing the early intervention Child Find process with the family and helping them to make the contact if they are interested.

- Monitor the progress of an infant or toddler that may have a delay if a family is not ready to make a referral or a decision and talk to the parents at a later date if necessary.

- Nurture families who have infants and young children and understand the stress they are enduring. Provide information, guidance and support that parents may need to make informed choices during the early stages of accessing services for an eligible child.



To learn more about
Alabama's Early Intervention System,
contact the Early Intervention Office,
located within the Alabama Department of Rehabilitation Services,
at 1-800-441-7607 or visit the web site at www.rehab.state.al.us.

Child Find

☐

Alabama's Early Intervention System **PLEASE PRINT CLEARLY**

Child Find Referral Form

1-800-543-3098

En Espanol: 1-866-450-2838

Child Find Fax # (334) 293-7393

www.rehab.alabama.gov/ei

(State Office Use Only)
New Case ID #: _____

INFANT/TODDLER INFORMATION

1. SSN#: _____ 2. Date of Birth: _____ 3. Sex: **F** ☐ **M** ☐

4. Last Name: _____ First Name: _____ MI/Name: _____

5. Is your child of Hispanic or Latino origin? Y ☐ N ☐ 6. Child's Primary Race: _____

* **If Primary Race is Two or More Races:** ☐ Hispanic/Latino ☐ American Indian/Alaska Native ☐ Asian
(Mark appropriate boxes)
☐ Black/African American ☐ Hawaiian/Pacific Islander ☐ White

7. Home Language: _____ 8. Medicaid: Y ☐ N ☐ Medicaid # _____

9. Private Insurance: Y ☐ N ☐ 10. CHIP/All Kids Y ☐ N ☐

CHILD RELATION INFORMATION

11. First Name: _____ Last Name: _____ MI: _____

12. Relation Type: _____ 13. Is this Primary relation? Y ☐ N ☐ 14. Is address same as child'? Y ☐ N ☐

15. Mailing Address: _____

City/State/Zip: _____ 16. County: _____

17. Physical Address: _____

City/State/Zip: _____ 18. County: _____

19. Primary contact #: ( ) _____ 20. Alternate contact #: ( ) _____

Alternate contact #: ( ) _____ Work Phone #: ( ) _____ Ext #: _____

REFERRAL SOURCE INFORMATION

21. Person making referral: _____ 22. Referral Source: _____

23. County: _____ 24. Phone: _____ 25. Fax: _____

26. Reason for referral: _____

27. How family became aware of Child Find: _____ Additional Information: _____

Refer to Service Coordinator/Caseload ID #: _____

Date Mailed/Faxed to Child Find: _____ Sender's Name/Phone #: _____

INCOMPLETE REFERRALS WILL NOT BE ACCEPTED (FILL IN ALL REQUIRED BLANKS)
BELOW - STATE OFFICE USE ONLY

*** Mail to: ADRS/EI, 602 S. Lawrence St., Montgomery, AL 36104 ** Child Find Fax Number: 334-293-7393**

Referral taken by: _____ **Date taken:** _____ **Processed by:** _____ **Official referral/entry date:** _____

☐ **ATTACHMENT:** _____ **Revised 02/13**

Children's Rehabilitation Service

Any child or adolescent younger than 21 years of age who is a resident of Alabama and has a special health care need is eligible for CRS. CRS provides specialty medical services to include medical clinics, evaluation clinics, medication, equipment, therapies, hospitalizations, and surgeries as well as support for families.

Calhoun County – Anniston CRS 1910 Coleman Road Anniston, AL 36207 Phone: 256-240-8801 or 1-800-289-9533 Counties: Calhoun, Cherokee, Clay, Cleburne, St. Clair, Talladega	**Jefferson County - Homewood CRS** 234 Goodwin Crest Drive Birmingham, AL 35209 Phone: 205-290-4550 or 1-888-430-7423 Counties: Cullman, Jefferson, Shelby, Walker
Clarke County – Jackson CRS 1506 College Avenue Jackson, AL 36545 Phone: 251-246-4025 or 1-800-283-8140 Counties: Choctaw, Clarke, Monroe, Washington	**Lee County – Opelika CRS** 516 W. Thomason Circle Opelika, AL 36801 Phone: 334-749-8339 or 1-800-568-8428 Counties: Chambers, Lee, Macon, Randolph, Russell, Tallapoosa
Colbert County – Muscle Shoals CRS 1450 E. Avalon Avenue Muscle Shoals, AL 35661 Phone: 256-381-1212 or 1-800-285-9924 Counties: Colbert, Franklin, Lauderdale, Lawrence, Marion, Winston	**Madison County – Huntsville CRS** 3000 Johnson Road Huntsville, AL 35805 Phone: 256-650-1701 or 1-800-283-8140 Counties: Jackson, Limestone, Madison, Marshall, Morgan
Covington County – Andalusia CRS 1082 Village Square Drive, Suite 2 Andalusia, AL 36420 Phone: 334-222-5558 or 1-800-723-8064 Counties: Butler, Conecuh, Covington, Crenshaw	**Montgomery County – Montgomery CRS** 602 South Lawrence Street Montgomery, AL 36104 Phone: 334-293-7500 or 1-800-568-9034 Counties: Autauga, Bullock, Chilton, Coosa, Elmore, Lowndes, Montgomery, Pike
Dallas County – Selma CRS 2906 Citizens Parkway Selma, AL 36701 Phone: 334-872-8422 or 1-800-967-6876 Counties: Dallas, Marengo, Perry, Wilcox	**Mobile County – Mobile CRS** 1610 Center Street, Suite A Mobile, AL 36604 Phone: 251-432-4560 or 1-800-879-8163 Counties: Baldwin, Escambia, Mobile
Etowah County – Gadsden CRS 1100 George Wallace Drive Gadsden, AL 35903 Phone: 256-547-8653 or 1-800-289-1353 Counties: Blount, Dekalb, Etowah	**Talladega County – Talladega CRS** office closed – clients referred to Anniston
Houston County – Dothan CRS 795 Ross Clark Circle NE, Suite 3 Dothan, AL 36303 Phone: 334-699-6600 or 1-800-677-9123 Counties: Barbour, Coffee, Dale, Geneva, Henry, Houston	**Tuscaloosa County – Tuscaloosa CRS** 1110 Dr. Edward Hillard Drive P.O. Drawer 2817 Tuscaloosa, AL 35403 Phone: 205-759-1279 or 1-800-723-0490 Counties: Bibb, Fayette, Greene, Hale, Lamar, Pickens, Sumter, Tuscaloosa

Alabama Community Based Sickle Cell Organizations

The Alabama NBS Program refers all infants identified with sickle cell trait and sickle cell disease to one of the local Community-Based Sickle Cell Organizations. Genetic counseling is offered to theses families.

Organization	Contacts	Address & Phone	Counties
Sickle Cell Disease Association of America Central Alabama Chapter Service Area I	Claudette Stallworth Executive Director Ms. Sharon Lewis	3813 Avenue I Ensley Birmingham, AL 35218 205-780-2355 Fax: 205-780-2368	Blount, Calhoun, Cherokee, Clay, Cleburne, Cullman, Etowah, Jefferson, Randolph, Shelby, St. Clair, Talladega, Walker
Sickle Cell Disease Association of America West Alabama Chapter Service Area II	Executive Director Jennifer Harris Sherman	P.O. Box 3151 Tuscaloosa, AL 35403 205-758-1761 Fax: 205-758-1781	Fayette, Green, Hale, Lamar,Marion, Pickens, Sumter, Tuscaloosa, Winston
Tri-County Sickle Cell Anemia Association, Inc. Service Area III	Mamie Danzey Executive Director Ms. Margaret Bolling	P.O. Box 3151 Selma, AL 36701 334-872-9362 Fax: 334-872-9383	Bibb, Chilton, Coosa, Dallas, Marengo, Perry, Wilcox
Sickle Cell Foundation of Greater Montgomery, Inc. Service Area IV	Tameka Westry Executive Director Monica Vandiver	3180 US Highway 80 West P.O. Box 9278 Montgomery, AL 36087 334-286-9122 Fax: 334-286-4804	Autauga, Butler, Chambers, Coffee, Crenshaw, Elmore, Lowndes, Montgomery, Tallapoosa
Southeast Alabama Sickle Cell Association Service Area V	Janie Cowan Executive Director Shelby B. Powell	P.O. Box 1079 Tuskegee, AL 36087 334-727-6120	Barbour, Bullock, Dale, Geneva, Henry, Houston, Lee, Macon, Pike, Russell
Sickle Cell Disease Association of America Mobile Chapter, Inc. Service Area VI	Executive Director Keava Boswell Jones	P.O. Box 40696 1453 Springhill Avenue Mobile, AL 36604 251-432-0301	Baldwin, Choctaw, Clarke, Conecuh, Covington, Escambia, Mobile, Monroe, Washington
North Alabama Sickle Cell Foundation, Inc. Service Area VII	McAuthor Johnson Executive Director Pamela Thompson	P.O. Box 813 Huntsville, AL 35804 256-536-2723 1-800-636-2723 Fax: 256-536-2714	Colbert, Dekalb, Franklin, Jackson, Lauderdale, Lawrence, Limestone, Madison, Marshall, Morgan

9/2014

Appendix

Alabama Newborn Screening Timeline

1964 PKU

1978 Congenital Hypothyroidism

1987 Hemoglobinopathies

1992 Galactosemia

1994 Congenital Adrenal Hyperplasia

1997 Voice Response System (VRS)

04/2004 Biotinidase Deficiency

10/2004 **Amino Acid Disorders:**
Citrullinemia (CIT)
Homocystinuria (HCY)
Maple Syrup Urine Disease (MSUD)
Tyrosinemia (TYR)
Argininosuccinate aciduria (ASA)

Organic Acid Disorders:
Propionic Acidemia (PROP)
Methylmalonic Acidemia (Vitamin B12 Disorders) (CBL, A,B)
Methylmalonic Acidemia (methylmalonyl-CoA mutase) (MUT)

Fatty Acid Disorders:
Medium chain acyl-CoA dehydrogenase deficiency (MCAD)
Carnitine Uptake Defect (CUD)

10/2006 **Organic Acid Disorders:**
Glutaric Acidemia (GA-1)
Isovaleric Acidemia (IVA)
Multiple carboxylase (MCD
3-Hydroxy 3-methylglutaric Aciduria (HMG)

04/2007 **Fatty Acid Disorders:**
Very long chain acyl-CoA dehydrogenase deficiency (VLCAD)
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
Trifunctional Protein Deficiency (TFP)

Organic Acid Disorders:
3-Methylcrotonyl-CoA carboxylase (3-MCC)
Beta ketothiolase (BKT)
Carnitine palymitoyltranferase II (CPT II)

01/2008 Universal Newborn Hearing Screening*

04/2008 Cystic Fibrosis (CF) (IRT/DNA)

2009 Cord Blood collection and testing discontinued

06/2013 Critical Congenital Heart Disease (CCHD)

***started voluntarily in 2001/ mandated 2008**

Newborn Screening Disorder Descriptions

3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency (HMG)

- organic acid disorder caused by a defect in processing the amino acid leucine
- very rare; incidence unknown
- brain damage, seizures, and death may occur without treatment
- treatment includes avoidance of fasting, low-leucine diet and L-carnitine supplementation

3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC)

- organic acid disorder caused by a defect in processing the amino acid leucine
- occurs 1 in 50,000 newborns
- brain damage, seizures, liver failure, coma and death may occur without treatment
- treatment with a low-protein diet and in some cases nutritional supplements

Argininosuccinate Acidemia (ASA)

- amino acid disorder resulting in build up of argininosuccinic acid and ammonia
- occurs less than 1 in 100,000 newborns
- brain swelling, coma, some degree of brain damage and sometimes death if untreated
- treatment consists of a low-protein diet, avoid fasting, meds to prevent ammonia build-up, nutritional supplements, and in some cases liver transplant

Beta-ketothiolase Deficiency (BKD)

- organic acid disorder resulting in build-up of acid triggered by some childhood illnesses
- rare; acutal incidence unknown
- coma, brain damage and death may occur if untreated
- treatment may include IV fluids to keep blood sugar levels up and acid levels down, avoidance of protein-rich foods, and long-term bicarbonate therapy

Biotinidase Deficiency

- an enzyme deficiency
- occurs 1 in 75,000 newborns
- seizures, hearing loss, and death in severe cases
- treatment includes daily doses of the vitamin biotin

Carnitine Uptake Deficiency (CUD)

- fatty acid disorder in which cells cannot bring in carnitine from the blood due to a missing transporter. Carnitine is needed for the transfer of fatty acids across membranes for cell energy
- occurs 1 in 50,000 newborns
- episodes of hypoglycemia (low blood sugar) and sudden unexpected death in infancy
- treatment includes carnitine replacement

Citrullinemia type I

- amino acid disorder resulting in build-up of citrulline that leads to ammonia build-up
- occurs 1 in 57,000 newborns
- seizures, coma, brain damage, and death can occur if untreated
- treatment includes low-protein diet, meds to rid the body of amino groups to prevent ammonia build-up, and nutritional supplements

Classical Galactosemia

- failure to metabolize the milk sugar galactose
- occurs 1 in 50,000 newborns
- may lead to damage of vital organs, blindness, severe mental retardation, infection, and death if not treated early
- treatment includes elimination of galactose from diet for life

Newborn Screening Disorder Descriptions

Congenital Adrenal Hyperplasia (CAH)

- lack of certain vital adrenal hormones called corticosteriods
- occurs 1 in 15,000 newborns
- masculinization of genitals, infertility in males and females, shock and/or death if untreated
- treatment may include steriods and special minerals depending if non-classical or classical form of CAH

Congenital Hypothyroidism

- lack of thyroid hormone
- occurs 1 in 3,000 newborns
- growth and brain development problems and mental retardation if not treated early
- treatment includes taking thyroid hormone daily

Critical Congenital Heart Disease (CCHD)

- seven heart defects classified as CCHD
- occurs 2 in 1,000 newborns
- significant risk for death or disability if not diagnosed and treated soon after birth
- pulse oximetry is the recommended screening method to detect CCHD

Cystic Fibrosis (CF))

- production of abnormally thick, sticky mucus in lungs and pancreas
- occurs 1 in 5,000 newborns
- recurrent cough, loose stools or intestinal obstruction (meconium illeus), electrolyte imbalance, pulmonary infections, airway obstruction, and/or growth failure
- pancreatic enzyme supplements, airway clearance techniques, aerolized meds

Glutaric Acidemia (GA-1)

- organic acid disorder resulting from a loss of an enzyme needed to break down amino acids
- occurs 1 in 40,000 newborns
- may develop normally for up to 18 months until viral illness triggers symptoms
- may lead to brain damage, seizures, low muscle tone, cerebral-palsy like symptoms, and death
- treatment can vary but may include dietary protein restriction and supplement with L-carnitine

Hearing Loss

- full or partial decrease in ability to detect or understand sound
- occurs 1-3 in 1,000 newborns
- delayed speech and language development
- treatment may include amplification, speech therapy, ear tubes, or surgical intervention (cochlear implants)

Hemoglobin S/beta thalassemia (Hb S/B Th)

- blood disorder which results from one sickle cell gene and one beta thalassemia gene
- occurs 1 in 50,000 newborns
- symptoms are often milder than sickle cell disease and vary among affected children
- treatment with penicillin may not be recommended for all affected children

Hemoglobin SC Disease (Hb S/C)

- blood disorder that results from one sickle cell gene and one hemoglobin C gene
- occurs 1 in 25,000 newborns
- symptoms may be milder than sickle cell disease and vary among affected children
- treatment with penicillin may not be recommended for all affected children

Newborn Screening Disorder Descriptions

Hemoglobin SS Disease (Hb SS)

- blood disease that causes sickling of blood cells and results in anemia
- occurs 1 in 5,000 newborns; higher incidence among African Americans (1 in 400)
- severe pain, damage to the vital organs, stroke, and sometimes death
- treatment may include penicillin, vaccinations to prevent infections, pain meds, and/or blood transfusions

Homocystinuria (HCY)

- amino acid disorder lacking an enzyme responsible for converting homocysteine into cystathionene, which is needed for normal brain development
- occurs less than 1 in 100,000 newborns
- mental retardation, eye problems, skeletal abnormalities, and/or stroke
- treatment consists of a special diet, one or more vitamins, and other supplements

Isovaleric Acidemia (IVA)

- organic acid disorder caused by an inability to process the amino acid leucine
- occurs 1 in 230,000 newborns
- coma, permanent brain damage and death can occur if untreated
- treatment includes a low-protein diet and nutritional supplements

Long Chain L-3-OH Acyl-CoA Dehydrogenase Deficiency (LCHAD)

- fatty acid disorder
- rare disorder; incidence unknown
- low muscle tone, developmental delay; heart, lung, liver failure may develop later in infancy following illness
- treatment includes a high carbohydrate/low-fat diet, nutritional supplements, and avoidance of fasting

Maple Syrup Urine Disease (MSUD)

- amino acid disorder affecting branch-chained amino acids
- occurs less than 1 in 100,000 newborns
- sweet smell to urine, intellectual disability, and/or death if not treated early
- low-protein diet for life and possible supplementation with a vitamin, thiamin

Medium Chain Acyl CoA Dehydrogenase Deficiency (MCADD)

- fatty acid disorder
- occurs 1 in 15,000 newborns
- seizures (caused by low blood sugar), liver failure, coma and death may occur
- identifying affected children before they become ill is vital to preventing a crisis
- treatment includes avoidance of fasting and nutritional supplements

Methylmalonic Acidemia (MMA)

- organic acid disorder caused by a defect in the processing of 4 essential amino acids
- occurs 1 in 80,000 newborns
- death during the first month of life and brain damage in survivors is common if untreated
- treatment includes a low-protein diet, vitamin B12 injections, and nutritional supplements

Methylmalonic Acidemia CblA/CblB

- organic acid disorder caused by a defect of vitamin metabolism leading to a build-up of acids
- occurs 1 in 100,000 newborns
- brain damage, seizures, paralysis, coma and death may occur if untreated
- treatment includes a low-protein diet and vitamin B12 injections

Newborn Screening Disorder Descriptions

Multiple Carboxylase Deficiency

- organic acid disorder caused by a defect of an enzyme required to activate several biotin dependent enzymes resulting in build up of lactic acid in the body
- occurs 1 in 100,000 newborns
- skin rashes, hair loss, brain damage, coma and death may occur if untreated
- treatment with biotin allows normal growth and development

Phenylketonuria (PKU)

- amino acid disorder that results in an inability to properly process the essential amino acid phenylalanine, which accumulates and damages the brain
- occurs 1 in 10,000 newborns
- may result in severe mental retardation if not identified early and treated
- treatment includes special formula and a low phenylalanine diet

Propionic Acidemia (PPA)

- organic acid disorder caused by a defect in the processing of four essential amino acids
- occurs 1 in 100,000 newborns
- seizures, abnormal muscle tone, frequent infections, heart problems, brain damage, coma and death
- treatment includes a low-protein diet and nutritional supplements (some children may still experience some symptoms even if treated)

Severe Combined Immonodeficiency (SCID)*

- group of very rare inherited disorders resulting in a defect in the immune system
- occurs 1 in 70,000 newborns
- may lead to deadly infections if untreated
- most effective treatment is a bone marrow transplant within first few months of life

addition to Alabama NBS Panel in 2015

Trifunctional Protein Deficiency (TFP)

- fatty acid disorder
- very rare; incidence unknown
- a seemingly healthy infant can die of what appears to be sudden infant death syndrome, and others may experience low muscle tone, seizures, heart failure, and coma
- treatment is based on strict avoidance of fasting, a low-protein diet, and nutritional supplements

Tyrosinemia Type I

- amino acid disorder resulting in absence of an enzyme which leads to build up of a toxin called succinylacetone in the liver
- occurs 1 in 100,000 newborns
- liver or kidney failure, nerve damage and death may occur if not treated
- treatment includes drug treatment and a low-protein diet

Very Long Chain Acyl CoA Dehydrogenase Deficiency (VLCAD)

- fatty acid disorder
- occurs 1 in 30,000 newborns
- heart failure, liver failure, death during first year of life
- treatment includes a high carbohydrate and low-fat diet, nutritional supplements, and avoidance of fasting

Alabama NBS Confirmed Disorders

Disorders	2008	2009	2010	2011	2012	2013	2014
Biotinidase Deficiency	0	1	0	0	0	0	0
Classical Galactosemia	0	0	4	0	3	1	2
Critical Congenital Heart Disease	*	*	*	*	3	2	1
Secondary Congenital Heart Disease	*	*	*	*	3	3	1
Cystic Fibrosis	7	23	16	19	14	12	19
Hearing Loss	43	29	31	68	56	54	pending
Endocrine Disorders							
Congenital Adrenal Hyperplasia	7	5	3	5	4	2	5
Congenital Hypothyroidism	16	30	33	29	32	36	37
Hemoglobinapathy							
Sickle Cell Disease	57	62	56	66	52	53	58
Sickle Cell Trait	1818	1940	1860	1835	1866	1817	1468
Amino Acid Disorders							
Argininosuccinate Aciduria (ASA)	0	0	0	0	0	0	1
Homocystinuria	1	0	0	0	0	0	0
Maple Syrup Urine Disease (MSUD)	0	0	0	0	0	1	0
Phenylketonuria	3	4	3	5	2	4	4
Tyrosinemia	-	-	1	0	0	0	0
Fatty Acid Disorders							
Carnitine Uptake Defect	2	3	0	2	1	1	1
Long chain Acyl-CoA Dehydrogenase Deficiency (LCAD)	0	0	0	0	0	1	0
Medium chain Acyl-CoA Dehydrogenase Deficiency (MCAD)	3	6	5	5	4	3	3
Trifunctional Protein Deficiency	0	1	0	0	0	0	0
Very long chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)	0	0	0	1	1	1	4
Organic Acid Disorders							
2-Methylbutyryl-CoA Dehydrogenase Deficiency	1	0	0	0	1	0	1
3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC)	0	1	0	0	2	0	0
Glutaric Acidemia	1	0	0	1	0	0	0
Isovaleric Acidemia	0	0	0	0	0	0	1
Methylmalonic Acidemia (MMA)	2	2	0	1	1	1	1
Propionic Acidemia	0	0	1	0	0	0	1

*Disorder not included in screening panel

Public Health Laws of Alabama

Section 22-20-3

Neonatal testing for certain diseases; rules and regulations for treatment thereof.

(a) It shall be the duty of the administrative officer or other persons in charge of each institution caring for infants 28 days or less of age, or the physician attending a newborn child or the person attending a newborn child that was not attended by a physician to cause to have administered to every such infant or child in his care a reliable test for hypothyroidism and a reliable test for phenylketonuria (PKU), such as the Guthrie test, or any other test considered equally reliable by the State Board of Health and a reliable test for sickle cell anemia, sickle cell trait, and/or abnormal hemoglobin and such other tests relating to mental retardation or other heritable diseases and conditions as are designated by the Board of Health. Provided, however, that the Board of Health shall designate only conditions that are detectable by mass screening of newborn infants. Initial mass screening tests and the recording of results shall be performed by the Public Health Laboratory at such times and in such manner as may be prescribed by the State Board of Health; confirmatory tests shall be undertaken by such laboratory facilities as are designated by the attending physician or parent; provided, that no such initial screening or confirmatory tests shall be given to any child whose parents object thereto on the grounds that such tests conflict with their religious tenets and practices. In the event a test is not given to a child on account of such objections by the parents, then no physician, nurse, laboratory technician, person administering tests, hospital, institution or other health care provider shall be liable for failure to administer the test.

(b) The State Board of Health shall promulgate such rules and regulations as it considers necessary to provide for the care and treatment of those newborn infants whose tests are determined positive, including but not limited to, advising dietary treatment for such infants. The State Board of Health shall promulgate any other rules and regulations necessary to effectuate the provisions of this section including the collection of a reasonable fee for the newborn child screening program.

(Acts 1965, No. 885, p. 1664; Acts 1979, No. 79-437, p. 703; Acts 1987, No. 87-672, p. 1202; Acts 1991, 1st Ex. Sess., No. 91-793, p. 188, §1.)

Alabama State Board of Health Administrative Code

ALABAMA STATE BOARD OF HEALTH
ALABAMA DEPARTMENT OF PUBLIC HEALTH
BUREAU OF FAMILY HEALTH SERVICES
ADMINISTRATIVE CODE

CHAPTER 420-10-1
CARE AND TREATMENT OF INFANTS IDENTIFIED THROUGH THE NEWBORN SCREENING PROGRAM

TABLE OF CONTENTS

420-10-1-.01 Purpose. The purpose of these rules is to provide administrative details and procedures for the care and treatment of newborns identified with phenylketonuria, hypothyroidism, galactosemia, congenital adrenal hyperplasia, hearing loss, hemoglobinopathy, biotinidase deficiency, cystic fibrosis, aminoacidopathies, fatty acid oxidation disorders, organic acidurias and acidemias, critical congenital heart disease and other heritable diseases.

Authors: Thomas M. Miller, M.D., Lucinda G. Ashley, R.N.-B.C.

Statutory Authority: Code of Ala. 1975, §§ 22-2-2, 22-20-3.

History: Filed December 21, 1987. **Amended:** Filed September 18, 2002; effective October 23, 2002. **Repealed and New Rule:** Filed December 17, 2003; effective January 21, 2004. **Amended:** Filed December 17, 2007; effective January 21, 2008. **Amended:** Filed May 17, 2013; effective June 21, 2013.

420-10-1-.02 Definitions.

(a) **Phenylketonuria** - A congenital disease due to a deficit in the metabolism of the amino acid phenylalanine.

(b) **Hypothyroidism** - A deficiency of thyroid gland activity with underproduction of thyroxin or the condition resulting from it.

(c) **Hemoglobinapathy** - Any hemoglobin phenotype which is other than AA.

(d) **Physician of Record** - The physician who requests the test.

(e) **Galactosemia** - An inherited error in the metabolism of galactose.

(f) **Congenital adrenal hyperplasia** - an inherited error in steroid biosynthesis.

(g) **Hearing loss** - the total or partial inability to hear sound in one or both ears.

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(h) **Biotinidase deficiency** – inherited deficiency caused by the lack of an enzyme involved in biotin synthesis.

(i) **Amino acid disorders** – inherited disorders in amino acid metabolism.

(j) **Fatty acid oxidation disorders** – inherited disorders in fatty acid metabolism.

(k) **Organic acid disorders** – inherited disorders in organic acid metabolism.

(l) **Cystic Fibrosis** – inherited disorder caused by a defective protein (cystic fibrosis transmembrane regulator, CFTR) involved in the salt balance of the body.

(m) **Critical Congenital Heart Disease (CCHD)** - a subset of congenital heart defects characterized by a diminished availability of oxygen to the body tissues that causes severe and life-threatening symptoms and requires intervention within the first days or first year of life.

Authors: Thomas M. Miller, M.D., William J. Callan, Ph.D., Lucinda G. Ashley, R.N.-B.C.

Statutory Authority: Code of Ala. 1975, §§ 22-2-2, 22-20-3.

History: Filed December 21, 1987; **Amended:** Filed September 21, 1992; effective October 26, 1992. **Amended:** Filed September 18, 2002; effective October 23, 2002. **Repealed and New Rule:** Filed December 17, 2003; effective January 21, 2004. **Amended:** December 17, 2007; effective January 21, 2008. **Amended:** Filed May 17, 2013; effective June 21, 2013.

420-10-1-.03 Designation of Additional Heritable Diseases. The State Board of Health hereby designates the following as a heritable disease subject to testing, reporting and notification requirements herein below specified. Phenylketonuria, hypothyroidism, galactosemia, congenital adrenal hyperplasia, hearing loss, hemoglobinapathy, biotinidase deficiency, cystic fibrosis, aminoacidopathies, fatty acid oxidation disorders and organic acidurias and acidemias, CCHD and other heritable disorders.

Authors: Thomas M. Miller M.D., William J. Callan, Ph.D., Lucinda G. Ashley, R.N.-B.C.

Statutory Authority: Code of Ala. 1975, §§ 22-2-2, 22-20-3.

History: Filed December 21, 1987; **Amended:** Filed September 21, 1992; effective October 26, 1992. **Repealed and New Rule:** Filed December 17, 2003; effective January 21, 2004. **Amended:** December 17, 2007; effective January 21, 2008. **Amended:** Filed May 17, 2013; effective June 21, 2013.

420-10-1-.04 Reporting And Notification.

(1) The Alabama Department of Public Health shall report all results of phenylketonuria, hypothyroidism, galactosemia, congenital adrenal hyperplasia, hearing loss, hemoglobinopathy, biotinidase deficiency, cystic fibrosis, aminoacidopathies, fatty acid oxidation disorders, organic acidurias and acidemias and other heritable disease

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testing to the submitting health care provider. Test results on transferred infants may be made available to both the transferring and receiving facilities.

(2) The submitting health care provider shall report all results, including positives, suspected positive results, and unsatisfactory specimens, to the physician of record (the physician indicated on the collection form) of the newborns tested and shall use such forms and follow such guidelines as shall be determined by the State Health Officer. The health care provider shall report the results of any hearing tests performed on the newborns to the Alabama Department of Public Health and shall use such forms and follow such guidelines as shall be determined by the State Health Officer.

(3) The Alabama Department of Public Health may release results of newborn screening tests, including hearing screening results, to any physician registered with the Alabama Voice Response System under the terms and conditions of the system without a signed release from the parent or guardian.

(4) The submitting health care provider shall screen all newborns in well baby nurseries for CCHD using pulse oximetry and shall use such forms and follow such guidelines as shall be determined by the State Health Officer.

(5) The submitting health care provider shall report the results of any failed pulse oximetry screening results to the Alabama Department of Public Health and shall use such forms and follow such guidelines as shall be determined by the State Health Officer.

Authors: Thomas M. Miller, M.D., William J. Callan, Ph.D., Lucinda G. Ashley, R.N.-B.C.

Statutory Authority: Code of Ala. 1975, §§ 22-2-2,22-20-3. **History:** Filed December 21, 1987. **Amended:** Filed September 21, 1995; effective October 26, 1992. **Amended:** Filed October 24, 1995; effective November 29, 1995. **Amended:** Filed September 18, 2002; effective October 23, 2002. **Repealed and New Rule:** Filed December 17, 2003; effective January 21, 2004. **Amended:** December 17, 2007; effective January 21, 2008. **Amended:** Filed May 17, 2013; effective June 21, 2013.

420-10-1-.05 Counseling And Management.

(a) The Alabama Department of Public Health shall make contact with the physician of record and the parent/guardian of newborns who test positive, for phenylketonuria, hypothyroidism, galactosemia, congenital adrenal hyperplasia, hearing loss, hemoglobinopathy, biotinidase deficiency, cystic fibrosis, aminoacidopathies, fatty acid oxidation disorders, organic acidurias and acidemias and other heritable disorders to notify them of positive test results and ascertain whether or not these newborns are under the care of a private physician. Additionally, the Alabama Department of Public Health shall make contact with the physician of record and the parent/guardian to advise them of the services available through the Alabama Department of Public Health. Newborns who are under the care of a private physician may additionally utilize these same services. The Alabama Department of Public Health may make contact with the family to make their services available or may assist the family in obtaining the services of a private physician. Services include health assessments, treatment, and referrals to tertiary care centers.

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(b) The Alabama Department of Public Health shall make contact with the submitting health care provider of newborns with failed pulse oximetry results to verify that appropriate screening, referral, and intervention services have been provided and if needed, may assist in obtaining the services. Services include health assessments, treatment, and referrals to tertiary care centers.

Authors: Thomas M. Miller, M.D., William J. Callan, Ph.D., Lucinda G. Ashley, R.N.-B.C.
Statutory Authority: Code of Ala. 1975, §§ 22-20-3.
History: Filed December 21, 1987. **Amended:** Filed September 21, 1992; effective October 26, 1992. **Amended:** Filed September 18, 2002; effective October 23, 2002. **Repealed and New Rule:** Filed December 17, 2003; effective January 21, 2004. **Amended:** December 17, 2007; effective January 21, 2008. **Amended:** Filed May 17, 2013; effective June 21, 2013.

420-10-1-.06 Fees. The Board shall assess and collect newborn screening fees from hospitals and birthing centers or third party payors. The newborn screening fee shall be set by the State Committee of Public Health based on the schedule of laboratory fees established by the Centers for Medicare and Medicaid Services (CMS) for use by Medicare and Medicaid. The Board shall bill the Medicaid Agency for Medicaid eligibles.

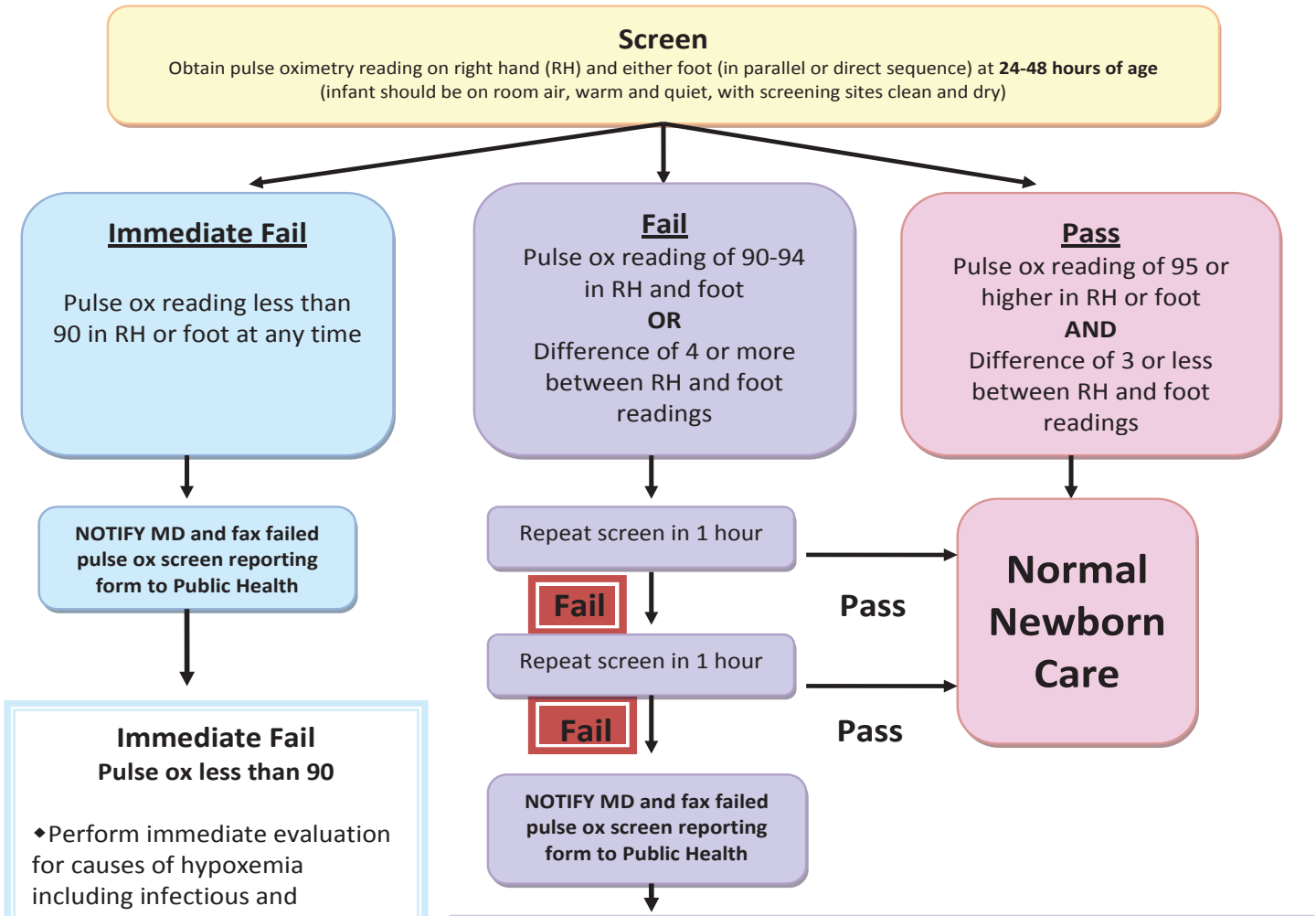
(1) Hospitals classified as "rural" by CMS or which have less than 105 beds and are located at least twenty (20) miles from the nearest acute care facility with obstetrical capabilities may have newborn screening fees waived for non-Medicaid eligible patients where there is no third party payor for such fees. The State Health Officer shall annually submit a list of hospitals to the Board which are eligible for waiver of fees.

(2) Additional reasonable and necessary fees may be charged to other payors by the hospital or physician in connection with this rule. The State Health Officer may waive fees deemed uncollectible because of a patient's inability to pay.

(3) There shall be only one (1) fee per birth collected from a hospital by the Board.

Authors: Lloyd Hofer, M.D., William J. Callan, Ph.D.
Statutory Authority: Code of Ala. 1975, §§ 22-20-3.
History: Filed February 19, 1992. **Amended:** Filed September 21, 1992; effective October 26, 1992. **Repealed and New Rule:** Filed December 17, 2003; effective January 21, 2004.

Pulse Oximetry Screening Algorithm



Immediate Fail

Pulse ox less than 90

- Perform immediate evaluation for causes of hypoxemia including infectious and pulmonary pathology.
- If no other etiology is found, immediate echocardiogram interpreted by a pediatric cardiologist is indicated. This may require transfer to an NICU with pediatric cardiology services.

Failed Screen

Pulse ox 90-94 or RH/foot difference of 4 or more x 3

- Perform comprehensive evaluation for causes of hypoxemia including infectious and pulmonary pathology.
- If no other etiology is found, consultation with pediatric cardiology or neonatology is indicated to arrange for a diagnostic echocardiogram to be interpreted by a pediatric cardiologist. This may require telemedicine, transfer to an NICU with pediatric cardiology services, or discussion with cardiology services to schedule a timely outpatient echocardiogram. Physician to physician communication recommended.

- This screening algorithm should not take the place of clinical judgment or customary clinical practice.
- A negative screen does not rule out heart disease.
- Optimal results are obtained using a motion-tolerant pulse oximeter that reports functional oxygen saturation, has been validated in low perfusion conditions, has been cleared by the FDA for use in newborns, has a 2% root mean-square accuracy, and is calibrated regularly.

- For more information see: Kemper, AR, Mahle, WT, Martin, GR et al; **Strategies for Implementing Screening for Congenital Heart Disease.** ***Pediatrics.*** 2011. available at: http://pediatrics.aappublications.org/content/early/2011/10/06/peds.2011-1317

Revised 6/13/12

Failed Pulse Ox Screen Reporting Form

PLACE LABEL OR WRITE-IN INFORMATION

Medical Record # _____

Patient Name: Last _____ First _____

Mother's Name: _____ Date of Birth _____/_____/_____

Hospital: _____ Medical Provider: _____

Alabama Newborn Screening Program

Fax failed screens to **334-206-3791**

Age at Initial Screening: _____ hours

Initial Screening:

Time _____

Pulse Ox Saturation of Right Hand _____

Pulse Ox Saturation of Foot _____

Difference between right hand and foot _____ ☐ Fail

Second Screening (*1 hour following initial screen if fail initial screen*)

Time _____

Pulse Ox Saturation of Right Hand _____

Pulse Ox Saturation of Foot _____

Difference between right hand and foot _____ ☐ Fail

Third Screening *(1 hour following second screening if fail second screen)*

Time _____

Pulse Ox Saturation of Right Hand _____

Pulse Ox Saturation of Foot _____

Difference between right hand and foot_____ ☐ Fail

Other etiology identified: ☐ Pulmonary ☐ Infection ☐ Unknown ☐ Other: _____

Transferred: _____

Provider referred to: _____

Screener's First Initial/Last Name: _____Date: _____/_____/_____

The following pages provide ACT Sheets which are available through the American College of Medical Genetics at www.acmg.net.

ACT Sheets provide short term actions a health professional should follow in communicating with the family and determining the appropriate steps in the follow-up of the infant that has screened positive.

American College of Medical Genetics ACT SHEET

Newborn Screening ACT Sheet
[Absent/ Reduced Biotinidase Activity]
Biotinidase Deficiency

Differential Diagnosis: Biotinidase deficiency (complete and partial); see C5-OH acylcarnitine for non-biotinidase associated conditions.

Condition Description: A multiple carboxylase deficiency resulting from a reduction in available biotin secondary to deficient activity of the biotinidase enzyme.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact family to inform them of the newborn screening result and ascertain clinical status.
- Evaluate infant if poor feeding, lethargy, or hypotonia are present.
- Consultation/referral to a metabolic specialist to determine appropriate follow-up.
- Undertake confirmatory testing in consultation with a metabolic specialist.
- Emergency treatment if symptomatic.
- Report findings to newborn screening program

Diagnostic Evaluation: Enzyme assay for biotinidase in serum or plasma reveals low activity. False positive findings are usually a processing/shipping problem. Urine organic acid analysis may show normal or increased 3-hydroxyisovaleric acid and 3-methylcrotonylglycine. Plasma acylcarnitine analysis may show normal or increased C5-OH acylcarnitine.

Clinical Considerations: The neonate is usually asymptomatic but episodic hypoglycemia, lethargy, hypotonia, and mild developmental delay can occur at any time from the neonatal period through childhood. Untreated biotinidase deficiency leads to developmental delay, seizures, alopecia, and hearing deficits. Biotin treatment is available and highly effective.

Local Resources:

University of Alabama at Birmingham, Department of Genetics, S. Lane Rutledge, M.D.
Newborn Screening Contact: Alicia Roberts, R.D.
Phone: 205-996-6983
1530 3rd Avenue South, Kaul 241
Birmingham, AL 35294

Newborn Screening ACT Sheet
[Elevated 17-hydroxyprogesterone (17-OHP)]
Congenital Adrenal Hyperplasia (CAH)

Differential Diagnosis: Congenital Adrenal Hyperplasia (CAH), 21-OH deficiency; stress or prematurity are possible secondary causes of increased 17-OHP.

Condition Description: Lack of adequate adrenal cortisol and aldosterone, and increased androgen production.

YOU SHOULD TAKE THE FOLLOWING ACTIONS IMMEDIATELY:

- Contact family to inform them of the newborn screening result and ascertain clinical status.
- Consult with pediatric endocrinologist, having the following information available (sex, age at NBS sampling, birth weight) and refer, if needed.
- Examine the newborn (ambiguous genitalia or non palpable testes, lethargy, vomiting, poor feeding).
- Initiate timely confirmatory/diagnostic testing as recommended by specialist.
- Emergency treatment as indicated (e.g. IV fluids, IM/IV hydrocortisone).
- Educate family about signs, symptoms and need for urgent treatment of adrenal crisis.
- Report findings to newborn screening program.

Diagnostic Evaluation: Diagnostic tests include serum 17-0HP (increased), serum electrolytes (reduced sodium and increased potassium), and blood glucose (reduced). Additional tests may be recommended by the specialist.

Clinical Considerations: Ambiguous genitalia in females who may appear to be male with non-palpable testes. Infants with Congenital Adrenal Hyperplasia are at risk for life-threatening adrenal crises, shock, and death in males and females. Finding could also be a false positive associated with stress or prematurity.

Local Referral Sites:

Children's of Alabama
Gail Mick, M.D.
Pediatric Endocrinology
1600 7th Avenue South, CPP 230
Birmingham, AL 35233
(205) 638-9107

University of South Alabama Medical Center
Anne Marie Kaulfers, M.D.
Pediatric Endocrinology
1504 Springhill Avenue, 4th Floor
Mobile, AL 36604
(251) 405-5147

Age of Collection and Birth Weight:	Results:	Action:
Full-term (>2500 grams) collected ≥ 24 hours of age	CAH > 65	1. Collect an immediate repeat newborn screen. 2. Evaluate infant and consult with pediatric endocrinologist if considered appropriate.
Preterm (<2500 grams) collected ≤ 24 hours of age	CAH > 150	
Any Age and Birthweight	2 borderline CAH (45-65)	1. Diagnostic testing to include 17-OHP is recommended.

American College of Medical Genetics **ACT SHEET**

Newborn Screening ACT Sheet
[Elevated TSH (Primary TSH test)]
Congenital Hypothyroidism

Differential Diagnosis: Primary congenital hypothyroidism (CH); transient CH.

Condition Description: Lack of adequate thyroid hormone production..

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact family to inform them of the newborn screening test result.
- Consult pediatric endocrinologist; refer to endocrinologist, if considered appropriate.
- Evaluate infant (see clinical considerations below).
- Initiate timely confirmatory/diagnostic testing as recommended by the specialist.
- Initiate treatment as recommended by consultant as soon as possible.
- Educate parents/caregivers that hormone replacement prevents mental retardation.
- Report findings to state newborn screening program.

Diagnostic Evaluation: Diagnostic tests should include serum free T4 and thyroid stimulating hormone (TSH); consultant may also recommend total T4 and T3 resin uptake. Test results include reduced free T4 and elevated TSH in primary hypothyroidism; if done, reduced total T4 and low or normal T3 resin uptake

Clinical Considerations: Most neonates are asymptomatic, though a few can manifest some clinical features, such as prolonged jaundice, puffy facies, large fontanels, macroglossia and umbilical hernia. Untreated congenital hypothyroidism results in developmental delay or mental retardation and poor growth.

Local Referral Sites

Birmingham:
Children's of Alabama
Gail Mick, M.D.
Pediatric Endocrinology
1600 7th Avenue South, CPP 230
Birmingham, AL 35233
205-638-9107

Mobile:
University of South Alabama Medical Center
Anne Marie Kaulfers, M.D.
Pediatric Endocrinology
1504 Springhill Avenue, 4th Floor
Mobile, AL 36604
251-405-5147

MEDICAL EMERGENCY:

Age of Collection:	Results:	Action Plan:
Sample collected > 24 hours of age	TSH≥40	1. Contact and inform the parent of need for immediate confirmatory testing - **immediate serum Free T4 and TSH.** 2. Assess for sign and symptoms. If symptomatic, refer to a local medical provider as soon as possible. 3. Notify pediatric endocrinologist (see above).
Sample collected < 24 hours of age	TSH > 150	

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Newborn Screening ACT Sheet
[Elevated IRT +/- DNA]
Cystic Fibrosis

Differential Diagnosis: Cystic fibrosis (CF); gastrointestinal abnormalities are also causes of increased IRT.

Condition Description: The cystic fibrosis transmembrane conductance regulator (CFTR) protein regulates chloride transport that is important for function of lungs, upper respiratory tract, pancreas, liver, sweat glands, and genitourinary tract. CF affects multiple body systems and is associated with progressive damage to respiratory and digestive systems.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact family to inform them of the newborn screening result and to ascertain clinical status (meconium ileus, failure to thrive, recurrent cough, wheezing and chronic abdominal pain).
- Contact CF Center for consultation with CF specialist.
- Determine sweat chloride (sweat test) through experienced sweat test laboratory.
- If cystic fibrosis is confirmed, clinical evaluation and genetic counseling are indicated.
- Report findings to newborn screening program.

Diagnostic Evaluation: Varies with screening test. Infants with highly elevated immunoreactive trypsinogen (IRT) may be considered screen positive. Elevated IRT results are followed with second tier tests for either additional IRT measurement or CFTR mutation panels. If screen positive, follow up with sweat chloride test to confirm diagnosis.

Clinical Considerations: Deficient chloride transport in lungs causes production of abnormally thick mucous leading to airway obstruction, neutrophil dominated inflammation and recurrent and progressive pulmonary infections. Pancreatic insufficiency found in 80 – 90% of cases. Some males may be infertile in adulthood.

Local Cystic Fibrosis Foundation Accredited Care Center (meets nationally accepted standards):

Hector Gutierrez, M.D.
Pediatric Pulmonology
Children's of Alabama/UAB (Pediatric) CF Care Center
1600 7th Avenue South, ACC 620
Birmingham, AL 35233
Contact: Staci Thrasher Self, MSW, LGSW
Phone: 205-638-5494

Alabama Newborn Screening Result:	Alabama Recommended Actions:
• Ultrahigh IRT > 170 • Collected < 24 hours of age • No *disease causing mutations reported • May have a **benign polymorphism (common variant) reported	Immediate repeat of newborn screen
• Ultrahigh IRT > 170 • Collected > 24 hours of age • No mutations of any type reported	Diagnostic sweat test indicated
• IRT value in the top 4% of levels received each day • One or more DNA mutations identified other than a **benign polymorphism (common variant)	Diagnostic sweat test indicated
*Disease-Causing Mutation – a difference in the genetic code that leads to an abnormal protein **Benign variant – change in the DNA code that has been studied and does not cause disease	

American College of Medical Genetics ACT SHEET

Newborn Screening ACT Sheet
[FS]
Sickle Cell Anemia (HbSS Disease or HbS/Beta Zero Thalassemia)

Differential Diagnosis: Homozygous sickle cell disease (Hb SS), sickle beta-zero thalassemia, or sickle hereditary persistence of fetal hemoglobin (Hb S-HPFH).

Condition Description: A red blood cell disorder characterized by presence of fetal hemoglobin (F) and hemoglobin S in the absence of hemoglobin A. The hemoglobins are listed in order of the amount of hemoglobin present (F>S). This result is different from FAS which is consistent with sickle carrier.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact the family to inform them of the screening result.
- Consult a specialist in hemoglobin disorders; refer if needed.
- Evaluate infant and assess for splenomegaly; do complete blood count (CBC) with mean corpuscular volume (MCV), and reticulocyte count.
- Order hemoglobin profile analysis (usually performed by electrophoresis).
- Initiate timely confirmatory/diagnostic testing as recommended by consultant.
- Initiate daily penicillin VK (125mg po bid) prophylaxis and other treatment as recommended by the consultant.
- Educate parents/caregivers regarding the risk of sepsis, the need for urgent evaluation if fever of ≥ 38.5° C (101° F) or signs and symptoms of splenic sequestration.

Diagnostic Evaluation: CBC, MCV, and reticulocyte count. Hemoglobin separation by electrophoresis, isoelectric focusing or high performance liquid chromatography (HPLC) shows FS pattern. DNA studies may be used to confirm genotype. Sickledex is not appropriate for confirmation of diagnosis in infants.

Clinical Considerations: Newborn infants are usually well. Hemolytic anemia and vaso-occlusive complications develop during infancy or early childhood. Complications include life-threatening infection, splenic sequestration, pneumonia, acute chest syndrome, pain episodes, aplastic crisis, dactylitis, priapism, and stroke. Comprehensive care including family education, immunizations, prophylactic penicillin, and prompt treatment of acute illness reduces morbidity and mortality. S-HPFH is typically benign.

Local Referral Sites:

Children's of Alabama
Thomas Howard, M.D.
Pediatric Hematology/Oncology
1600 7th Avenue South, ACC 512
Birmingham, AL 35233
Sharon Carlton: 205-638-2390

University of South Alabama Sickle Cell Center
Felicia Wilson, M.D.
Pediatric Hematology/Oncology
1504 Springhill Avenue, Suite 5230
Mobile, AL 36604
Andretta McCovey: 251-405-5121

St. Jude's Clinic
Carolyn Russo, M.D.
Pediatric Hematology
910 Adams Street, Suite 310
Huntsville, AL 35801
256-265-5833

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Newborn Screening ACT Sheet
[FSC]
Hemoglobin SC Disease (HbSC)

Differential Diagnosis: Hemoglobin SC disease.

Condition Description: A red cell disorder characterized by the presence of fetal hemoglobin (F) and hemoglobins S and C in the absence of Hb A. The hemoglobins are listed in order of the amount of hemoglobin present (F>S>C). This result is different from FAS which is consistent with sickle carrier.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact the family to inform them of the screening result.
- Consult a specialist in hemoglobin disorders; refer if needed.
- Evaluate infant and assess for splenomegaly; do complete blood count (CBC).
- Order hemoglobin profile analysis (usually performed by electrophoresis).
- Initiate timely confirmatory/diagnostic testing as recommended by consultant.
- Initiate treatment as recommended by consultant.
- Educate parents/caregivers regarding the risk of sepsis, the need for urgent evaluation for fever of ≥38.5° C (101° F) and signs and symptoms of splenic sequestration.
- Report findings to newborn screening program.

Diagnostic Evaluation: CBC. Hemoglobin separation by electrophoresis, isoelectric focusing or high performance liquid chromatography (HPLC) shows FSC. DNA studies may be used to confirm genotype.

Clinical Considerations: Newborn infants are usually well. Hemolytic anemia and vaso-occlusive complications develop during infancy or early childhood. Complications include life-threatening infection, splenic sequestration, pneumonia, acute chest syndrome, pain episodes, aplastic crisis, dactylitis, priapism, and stroke. Comprehensive care including family education, immunizations, prophylactic penicillin, and prompt treatment of acute illness reduces morbidity and mortality.

Local Referral Sites:

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Newborn Screening ACT Sheet
[FSA]
Hemoglobin S/Beta plus Thalassemia (HbSβ+ Disease)

Differential Diagnosis: Sickle beta plus thalassemia. The hemoglobins are listed in order (F>S>A) of the amount of hemoglobin present. This result is different from FAS which is consistent with sickle carrier (trait).

Condition Description: Individuals with sickle beta plus thalassemia, a form of sickle cell disease, are compound heterozygotes for the Hb S and beta-thalassemia mutations in the beta-globin genes.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact the family to inform them of the screening result.
- Perform a physical exam on the infant and assess for splenomegaly.
- Obtain a blood sample for confirmatory testing and a complete blood count (CBC) with reticulocyte count.
- Order hemoglobin profile analysis (usually performed by electrophoresis).
- Initiate penicillin (PenVK 125mg po bid) prophylaxis.
- Educate parents/caretakers regarding the risk of sepsis and advise that infant be immediately evaluated if a fever of ≥ 38.5° C (101° F) is present.
- Contact a specialist in hemoglobin disorders for consultation on diagnostic evaluation and management.

Diagnostic Evaluation: CBC. Hemoglobin separation by electrophoresis, isoelectric focusing or high performance liquid chromatography (HPLC) shows FSA. DNA studies may be used to confirm genotype.

Clinical Considerations: Infants are usually normal at birth. Later potential clinical problems include mild to moderate hemolytic anemia, life-threatening infection, vaso-occlusive pain episodes, dactylitis, and chronic organ damage. Prompt treatment of infection and splenic sequestration is associated with decreased mortality in the first three years of life.

Local Referral Sites:

Children's of Alabama
Thomas Howard, M.D.
Pediatric Hematology/Oncology
1600 7th Avenue South, ACC 512
Birmingham, AL 35233
Sharon Carlton: 205-638-2390

University of South Alabama Sickle Cell Center
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Newborn Screening ACT Sheet
[FAS]
Sickle Cell Carrier (HbAS)

Differential Diagnosis: Sickle Cell Carrier. The hemoglobins are listed in order of the amount of hemoglobin present (F>A>S). This result is different from FS which is consistent with sickle cell anemia or sickle cell beta zero thalassemia (HbS B^0), or FSA which is consistent with sickle beta-plus thalassemia.

Condition Description: Generally benign genetic carrier state (trait) characterized by the presence of fetal hemoglobin (F), and hemoglobin A and S.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact the family to inform them of the screening result to offer education and reassurance that infants and young children do not have clinical problems related to the carrier state for hemoglobin S.
- Repeat screen or confirm result by alternate assay.
  - Order hemoglobin profile analysis (usually performed by electrophoresis).
- Offer family members referral for hemoglobin disorder testing and genetic counseling.
- Report findings to state newborn screening program.

Diagnostic Evaluation: Hemoglobin separation by electrophoresis, isoelectric focusing or high performance liquid chromatography (HPLC) shows FAS. DNA studies may be used to confirm genotype. Sickledex is not adequate for confirmation of the diagnosis.

Clinical Considerations: Newborn infants are usually normal. Prognosis is good, with a normal life expectancy. Carriers are at risk for having children affected by sickle cell disease. Older children and adults may have hematuria. Splenic infarction and an increased risk of sudden death associated with severe hypoxia, extreme physical exertion and dehydration have been reported.

Local Referral Sites:

Children's of Alabama
Thomas Howard, M.D.
Pediatric Hematology/Oncology
1600 7th Avenue South, ACC 512
Birmingham, AL 35233
Sharon Carlton: 205-638-2390

University of South Alabama Sickle Cell Center
Felicia Wilson, M.D.
Pediatric Hematology/Oncology
1504 Springhill Avenue, Suite 5230
Mobile, AL 36604
Andretta McCovey: 251-405-5121

St. Jude's Clinic
Carolyn Russo, M.D.
Pediatric Hematology
910 Adams Street, Suite 310
Huntsville, AL 35801
256-265-5833

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Newborn Screening ACT Sheet
[FA + Barts Hb]
Alpha (α) Thalassemia (phenotype varies with % Barts Hb)

Differential Diagnosis: Hemoglobin A/Barts, alpha thalassemia carrier, hemoglobin H disease, alpha thalassemia major.

Condition Description: A red blood cell disorder characterized by presence of fetal hemoglobin (F) and hemoglobin A, as well as one or more alpha globin mutations (resulting in hemoglobin Barts).

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact family family to inform them of the screening result.
- Evaluate infant, assess for splenomegaly, and do complete blood count (CBC) for Hb, and mean corpuscular volume (MCV).
- Order hemoglobin profile analysis (usually performed by electrophoresis).
- Consult a specialist in hemoglobin disorders; refer if needed.
- Initiate timely confirmatory/diagnostic testing as recommended by consultant.
- Report findings to newborn screening program.

Diagnostic Evaluation: CBC and MCV. Hemoglobin separation by electrophoresis, isoelectric focusing (IEF), or high performance liquid chromatography (HPLC), shows FA+Barts pattern. DNA studies may be used to confirm genotype.

Clinical Considerations: Severity depends on the number of the four alpha globin genes that are lost. Presence of less than 25% Hb Barts indicates loss of one (silent carrier) or two (alpha thalassemia trait) genes. Individuals are asymptomatic with laboratory features that are normal or may resemble iron deficiency anemia. Hemoglobin Barts above 25% in the newborn indicates a hemoglobin H disease, a clinically significant form of alpha thalassemia, is likely. Deletion or dysfunction of 3 of the 4 alpha globin genes manifests as Hb H disease. Hb H disease is characterized by splenomegaly and anemia that may require intermittent transfusions. Absence of all four alpha globin genes results in hydrops fetalis and is usually fatal, in utero or shortly after birth.

Sickle Cell Local Referral Sites:

Children's of Alabama
Thomas Howard, M.D.
Pediatric Hematology/Oncology
1600 7th Avenue South, ACC 512
Birmingham, AL 35233
205-638-2390

University of South Alabama Sickle Cell Center
Felicia Wilson, M.D.
1504 Springhill Avenue, Suite 5230
Mobile, AL 36604
251-415-5172 or 251-405-5147 (3)

St. Jude's Clinic at Huntsville Hospital
Carolyn Russo, M.D.
910 Adams Street, Suite 310
Huntsville, AL 35801
256-265-5833 or 1-866-595-5449

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Newborn Screening ACT Sheet
[Absent/Reduced Galactose-1-Phosphate Uridyltransferase (GALT)]
Classical Galactosemia

Differential Diagnosis: Galactosemia (galactose-1-phosphate uridyltransferase [GALT] deficiency); GALT heterozygotes; GALT variants; artifactual reductions due to enzyme inactivation by high temperature and/or humidity.

Condition Description: In galactosemia, GALT deficiency results in accumulation of galactose-1-phosphate (Gal-1-P) and galactose, causing multi-organ disease.

YOU SHOULD TAKE THE FOLLOWING ACTIONS IMMEDIATELY:

- Contact family to inform them of the newborn screening result, ascertain clinical status, arrange immediate clinical evaluation, stop breast or cow's milk and initiate non-lactose feeding (powder-based soy formula).
- Consult with metabolic specialist; refer if considered appropriate.
- Evaluate the infant (jaundice, poor feeding, vomiting, lethargy, bulging fontanel, and bleeding) and arrange diagnostic testing as directed by metabolic specialist.
- Emergency treatment as recommended by metabolic specialist. If baby is sick, stop cow's milk and initiate non-lactose feedings.
- Educate family about importance of diet change.
- Report findings to newborn screening program.

Diagnostic Evaluation: Quantification of erythrocyte galactose-1-phosphate (Gal-1-P) and GALT. Classical galactosemia shows <1% GALT activity and markedly increased Gal-1-P. Transfusions in infant can invalidate the results of erythrocyte enzyme assays. Enzyme variants may be distinguished by GALT electrophoresis or mutation analysis.

Clinical Considerations: Classical galactosemia presents in the first few days of life and may be fatal without treatment. Signs include poor feeding, vomiting, jaundice and, sometimes, lethargy and/or bleeding. Neonatal *E. coli* sepsis can occur and is often FATAL. Treatment is withdrawal of milk and, if symptomatic, emergency measures.

Local Resources:

University of Alabama at Birmingham, Department of Genetics, S. Lane Rutledge, M.D.
Newborn Screening Contact: Alicia Roberts, R.D.
Phone: 205-996-6983
1530 3rd Avenue South, Kaul 241
Birmingham, AL 35294

*If the initial newborn screening galactosemia result is abnormal, stop breast or infant formula feeding and initiate a non-galactose feeding (powder-based soy formula).

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Newborn Screening ACT Sheet
[Congenital Hearing Loss >30db]
Congenital Hearing Loss

Differential Diagnosis: Extensive. Includes 40% environmental (mostly bacterial/viral) and 60% genetic (30% syndromal and 70% non-syndromal representing over 100 genes).

Condition Description: Defined as hearing loss that is permanent, bilateral or unilateral, sensorineural or conductive, and averaging loss of 30 decibels or more in the frequency range important for speech recognition.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact family to inform them of the newborn screening result.
- Ensure coordinated and comprehensive multidisciplinary hearing loss evaluation and care.
- Initiate timely diagnostic evaluation by a multidisciplinary hearing loss team, including evaluation by a genetic specialist.
- Report findings to state Early Hearing Detection and Intervention (EHDI) program.

Diagnostic Evaluation: Hearing loss is confirmed and followed up by a comprehensive hearing loss team evaluation and testing for an etiologic diagnosis. Testing algorithms are prioritized around family history and likelihood of a syndromal condition. If familial and/or non-syndromal, *GJB2* (Connexin 26) and *GJB6* (Connexin 30) gene testing is done. Cytomegalovirus (CMV) and mitochondrial etiologies are also possible. Confirmatory work should be completed by age 3 months and early intervention services initiated before 6 months of age.

Clinical Considerations: Hearing loss may indicate a genetic syndrome with involvement of other organ systems. Untreated hearing loss can result in lifelong deficits in speech and language development, so it is critical that all infants who fail newborn screening have follow-up testing.

Rovetta Hanna BSN, RN
Early Hearing Detection and Intervention Coordinator
Alabama Department of Public Health
Newborn Screening Program
334-206-2944

The Alabama Department of Public Health complies with the guidelines set by the Joint Committee on Infant Hearing (JCIH) for newborn hearing screening and follow-up. These guidelines specify that infants who fail an automated auditory brainstem response (AABR) hearing screen must have a repeat AABR for follow-up screening, and both ears should be tested even if only one ear failed.

Infants who fail an initial otoacoutstic emissions (OAE) hearing screen may be re-screened with either an OAE or AABR, since the AABR is a more sensitive and comprehensive test. No more than two valid initial attempts should be performed. If the infant fails both, then a referral for a diagnostic hearing evaluation should be made as soon as possible.

For further information, please visit the following link: www.jcih.org.

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Newborn Screening ACT Sheet
[Elevated C8 with Lesser Elevations of C6 and C10 Acylcarnitine]
Medium-chain Acyl-CoA Dehydrogenase (MCAD) Deficiency

Differential Diagnosis: Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency.

Condition Description: MCAD deficiency is a fatty acid oxidation (FAO) disorder. Fatty acid oxidation occurs mainly during prolonged fasting and/or periods of increased energy demands (fever, stress), when energy production relies increasingly on fat metabolism. In an FAO disorder, fatty acids and potentially toxic derivatives accumulate because of a deficiency in one of the mitochondrial FAO enzymes.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact family to inform them of the newborn screening result and ascertain clinical status (poor feeding, vomiting, lethargy).
- Consult with pediatric metabolic specialist.
- Evaluate the newborn (poor feeding, lethargy, hypotonia, hepatomegaly). If signs are present or infant is ill, transport infant to hospital for emergency treatment that would include IV glucose and any further treatment in consultation with the metabolic specialist.
- If infant is normal initiate timely confirmatory/diagnostic testing, as recommended by specialist.
- Educate family about need for infant to avoid fasting and the need for immediate medical attention if the infant even becomes mildly ill (poor feeding, vomiting, or lethargy).
- Report findings to newborn screening program.

Diagnostic Evaluation: Plasma acylcarnitine analysis will show a characteristic pattern consistent with MCADD. Urine organic acid analysis may also show an abnormal profile. Diagnosis may be confirmed by mutation analysis of the MCAD gene.

Clinical Considerations: MCAD deficiency is usually asymptomatic in the newborn although it can present acutely in the neonate with hypoglycemia, metabolic acidosis, hyperammonemia, and hepatomegaly. MCAD deficiency is associated with high mortality unless treated promptly; milder variants exist. Hallmark features include vomiting, lethargy, and hypoketotic hypoglycemia. Untreated MCAD deficiency is a significant cause of sudden death.

Local Referral Site:

University of Alabama at Birmingham
Department of Genetics
S. Lane Rutledge, MD
1530 3rd Avenue South, Kaul 241
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205-996-6983

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Newborn Screening ACT Sheet
[Increased Phenylalanine]
Phenylketonuria (PKU)

Differential Diagnosis: Phenylketonuria (Classical PKU); non-PKU mild hyperphenylalaninemia; pterin defects; transient hyperphenylalaninemia.

Condition Description: In PKU the phenylalanine from ingested protein cannot be metabolized to tyrosine because of deficient liver phenylalanine hydroxylase (PAH). This causes elevated phenylalanine. Pterin defects result from deficiency of tetrahydrobiopterin (BH4), the cofactor for PAH and other hydroxylases. This produces not only increased phenylalanine but also neurotransmitter deficiencies.

YOU SHOULD TAKE THE FOLLOWING ACTIONS IMMEDIATELY:

- Contact family immediately to inform them of the newborn screening result.
- Consult with pediatric metabolic specialist.
- Evaluate the newborn and refer as appropriate.
- Initiate confirmatory/diagnostic tests in consultation with metabolic specialist.
- Provide the family with basic information about PKU and dietary management.
- Report findings to newborn screening program.

Diagnostic Evaluation: Plasma amino acid analysis which shows increased phenylalanine without increased tyrosine (increased phenylalanine:tyrosine ratio). Urine pterin analysis and red blood cell DHPR assay will identify pterin defects. Consider PAH mutation testing.

Clinical Considerations: Asymptomatic in the neonate. If untreated PKU will cause irreversible mental retardation, hyperactivity, autistic-like features, and seizures. Treatment will usually prevent these symptoms. Pterin defects cause early severe neurologic disease (developmental delay/seizures) and require specific therapy.

Local Referral Site:

University of Alabama at Birmingham
Department of Genetics
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205-996-6983

American College of Medical Genetics **ACT SHEET**

Newborn Screening ACT Sheet
[Decreased C0 and other Acylcarnitines]
Carnitine uptake Defect (CUD)

Differential Diagnosis: Carnitine uptake defect (CUD), maternal carnitine deficiency and prematurity.

Condition Description: CUD is caused by a defect in the carnitine transporter that moves carnitine across the plasma membrane. Reduced carnitine limits acylcarnitine formation preventing transport of long-chain fatty acids into mitochondria, thereby limiting energy production. Tissues with high energy needs (skeletal and heart muscle) are particularly affected.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact family to inform them of the newborn screening result and ascertain clinical status (poor feeding, lethargy, tachypnea).
- Consult with pediatric metabolic specialist.
- Evaluate the newborn (tachycardia, hepatomegaly, reduced muscle tone); initiate emergency treatment as indicated by metabolic specialist.
- Initiate timely confirmatory/diagnostic testing as recommended by specialist.
- Educate family about signs, symptoms, and need for urgent treatment if infant becomes ill.
- Report findings to newborn screening program.

Diagnostic Evaluation: Plasma carnitine analysis will reveal decreased free and total carnitine (C0) in plasma in an affected infant. If the total and free carnitine are normal in the infant, it may suggest a maternal carnitine deficiency and plasma carnitine analysis in the mother is indicated. Transporter assays in fibroblasts and *SLC22A5* (OCTN2 carnitine transporter) gene sequencing establish the diagnosis. Prematurity should be considered in the differential diagnosis.

Clinical Considerations: Carnitine transporter defect has a variable expression and variable age of onset. Characteristic manifestations include lethargy, hypotonia, hepatomegaly, and cardiac decompensation due to cardiomyopathy. Hypoglycemia is typical in acute episodes. These are rarely present in the neonatal period.

Local Referral Site:
S. Lane Rutledge, M.D.
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KAUL 210B
1530 3rd Avenue South
Birmingham, AL 35294-0024
Phone: 205-996-6983
Fax: 205-975-6390

Alabama Newborn Screening Program
Rachael Montgomery, BSN, RN 334-206-5955

PLEASE CHECK WITH US BEFORE RELAYING FOLLOW-UP NEWBORN SCREENING RESULTS TO THE FAMILY AS A REPEAT CARNITINE SCREENING MAY BE REPORTED AS NORMAL BUT STILL BE BELOW THE REPEAT CUTOFF VALUE.

Dr. Rutledge recommends the following for abnormal carnitine values: Alert level = carnitine less than 5.54

If equal to or more than 34 weeks gestation:
1. Send a plasma carnitine level (total/free carnitine).
2. A carnitine level should be obtained on an infant's mother for an infant with an initial low level whose total and free carnitine normalizes as this may suggest a maternal carnitine deficiency.

If less than 34 weeks gestation or sick infant in the NICU no matter gestational age:
1. Carnitine supplementation as directed.
2. Send a plasma carnitine level (total/free carnitine) after carnitine supplementation is discontinued.
3. May need further evaluation if abnormal levels persist.

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Carnitine Supplementation Protocol

Carnitine Supplementation for Sick Infant

1) Recommend carnitine supplementation as follows:

   a. Po/enteral 100 mg/kg/day divided TID x 3 days then 30mg/kg/day divided BID for 5-7 days

   b. IV 30mg/kg/day (can put with TPN or divided BID x 3 days) then 20mg/kg/day in TPN or divided BID for 5-7 days

2) Complete a plasma carnitine profile 5-7 days after carnitine supplementation is discontinued

3) May need to initiate metabolic evaluation if abnormal levels persists when infant on full feeding

Signature:_____ Date:_____

S. Lane Rutledge, M.D.
Professor Genetics, Pediatrics, and Neurology
University of Alabama at Birmingham

Low Carnitine Protocol
11/12/2014

American College of Medical Genetics **ACT SHEET**

Newborn Screening ACT Sheet
[Increased Tyrosine]
Tyrosinemia

Differential Diagnosis: Tyrosinemia I (hepatorenal); tyrosinemia II (oculocutaneous); tyrosinemia III; transient hypertyrosinemia; liver disease.

Condition Description: In the hepatorenal form, tyrosine (from ingested protein and phenylalanine metabolism) cannot be metabolized by fumarylacetoacetate hydrolase to fumaric acid and acetoacetic acid. The resulting fumarylacetoacetate accumulates and is converted to succinylacetone, the diagnostic metabolite, which is liver toxic and leads to elevated tyrosine. Tyrosinemias II and III are due to other defects in tyrosine degradation.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact family to inform them of the newborn screening result.
- Consult with pediatric metabolic specialist.
- Evaluate the newborn and refer as appropriate.
- Initiate confirmatory/diagnostic tests in consultation with metabolic specialist.
- Provide family with basic information about tyrosinemia.
- Report findings to newborn screening program.

Local Referral Site:

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Alabama Newborn Screening Program
Rachael Montgomery, BSN, RN 334-206-5955

**The most common type of tyrosinemia found by newborn screening is transient tyrosinemia. This is a harmless condition, often seen in newborns due to immaturity of one of the enzymes of tyrosine metabolism. Vitamin C is a cofactor for the enzyme and giving vitamin C to these children often normalizes enzyme activity and tryosine levels.*

Alert Value = tyrosine 445.75 or greater
Dr. Rutledge recommends the following for an elevated tyrosine level:

If greater than 34 weeks gestation and NOT on TPN at time of collection:
1. Obtain clinical status of infant
2. *Proceed with vitamin C protocol as directed if no liver involvement
3. If liver involvement, please order the following diagnostic labs: urine for succinylacetone, plasma amino acids, and liver function test. Please forward copies of results to newborn screening at fax (334) 206-3791.

If ON TPN at time of collection:
1. Obtain clinical status of infant
2. For infant with NO evidence of liver disease, collect a plasma amino acid two days or more off TPN.
3. If an infant has liver involvement, order a urine succinylacetone and plasma amino acids and fax results to the Newborn Screening Program at (334) 206-3791.

If infant is less than 34 weeks and NOT on TPN at time of collection:
1. *Proceed with vit C protocol as directed (see attached vitamin C protocol)

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Vitamin C Protocol

Protocol for Vitamin C (Ascorbic Acid) Administration R/T Elevated Tyrosine levels

Rationale:
Newborns may have immaturity of one of the enzymes of tyrosine metabolism. Vitamin C is a cofactor for the enzyme and giving Vitamin C to these children often normalizes enzyme activity and tyrosine levels.

1) SUGGESTED DOSING:
Give **POLYVISOL** at appropriate dose for baby's age and weight
Give p.o. daily x 3 weeks

OR

Vitamin C : 50mg p.o. daily x 3 weeks

2) **Complete a plasma amino acid profile one week after vitamin C administration is complete.**

It is important that diagnostic testing is repeated **AFTER** the infant has completed 3 weeks of vitamin C in order to make sure that the tyrosine level remains normal. **It could signify more serious underlying disorders if the level is not normal after vitamin C administration**.

Please contact the Newborn Screening Program with any questions:
Rachael Montgomery, BSN, RN 334-206-5955
Cindy Ashley, BSN, RN 334-206-2971

Signature:_____ Date:_____
S. Lane Rutledge, M.D.
Professor Genetics, Pediatrics, and Neurology
University of Alabama at Birmingham

Tyrosine Protocol
11/12/2014

Alabama Newborn Screening Program

Alabama's Listening!

UNIVERSAL NEWBORN HEARING SCREENING

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Alabama Department of Public Health Newborn Screening Program

www.adph.org/newbornscreening 1-866-928-6755