

ALABAMA



Newborn Screening PROGRAM



Newborn Screening Reference Manual for Medical Providers 2019

**Alabama Newborn Screening Laboratory
Bureau of Clinical Laboratories
8140 AUM Drive
P.O. Box 244018
Montgomery, AL 36124-4018
Phone: (334) 260-3400
www.alabamapublichealth.gov/bcl**

**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
Phone: 1-866-928-6755
www.alabamapublichealth.gov/newbornscreening**

TABLE OF CONTENTS

SECTION 1 - PROGRAM OVERVIEW

Alabama Newborn Screening Program	6
Health Insurance Portability and Accountability Act (HIPAA)	7
Alabama Newborn Screening Panel of Disorders	8
Medical Provider Responsibilities	9
Alabama Newborn Screening Medical Consultants	10
Secure Remote Viewer Instructions	11
Secure Remote Viewer Registration Form	12
Newborn Screening Education Material	13
Newborn Screening Refusal Form	14
Alabama Birthing Hospitals	15
Why Use An Accredited Cystic Fibrosis Center	16

SECTION 2 - SPECIMEN COLLECTION

One Drop, One Circle, One Time	18
Newborn Screening Blood Collection Guidelines	19-27
Sick Infant Blood Collection Guidelines	26
Hemoglobinopathy Repeats	28
Newborn Screening Collection Tips	29
Whatman® Neonatal Screening Reference Form	30-31
Whatman® Simple Spot Check Reference Form	32
Alabama Newborn Screening Kit Reorder Form	33
Newborn Screening Provider Updated Form	34

SECTION 3 - NEWBORN HEARING SCREENING

Newborn Hearing Screening Overview	36
Inpatient Newborn Hearing Screening Protocol	37
Joint Committee on Infant Hearing	38
Newborn Hearing Screening Hospital Algorithm	39
Instructions for Entering Hearing Demographic Information	40
American Academy of Pediatrics Early Hearing Detection & Intervention (EHDl) Guidelines for Pediatric Providers	41
Re-screen Newborn Hearing Results Form	42

Diagnostic Hearing Evaluation Form	43
Children's Rehabilitation Services Newborn Hearing Assessment Clinics	44
Center for Disease Control and Prevention's EHDl Program Update	45
Alabama Newborn Hearing Provider Directory	46
Newborn Hearing Screening Checklist	47-49
Newborn Hearing Screening Frequently Asked Questions	50

SECTION 4 - PULSE OXIMETRY SCREENING

Critical Congenital Heart Disease (CCHD)	52
Pulse Oximetry Screening Equipment	53
Pulse Oximetry Training	54-61
Knowledge Assessment	58-59
Competency Checklist	60
Training Log	61
Pulse Oximetry Screening	62
Pulse Oximetry Screening Algorithm	63
Pulse Oximetry Reporting Form	64

SECTION 5 - RESOURCES

Public Health Districts	66
Alabama County Health Departments	67
Alabama Early Intervention System (AEIS)	68-69
Child Find Referral Form	70
Children's Rehabilitation Service (CRS)	72-73
Alabama Community-Based Sickle Cell Organizations	74

APPENDIX

Alabama Newborn Screening Timeline	76
Newborn Screening Disorder Descriptions	77-80
Alabama Newborn Screening Confirmed Disorders	81
Alabama Newborn Screening Public Health Law	82
Alabama State Board of Health Administrative Code	83-88
American College of Medical Genetics ACT Sheets	89-107

SECTION 1 – PROGRAM OVERVIEW

Alabama Newborn Screening Program	6
HIPAA	7
Alabama Newborn Screening Panel of Disorders	8
Medical Provider Responsibilities	9
Alabama Newborn Screening Medical Consultants	10
Secure Remote Viewer Instructions.....	11
Secure Remote Viewer Registration Form	12
Newborn Screening Education Material.....	13
Newborn Screening Refusal Form.....	14
Alabama Birthing Hospitals	15
Why Use An Accredited Cystic Fibrosis Center.....	16

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 (NBS) 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 many of the complications associated with genetic and endocrine disorders. Early diagnosis can reduce morbidity, premature death, and developmental disabilities, including intellectual impairment. The Alabama NBS panel includes 31 of 35 disorders recommended by the U.S. Department of Health and Human Services Secretary's Advisory Committee on Heritable Disorders in Newborns and Children. Each year, the Alabama NBS Program identifies approximately 150-200 infants with a metabolic, endocrine, hematological, or other congenital disorders that may not be apparent at birth.

The Alabama Newborn Early Hearing Detection and Intervention (EHDI) 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 with hearing loss are enrolled in early intervention services by six months of age.

The Bureau of Clinical Laboratories (BCL) NBS Laboratory performs blood analysis that aids in the diagnosis of 29 primary genetic disorders. In addition, screening is performed for over 15 secondary disorders, bringing the total to more than 45 disorders. All newborns identified with an abnormal result 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 to coordinate prompt diagnostic testing and develop an appropriate treatment plan, when necessary. Treatment may include medications, dietary restrictions and/or supplements, and surgical intervention.

HIPAA

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 screening 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 screening 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 r. 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 1, 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.

In conclusion, state law gives the State Board of Health the authority to designate newborn screenings and the authority to promulgate “such rules and regulations as it considers necessary to provide for the care and treatment of those newborn infants.” Ala. Code §22-20-3(b). Pursuant to this authority, the Board of Health has adopted the above-described regulations that required the reporting of all newborn screenings. Because HIPAA does not pre-empt laws for the conduct of public health surveillance, investigation, or intervention and HIPAA allows disclosures for public health activities, you may continue to release newborn screening information without patient authorization to Public Health for the conduct of public health activities. Furthermore, you may rely on Public Health’s judgment as to the minimum amount of information necessary in the disclosure request.

If you have any concerns or questions regarding these matters, please do not hesitate to contact me at 334-206-5209 or pamela.kendrick@adph.state.al.us.

Sincerely,
Pam Kendrick, Privacy Officer

ALABAMA NBS PANEL OF DISORDERS

There are thirty-one primary disorders which are currently included in the Alabama Newborn Screening Panel and over forty-five total disorders including the secondary conditions. Please see the appendix for a brief description and timeline of each primary disorder.

1. 3-Hydroxy-3-methylglutaric aciduria (HMG)
2. 3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC)
3. Argininosuccinic aciduria (ASA)
4. β -Ketothiolase deficiency (BKT)
5. Biotinidase deficiency (BIOT)
6. Carnitine uptake/ transport defect (CUD)
7. Citrullinemia type I (CIT)
8. Classic galactosemia (GALT)
9. Classic phenylketonuria (PKU)
10. Congenital adrenal hyperplasia (CAH)
11. Critical congenital heart disease (CCHD)
12. Cystic fibrosis (CF)
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 hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
21. Maple syrup urine disease (MSUD)
22. Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
23. Methylmalonic acidemia, cobalamin disorders (Cbl A, B)
24. Methylmalonic acidemia, methylmalonyl-CoA mutase (MUT)
25. Holocarboxylase synthase deficiency (MCD)
26. Primary congenital hypothyroidism (CH)
27. Propionic acidemia (PROP)
28. Severe combined immunodeficiencies (SCID)
29. Trifunctional protein deficiency (TFP)
30. Tyrosinemia type I (TYR I)
31. Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

MEDICAL PROVIDER RESPONSIBILITIES



Medical providers are notified of newborn screening results by mail as long as they are identified on the specimen collection form.



Medical providers are notified by immediate phone call for potential positives and/or abnormal results that are outside of set cutoff values.



Providers are encouraged to use Alabama's Secure Remote Viewer (SRV) to access newborn screening results.

Ensure that all newborn patients in their care have received complete and valid newborn screening results and that any invalid or screen positive results have been appropriately addressed.

Contact families about out-of-range or invalid screening results in a knowledgeable and sensitive fashion by educating themselves on the medical aspects of conditions included in the screening panel.

Facilitate repeat or confirmatory testing and appropriate subspecialty care and report the results of confirmatory tests and diagnosis to the Alabama Newborn Screening Program.

Collect a repeat newborn screen as soon as possible if the first test is unsatisfactory, and collect a routine repeat screen at 2-6 weeks of age on all infants since TSH elevations could be delayed.

Ensure that the recommended hearing screening method is used for the rescreening of all infants who fail their initial hearing screen.

Obtain a signed statement for parent refusal of newborn screenings, when applicable.

Ensure that medical provider contact information stays current with the state lab so that collection forms and test reports can be provided to the correct provider in a timely manner.

It is recommended that pediatric providers offer and explain newborn screening to families of children under their care. Pediatric providers may face professional liability for failing to adequately inform parents of each newborn screening test (Mallory vs. Meier, et al.). Newborn hearing results are reported electronically by birthing hospitals and may not always link to a blood spot record and appear on the lab report. Please be sure to verify newborn hearing screening is completed (see section 3).

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

ALABAMA NBS MEDICAL CONSULTANTS

ENDOCRINE - CH/CAH

1. USA Medical Center, Endocrinology
Samar Bhowmick, M.D. (251) 405-5147
Anne Marie Kaulfers, M.D., Christina Hair, R.N. (251) 410-5437
2. Children's of Alabama, Endocrinology
Gail Mick, M.D. (205) 638-9107
Leslie Pitts, CRNP (205) 638-6442

HEMOGLOBINOPATHIES - SICKLE CELL DISEASE, TRAIT CONDITIONS AND OTHER HEMOGLOBINOPATHIES

1. USA Sickle Cell Center
Felicia Wilson, M.D. (251) 405-5147 (#3)
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 (256) 265-5833

CYSTIC FIBROSIS

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

1. UAB Genetics
Maria Descartes, M.D. (205) 934-4983
Alicia Roberts, R.D. (205) 996-6983

CRITICAL CONGENITAL HEART DISEASE (CCHD)

1. UAB Pediatric Cardiology (205) 934-3460 (direct), (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. Diagnostic & Medical Clinic (Mobile) (251) 435-1200

SEVERE COMBINED IMMUNODEFICIENCY (SCID)

1. Children's of Alabama
T. Prescott Atkinson, M.D., Allergy, Asthma, Immunology (205) 638-9586
2. UAB – Lowder Pediatric Blood and Marrow Transplantation Program
Infants will 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 Bureau of Clinical Laboratories (BCL). 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 Alabama Bureau of Clinical Laboratories 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 three options for the account's username. 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-ups 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 numbers on filter form followed by last 2 digits of birth year).
7. Once the search criteria have been entered select the Perform Search button at the bottom of the page.
8. If the minimum criteria have not been entered "Invalid Search Criteria" will be displayed.
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 Bureau of Clinical Laboratories.

PLEASE PRINT

Name of Physician (first and last name) _____

Name of Facility _____

Mailing Address _____

Area Code/Telephone Number _____

E-Mail Address _____

THIS IS REQUIRED: The registrant will receive an invitation via email.

Username (list three options) _____

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.

NEWBORN SCREENING EDUCATION MATERIAL

Newborn screening material may be ordered directly online at the following site <https://www.alabamapublichealth.gov/Extranet/Forms/Form.asp?ss=s&formID=5561> 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 volume of materials 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 Number _____

Contact Person _____

Education Material Number: ADPH-FHS- _____

Quantity Requested _____

FHS-533 comes in packets of 50 and FHS-537/538 comes in packets of 100, limit of 10 packets

FHS-533



Description: Booklet that includes bloodspot, hearing, and pulse ox screening information. Spanish version also available.

FHS-537 (5x8 card)



Description: Single 5x8 card for expecting parents. Includes four statements parents need to know about newborn screening. English on one side and Spanish on other side.

FHS-538



Description: Pamphlet with hearing information for parents. Spanish version also available.

NEWBORN SCREENING REFUSAL FORM

The American Academy of Pediatrics and the Alabama Department of Public Health strongly recommend Newborn Screening for all infants. Parents have a right to refuse newborn screening. Parents should be provided education regarding the risks of not screening their baby and should sign a refusal form for informed consent if refusing any part of the newborn screening.

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 choose not to have my child receive the newborn bloodspot screening from the Alabama Department of Public Health for life threatening diseases screened for by the Newborn Screening Program.
- ☐ I choose not to have my child screened for hearing loss.
- ☐ I choose not to have my child screened for critical congenital heart disease.

I have been provided information about newborn screening in my state and the importance of early identification of the disorders. I 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.
- **If my child does not participate in newborn screening, the consequences of a late diagnosis may include delayed development, intellectual disability, 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 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	Births in 2016
Huntsville Hospital	4,383
UAB Hospital	4,297
Brookwood Medical Center	4,006
Baptist Medical Center East	3,634
St. Vincent's Birmingham	3,347
USA Children's & Women's Hospital	2,670
East Alabama Medical Center	2,077
DCH Regional Medical Center	1,935
Northeast AL Regional Medical Center	1,890
Providence Hospital	1,585
Northport Medical Center	1,540
Southeast AL Medical Center	1,506
Flowers Hospital	1,317
Thomas Hospital	1,258
North Alabama Medical Center*	1,228
Jackson Hospital	1,210
Mobile Infirmary Medical Center	1,115
Gadsden Regional Medical Center	1,043
Shelby Baptist Medical Center	1,008
Medical Center Enterprise	969
Decatur Morgan Hospital-Parkway	957
Grandview Medical Center	957
Madison Hospital	885

Birthing Hospital	Births in 2016
Crestwood Medical Center	873
Springhill Medical Center	873
Helen Keller Hospital	804
Walker Baptist Medical Center	773
Baptist Medical Center South	747
DeKalb Regional Medical Center	699
Marshall Medical Center South	626
South Baldwin Regional Medical Center	610
Vaughan Regional Medical Center	606
Cullman Regional Medical Center	600
Athens-Limestone Hospital	523
Coosa Valley Medical Center	472
Marshall Medical Center North	437
Russell Medical Center	415
UAB Medical West	396
Andalusia Regional Hospital	389
Highlands Medical Center	371
Princeton Baptist Medical Center	319
D.W. McMillan Memorial Hospital	254
North Baldwin Infirmary	213
Grove Hill Memorial Hospital	210
Monroe County Hospital	190
Bibb Medical Center	60

*Formerly Eliza Coffee Memorial Hospital

Per the Center for Health Statistics, Births by Hospital of Occurrence, Alabama 2016 Report

WHY USE AN ACCREDITED CYSTIC FIBROSIS CENTER?

Accredited Cystic Fibrosis (CF) Care Centers are required to meet nationally accepted standards that have been developed by the Cystic Fibrosis Foundation (CFF) and the Clinical and Laboratory Standards Institute (CLSI).

National standards for diagnostic sweat testing are imperative to ensure the results are consistently accurate and reliable. CFF accredited centers offer a multidisciplinary approach to the management of cystic fibrosis and include the following clinic personnel:

- Physicians
- Registered nurses
- Respiratory therapists
- Dietitians/nutritionists
- Social workers
- Geneticist or genetic counselors

Having these specialists available in a single location increases the convenience of treatment for CF. Families are able to make a single appointment at the CF Center, rather than separate appointments for each specialist.

Babies referred to an accredited CF Center in Alabama also get referred to Children's Rehabilitation Service (CRS) which offers medical, financial, and support services to families and children facing a variety of special health care needs. These clinics provide state-of-the-art care for infants and children with CF in Alabama. Every county in Alabama is served through a network of 14 community based CRS offices.

For your convenience, contact information for the CFF accredited center in Alabama is included below. Transportation assistance is available to families who qualify.

University of Alabama at Birmingham/Children's of Alabama Cystic Fibrosis Center

Dr. Hector Gutierrez, Pediatric Pulmonologist

1600 7th Avenue South, Lowder 620

Birmingham, AL 35233 • (205) 638-5494

SECTION 2 - SPECIMEN COLLECTION

One Drop, One Circle, One Time 18

Newborn Screening Blood Collection Guidelines19-27

Sick Infant Blood Collection Guidelines26

Hemoglobinopathy Repeats28

Newborn Screening Collection Tips29

Whatman® Neonatal Screening Reference Form 30-31

Whatman® Simple Spot Check Reference Form.....32

Alabama Newborn Screening Kit Reorder Form.....33

Newborn Screening Provider Update Form.....34



**ONE DROP
ONE CIRCLE
ONE TIME**

NEWBORN SCREENING COLLECTION GUIDELINES

ALABAMA DEPARTMENT OF PUBLIC HEALTH BUREAU OF CLINICAL LABORATORIES

Newborn Screening Collection Guidelines

Revised 4/1/2019



ALABAMA
PUBLIC
HEALTH

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
Phone: 334-260-3476 • FAX: 334-260-3439

Sharon P. Massingale, PhD, HCLD/CC(ABB), Public Health Laboratory Director
Aretha M. Williams, PhD, Laboratory Assistant Director
Danita M. Rollin, BS, MT (ASCP), Newborn Screening Laboratory Division Manager

Contacts: Deannie Morris, Lynn Green and Derek Kennedy

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, Severe Combined Immunodeficiency, 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, Severe Combined Immunodeficiency, 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. Refer to the Alabama Newborn Screening Sick Infant Blood Collection Guidelines on page 26.
Transitioning Infants	Infants admitted to NICU for short term observation but who 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.
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.

NEWBORN SCREENING COLLECTION GUIDELINES

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.

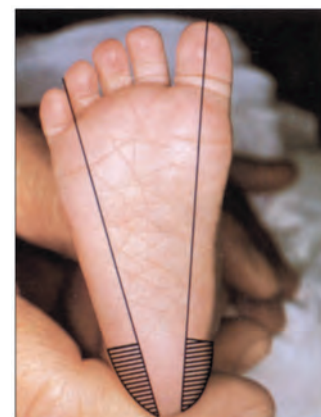
COLLECTION OF FILTER PAPER BLOODSPOT SPECIMEN

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 2.0 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, a towel, or a 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 <2.0 mm. **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:

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

TIMING & TRANSPORT (i)

1. Specimens should be shipped or transported by mail, major courier services*, or other express delivery services to the public health laboratory as soon as they are dry (minimum of three hours) and no later than 24 hours after collection. If mailed to the lab, please send to the following address:

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

*Daily courier transport is recommended whenever possible to control environmental conditions and minimize delays in shipment.

2. Appropriate documentation for all stages in specimen transit should be tracked and maintained, from collection to delivery.
3. Dried blood spots (DBS) are nonregulated and an exempt human specimen, posing no occupational exposure to blood or other potentially infectious material. Standard precautions should be followed in preparing these specimens for shipment.
4. It is **NOT** recommended that DBS specimens be packaged in airtight, leak-proof sealed containers (e.g., plastic or foil bags) because the lack of air exchange causes heat buildup and moisture accumulation that is detrimental to the stability of the DBS specimen.
5. Do **NOT** place in outside mailboxes or drop boxes because fluctuating temperature and humidity may damage specimens.
6. The inclusion of desiccant packs may aid in preventing moisture accumulation.
7. The use of preaddressed envelopes for mailing may help decrease the transport time, and thus decrease time from collection to diagnosis in affected newborns.
8. To mail DBS specimens, please use the basic triple-packaging system:
 - Primary container – filter paper that contains absorbed and dried blood
 - Secondary container – fold over flap envelope to secure the contents
 - Third container – outer envelope of sturdy, high quality paper

CLSI. *Blood Collection on Filter Paper for Newborn Screening Programs; Approved Standard—Sixth Edition*. CLSI document NBS01-A6. Wayne, PA: Clinical and Laboratory Standards Institute; 2013.

NEWBORN SCREENING COLLECTION GUIDELINES

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 expire 3-2020.

FORM A

ALABAMA NEWBORN SCREENING PROGRAM

Alabama Department of Public Health
Bureau of Clinical Laboratories
6140 AUM Drive, PO Box 244018
Montgomery, AL 36124-4018

STATE LAB (334) 260-3400

Infant's Last Name 1		Infant's First Name 1		Medical Record # 2	Infant's Medicaid # 3
Date of Birth 4	Time of Birth (Military) 5	Birth Weight 6 (gms) (Current Wt. / mth.)	Multiple Birth Order 7	Weeks Gestation 8	
Date of Collection 9	Time of Collection (Military) 10	<input type="checkbox"/> Male 11 <input type="checkbox"/> Female 12	Last Transfusion 13 MM DD YY TIME		
<input type="checkbox"/> Home Birth 14	Infant's Age 15	<input type="checkbox"/> White <input type="checkbox"/> Black <input type="checkbox"/> Other <input type="checkbox"/> Asian 16 <input type="checkbox"/> Hispanic	<input type="checkbox"/> First Test <input type="checkbox"/> Retest - Prior 17	<input type="checkbox"/> Routine Second Test <input type="checkbox"/> Retest - Prior Abnormal (Requested by State)	
Mother's Last Name 18		Mother's First Name 18		Mother's Social Security Number 18	
Mailing Address 18		Mother's Phone Number 18		Mother's Medicaid Number 18	
City 18	County 18	State 18	Zip 18		
Ordering Physician (Last) 19 (First) (MI)		PULSE OXIMETRY SCREENING: 22 at screening (hrs) <input type="checkbox"/> Pass <input type="checkbox"/> Fail <input type="checkbox"/> Not Performed <input type="checkbox"/> Refused <input type="checkbox"/> Expired <input type="checkbox"/> NICU <input type="checkbox"/> On O2			
NPI # 20		Notes			
Referral Physician 21					
SUBMITTER ADDRESS 23		24 -Laboratory use only- Do not write on or affix labels in this area			
AL Zip					

FORMS MUST BE FILLED OUT COMPLETELY IN BLUE OR BLACK INK - PRINT LEGIBLY

SN 550004

INSURANCE INFORMATION - Complete Form (Instructions on Back) - DO NOT REMOVE

SPECIMEN SHOULD BE COMPLETELY DRY BEFORE COVERING



NO BLOOD ON FLAP
FLAP MUST REMAIN INTACT

FORM B

ALABAMA NEWBORN SCREENING PROGRAM

Alabama Department of Public Health
Bureau of Clinical Laboratories
6140 AUM Drive, PO Box 244018
Montgomery, AL 36124-4018

STATE LAB (334) 260-3400

Infant's Last Name 1		Infant's First Name 1		Medical Record # 2	Infant's Medicaid # 3
Date of Birth 4	Time of Birth (Military) 5	Birth Weight 6 (gms) (Current Wt. / mth.)	Multiple Birth Order 7	Weeks Gestation 8	
Date of Collection 9	Time of Collection (Military) 10	<input type="checkbox"/> Male 11 <input type="checkbox"/> Female 12 TPN	Last Transfusion 13 MM DD YY TIME		
<input type="checkbox"/> Home Birth 14	Infant's Age 15	<input type="checkbox"/> White <input type="checkbox"/> Black <input type="checkbox"/> Other <input type="checkbox"/> Asian 16 <input type="checkbox"/> Hispanic	<input type="checkbox"/> First Test <input type="checkbox"/> Retest - Prior 17	<input type="checkbox"/> Routine Second Test <input type="checkbox"/> Retest - Prior Abnormal (Requested by State)	
Mother's Last Name 18		Mother's First Name 18		Mother's Social Security Number 18	
Mailing Address 18		Mother's Phone Number 18		Mother's Medicaid Number 18	
City 18	County 18	State 18	Zip 18		
Ordering Physician (Last) 19 (First) (MI)		Notes			
NPI # 20					
Referral Physician 21					
SUBMITTER ADDRESS 23		24 -Laboratory use only- Do not write on or affix labels in this area			
AL Zip					

FORMS MUST BE FILLED OUT COMPLETELY IN BLUE OR BLACK INK - PRINT LEGIBLY

SN 775002

INSURANCE INFORMATION - Complete Form (Instructions on Back) - DO NOT REMOVE

SPECIMEN SHOULD BE COMPLETELY DRY BEFORE COVERING



NO BLOOD ON FLAP
FLAP MUST REMAIN INTACT

SEE BACK OF FORM FOR SPECIMEN COLLECTION INSTRUCTIONS 2020-03-31

SEE BACK OF FORM FOR SPECIMEN COLLECTION INSTRUCTIONS 2020-03-31

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 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. <i>Mother's social security number should be entered accurately.</i> 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. <i>This information is required to be provided and complete.</i>
20	NPI field – enter the National Provider Identification 9 digit number for the ordering physician . <i>This information is required to be provided and complete.</i>
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	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
23	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. <i>This information is required to be complete and accurate.</i>
24	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.
25	INSURANCE FORM – Insurance information MUST be entered completely and accurately. This sheet should not be removed from the NBS form.

EXAMPLES

SEE BACK OF FORM FOR SPECIMEN COLLECTION INSTRUCTIONS 2020-03-31

ALABAMA NEWBORN SCREENING PROGRAM

Alabama Department of Public Health
Bureau of Clinical Laboratories
8140 AUM Drive, PO Box 244018
Montgomery, AL 36124-4018
STATE LAB (334) 260-3400

Infant's Last Name DOE		Infant's First Name BABY		Medical Record # 134521	Infant's Medicaid # 5000001234567
Date of Birth 01 05 19	Time of Birth (Military) 0 3 2 0	Birth Weight 2250 (gms) (Current WT. if > 1 mth.)		Multiple Birth Order TWIN A	Weeks Gestation 38wks
Date of Collection 01 06 19	Time of Collection (Military) 0 3 0 0	<input type="checkbox"/> Male <input checked="" type="checkbox"/> Female <input type="checkbox"/> TPN <input type="checkbox"/> White <input type="checkbox"/> Black <input type="checkbox"/> Other <input checked="" type="checkbox"/> Asian <input type="checkbox"/> Hispanic		Last Transfusion 01/05/19 MM DD YY TIME @1400	
<input type="checkbox"/> Home Birth Infant's Age 24 HRS		<input type="checkbox"/> First Test <input type="checkbox"/> Routine Second Test <input type="checkbox"/> Retest - Prior Unsat <input type="checkbox"/> Retest - Prior Abnormal (Requested by State)			
Mother's Last Name DOE		Mother's First Name JANE		Mother's Social Security Number 4 9 9 9 9 9 9 9 9	
Mailing Address 123 NEW BABY DR		Mother's Phone Number 3 3 3 2 9 5 3 3 3		Mother's Medicaid Number 5 0 0 0 0 9 9 9 9 9 9 9	
City HUNTSVILLE		County MADISON		State AL	Zip 35801
Ordering Physician (Last) HOWSER, DOOGIE		(First) 		(MI) 	
NPI # 1 4 1 1 3 4 4 1 1 1		Referral Physician PEDIATRIC PEDS		PULSE OXIMETRY SCREENING: Age at screening 36 (hrs) <input checked="" type="checkbox"/> Pass <input type="checkbox"/> Fail <input type="checkbox"/> Not Performed <input type="checkbox"/> Refused <input type="checkbox"/> Expired <input type="checkbox"/> NICU <input type="checkbox"/> On O2 Notes	
SUBMITTER ADDRESS EASTMAN HOSPITAL		456 HOSPITAL DR		HUNTSVILLE AL 35801	

FORMS MUST BE FILLED OUT COMPLETELY IN BLUE OR BLACK INK - PRINT LEGIBLY

SN 693952

INSURANCE INFORMATION - Complete Form (Instructions on Back) - DO NOT REMOVE

SPECIMEN
SHOULD BE
COMPLETELY DRY
BEFORE COVERING



NO BLOOD
ON FLAP

FLAP MUST
REMAIN INTACT

DO NOT USE
-Laboratory use only
Do not write on or affix labels in this area
THIS AREA

SEE BACK OF FORM FOR SPECIMEN COLLECTION INSTRUCTIONS 2020-03-31

ALABAMA NEWBORN SCREENING PROGRAM

Alabama Department of Public Health
Bureau of Clinical Laboratories
8140 AUM Drive, PO Box 244018
Montgomery, AL 36124-4018
STATE LAB (334) 260-3400

Infant's Last Name DOE		Infant's First Name BAMBI		Medical Record # 345-2	Infant's Medicaid # 5000001234567
Date of Birth 01 05 19	Time of Birth (Military) 0 3 2 0	Birth Weight 2250 (gms) (Current WT. if > 1 mth.)		Multiple Birth Order TWIN A	Weeks Gestation 38wks
Date of Collection 01 20 19	Time of Collection (Military) 1 3 1 5	<input type="checkbox"/> Male <input checked="" type="checkbox"/> Female <input type="checkbox"/> TPN <input type="checkbox"/> White <input type="checkbox"/> Black <input type="checkbox"/> Other <input checked="" type="checkbox"/> Asian <input type="checkbox"/> Hispanic		Last Transfusion 01/05/19 MM DD YY TIME @1400	
<input type="checkbox"/> Home Birth Infant's Age 2wks		<input type="checkbox"/> First Test <input checked="" type="checkbox"/> Routine Second Test <input type="checkbox"/> Retest - Prior Unsat <input type="checkbox"/> Retest - Prior Abnormal (Requested by State)			
Mother's Last Name DOE		Mother's First Name JANE		Mother's Social Security Number 4 9 9 9 9 9 9 9 9	
Mailing Address 123 NEW BABY DR		Mother's Phone Number 3 3 3 2 9 5 3 3 3		Mother's Medicaid Number 5 0 0 0 0 9 9 9 9 9 9 9	
City HUNTSVILLE		County MADISON		State AL	Zip 35801
Ordering Physician (Last) MCSTUFFINS, DOC		(First) 		(MI) 	
NPI # 1 0 1 0 1 0 1 0 1 0		Referral Physician MCSTUFFINS, DOC		PULSE OXIMETRY SCREENING: Age at screening 36 (hrs) <input type="checkbox"/> Pass <input type="checkbox"/> Fail <input type="checkbox"/> Not Performed <input type="checkbox"/> Refused <input type="checkbox"/> Expired <input type="checkbox"/> NICU <input type="checkbox"/> On O2 Notes	
SUBMITTER ADDRESS PEDIATRIC PEDS		1001 PEDS DR		HUNTSVILLE AL 35801	

FORMS MUST BE FILLED OUT COMPLETELY IN BLUE OR BLACK INK - PRINT LEGIBLY

SN 944202

INSURANCE INFORMATION - Complete Form (Instructions on Back) - DO NOT REMOVE

SPECIMEN
SHOULD BE
COMPLETELY DRY
BEFORE COVERING



NO BLOOD
ON FLAP

FLAP MUST
REMAIN INTACT

DO NOT USE
-Laboratory use only
Do not write on or affix labels in this area
THIS AREA

SICK INFANT BLOOD COLLECTION GUIDELINES

Sick Infant and Well Baby Newborn Screening Blood Collection Algorithm, February 22, 2016

The following newborn screening algorithm 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.

BIRTH OF PRETERM, LBW OR SICK NEWBORN

Serial screening, with the collection of three specimens, is proposed as the most expedient and efficient paradigm for this population (CLSI Preterm, LBW, and Sick Newborns, page 19)

Transfer/Arrival NICU NBS Specimen

Collect the **"arrival NICU"** NBS specimen on admission to the NICU (if not already collected) **regardless of age*** before any other treatments are begun (transfusions, TPN or antibiotics). If transferred, **the transfer hospital** should collect a specimen on Form A before transported unless infant is unstable. **The receiving hospital**, on admission, should collect a specimen on a second test form (Form B) and mark the "First Test" box.

***For most preterm and LBW newborns, admission to the NICU occurs immediately after birth, usually 1 to 2 hours of age, or up to 24 hours of age. If an infant is 24 hours of age or older on admission to the NICU, repeat screening should be done according to local program recommendations for normal infants unless there were abnormalities on the initial specimen (CLSI, page 20).**

Acute NICU NBS Specimen

Collect the **"acute NICU"** NBS specimen at **48-72 hours of life** on infants initially tested at **<24 hours of age at first screen**.

- **If receiving blood** - wait and collect 72 hours after the last transfusion.
- **If on TPN** - collect acute screen plus an additional screen when TPN is discontinued.

Final NICU NBS Specimen

Collect the **"final routine NICU"** NBS specimen at **28 days of age** or at discharge, whichever comes first, for any infant in the NICU > 2 weeks of age. All NICU infants discharged before 2 weeks of age should have the recommended routine NBS specimen collected by their pediatrician at 2-6 weeks of age.

BIRTH OF FULL-TERM OR WELL NEWBORN

Initial NBS Specimen

Collect an initial NBS specimen at **24-48 hours of age** (mail within 24 hours).

- Collect the first sample on a First Test Form (A Form) and any subsequent samples on a second test form (B Form).
- 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 second test before one week of age.

Routine Repeat NBS Specimen

Collect a recommended routine second NBS specimen at **2-6 weeks of age**.

- This specimen is not routinely tested for Hemoglobinopathies or Severe Combined Immunodeficiencies.
- Collect on a Second Test Form (B Form)

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, being alert to the effects that treatments and the infant's condition may have on the screening test results.

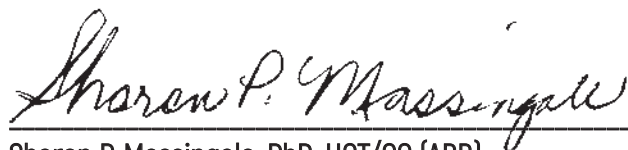
If a well infant has had 2 newborn screens collected or a sick infant has had 4 screens collected (check Secure Remote Viewer to determine), please contact the NBS Lab at 334-260-3476, or the NBS Follow-up Program at 334-206-5556 for follow-up issues.

NEWBORN SCREENING COLLECTION GUIDELINES

Alabama Department of Public Health Bureau of Clinical Laboratories Newborn Screening Blood Collection Guidelines

These guidelines have been provided for newborn screening providers in order to inform and instruct on the proper techniques of collecting a high-quality specimen, for handling it after it has been collected, and for transporting it to the testing facility. These guidelines 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, Bureau of Clinical Laboratories.

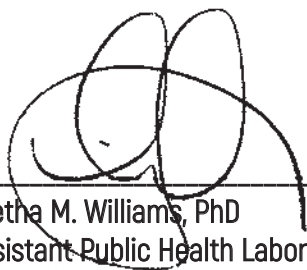
For further guidance please refer to the *CLSI® Blood Collection on Filter Paper for Newborn Screening Programs; Approved Standard*, which addresses the issues associated with specimen collection, the filter paper collection device, the application of blood to the filter paper, and uniform techniques for collecting the best possible specimen for use in newborn screening programs.



Sharon P. Massingale, PhD, HCT/CC (ABB)
Public Health Laboratory Director
Bureau of Clinical Laboratories

4-19-19

Date



Aretha M. Williams, PhD
Assistant Public Health Laboratory Director
Bureau of Clinical Laboratories

4-19-19

Date



Danita M. Rollin, BS, MT (ASCP)
Newborn Screenings Laboratory Division Manager
Bureau of Clinical Laboratories

4-12-19

Date

NBS 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 is 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 filter 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 **well-trained** individuals to collect newborn screening blood in order to reduce unsatisfactory specimens.
4. Track the performance of these collectors and re-train or substitute as necessary if unsatisfactory or invalid results occur.
5. **Perform a quality control inspection** 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.**
 - 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.

Neonatal Screening

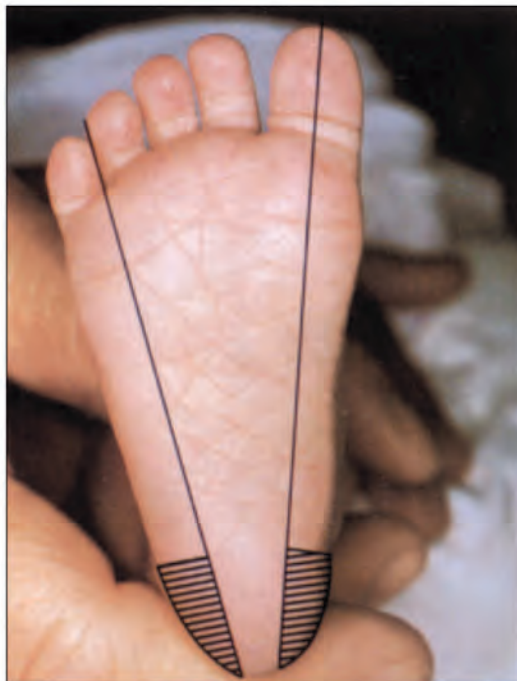
Blood Specimen Collection and Handling Procedure



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



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



- 3** Hatched area (//) indicates safe areas for puncture site.



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



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



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



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



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

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



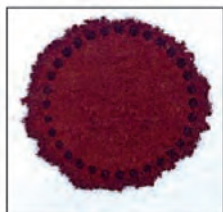
10 Mail completed form to testing laboratory within 24 hours of collection.

Information provided by The New York State Department of Health.

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

51684 3/08

SIMPLE SPOT CHECK



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. 	<ul style="list-style-type: none"> 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. 	<ul style="list-style-type: none"> Applying blood with a capillary tube or other device.
3. Specimen not dry before mailing. 	<ul style="list-style-type: none"> Mailing specimen before drying for a minimum of four hours.
4. Specimen appears supersaturated. 	<ul style="list-style-type: none"> 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. 	<ul style="list-style-type: none"> 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. 	<ul style="list-style-type: none"> 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. 	<ul style="list-style-type: none"> Touching the same circle on filter paper to blood drop several times. Filling circle on both sides of filter paper.
8. No blood. 	<ul style="list-style-type: none"> Failure to obtain blood specimen.



BUREAU OF CLINICAL LABORATORIES

SHARON P. MASSINGALE, PhD., 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
SCOTT HARRIS, M.D., M.P.H. • STATE HEALTH OFFICER

BUREAU OF CLINICAL LABORATORIES
SHARON P. MASSINGALE, PhD., 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 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# _____

Mail: P.O. Box 244018
Montgomery, AL 36124-4018

Phone: 334/260-3400
Fax: 334/274-9800

Shipping: 8140 AUM Drive
Montgomery, AL 36117-7001

SECTION 3 - NEWBORN HEARING SCREENING

Newborn Hearing Screening Program Overview	36
Inpatient Newborn Hearing Screening Protocol.....	37
Joint Committee on Infant Hearing	38
Newborn Hearing Screening Hospital Algorithm	39
Instructions for Entering Hearing Demographic Information	40
American Academy of Pediatrics Early Hearing Detection & Intervention (EHDI) Guidelines for Pediatric Providers	41
Re-screen Newborn Hearing Results Form	42
Diagnostic Hearing Evaluation Form	43
Children's Rehabilitation Services Newborn Hearing Assessment Clinics ...	44
CDC's EHDI Program Update	45
Alabama Newborn Hearing Provider Directory	46
Newborn Hearing Screening Checklist	47-49
Newborn Hearing Screening Frequently Asked Questions	50



NEWBORN HEARING SCREENING PROGRAM OVERVIEW

The Alabama Universal Newborn Hearing Screening Program, also known as *“Alabama’s Listening!”* or the *Early Hearing Detection and Intervention (EHDI) Program*, is administered through the Alabama Department of Public Health and was established in February 2001 to address the hearing health care needs of Alabama’s babies. The health care needs of infants with hearing loss include timely screening, diagnosis, and referral to Early Intervention (EI) services. The program follows the Joint Committee on Infant Hearing 1-3-6 guidelines: screening before **one** month of age, diagnostic evaluation by an audiology professional before **three** months of age, and referral to EI services before **six** months of age.

The Alabama EHDI Program collaborates with many partners to include state birthing hospitals, pediatric health care providers, Alabama Department of Rehabilitation Services, the National Center for Hearing Assessment and Management (NCHAM), Early Head Start Programs, the Centers for Disease Control and Prevention (CDC), and the Health Resources and Services Administration (HRSA). The goal of the Alabama EHDI Program is to ensure all infants with hearing loss are identified as early as possible and provided with timely and appropriate audiological, educational, and medical intervention in order to improve a child’s speech and language development, as well as thinking, learning, and social skills.

INPATIENT NEWBORN HEARING SCREENING PROTOCOL

Each birthing hospital is responsible for creating and implementing policies and procedures that are in line with state and national recommendations. These policies and procedures should follow state law and include, but not be limited to the following: 1) screening procedure, 2) reporting procedure, 3) guidelines for training personnel, 4) performance maintenance, 5) quality improvement indicators, and 6) data management.

The Alabama State Law, Section 22-20-3, provides legal authority for institutions caring for infants 28 days or less of age to administer a reliable test for newborn screening to include the newborn hearing screening. The law allows for parents to refuse testing on the grounds that such tests conflict with their religious tenets and practices. A written refusal should be obtained if a parent objects to newborn screening (page 14). In addition, the Alabama State Board of Health Administrative Code, Chapter 420-10-1, *Care and Treatment of Infants Identified Through the Newborn Screening Program*, mandates reporting of any hearing tests performed on the newborns to the Alabama Department of Public Health and use of forms and guidelines as determined by the State Health Officer.

Inpatient Screening Protocol Recommendations:

- Identify staff responsible for screening, reporting, and training personnel.
- Document all job descriptions, qualifications, and roles, as well as orientation, minimum length of training, and competency validation.
- It is recommended that the discharge planner be responsible for notifying parents of the newborn's hearing results, and responsible for scheduling outpatient hearing screening as necessary.
- Identify the name, model, and type of testing equipment being used for screening purposes. Care, use, troubleshooting, maintenance and servicing of the testing equipment should be included.
- A copy of the policy and procedure manual for newborn hearing screening should be located in close proximity to the screening site.
- Birthing hospitals should also perform in-house quality assurance/improvement on a quarterly basis.
- Hospitals should use a general consent to perform hearing screening. It is advised that each facility consult with their legal representation to ensure that the consent is appropriate to cover this service.
- Identify the optimal testing environment as well as the desired condition or state of the newborn during testing.
- Identify risk indicators associated with hearing loss if known.

Resources:

- An Audiology Provider Directory is available at www.ehdipals.org
- Training may be provided by:
 - o State Newborn Hearing Screening Coordinator at (334) 206-2944
 - o Video training is also available on the website: www.alabamapublichealth.gov/newbornscreening/newborn-hearing-screening.html
 - o Interactive Web Based Newborn Hearing Screening Training Curriculum strongly recommended for all hospital staff performing the newborn hearing screening: www.infanthearing.org/nhstc/index.html

JOINT COMMITTEE ON INFANT HEARING

The Joint Committee on Infant Hearing (JCIH) 2007 Position Statement serves as the national standard for Early Hearing Detection and Intervention Programs. JCIH endorses early detection and intervention for infants with hearing loss to ensure opportunities to maximize linguistic competence and literacy development so that infants and children do not fall behind their peers in communication, cognition, reading, and social-emotional development. According to JCIH, such delays may result in lower educational and employment levels in adulthood.

Included is a link to the 2007 JCIH Position Statement along with an outline of important points: www.jcih.org/posstatemts.htm

- Separate protocols are recommended for NICU and well-baby nurseries.
- NICU babies greater than five days are to have Automated Brainstem Response (ABR) included as part of their screen so that neural hearing loss will not be missed.
- For infants who do not pass automated ABR testing in the NICU, referral should be made directly to an audiologist for rescreening.
- All infants who do not pass the initial hearing screening and the subsequent rescreening should have appropriate audiological and medical evaluations to confirm the presence of hearing loss no later than 3 months of age.
- Screening results should be conveyed immediately to families so they understand the outcome and importance of follow-up when indicated.
- A complete evaluation of both ears is recommended for each rescreening, even if only one ear did not pass the initial screen.
- For readmissions of infants in the first month of life, if there are conditions present which are associated with potential hearing loss (e.g. hyperbilirubinemia requiring exchange transfusion or culture + sepsis), a repeat hearing screen is recommended prior to discharge.
- Audiologists with skills and expertise in evaluating infants with hearing loss should provide diagnostic evaluation before three months of age.

NEWBORN HEARING SCREENING HOSPITAL ALGORITHM

Based on the Joint Committee on Infant Hearing (JCIH) Guidelines

Initial newborn hearing screening is performed
24-48 hours of age or before the baby leaves the hospital

Before you start the initial newborn hearing screening, is the baby's...

☐ Information entered exactly as entered on the blood spot form? (Refer to the instructions for entering demographic information into the hearing device)

☐ Testing method appropriate and all supplies gathered for testing both ears?

It is recommended to perform **only** two inpatient hearing screens, one initial and one rescreen if needed.

Otoacoustic Emissions (OAE)

- Measures hair cells of the outer ear
- Does not detect neural hearing loss
- Should only be used for well babies

Automated Auditory Brainstem Response (AABR)

- Measures inner ear and brain response to sound
- Detects neural hearing loss
- May be used for all infants, must be used for all NICU.

DID NOT PASS IN ONE OR BOTH EARS

Re-screen both ears with OAE and/or AABR, even if only one ear did not pass.

PASS BOTH EARS

No further testing required

DID NOT PASS IN ONE OR BOTH EARS

Re-screen both ears with AABR **only**, even if only one ear did not pass. A referral should be made **directly** to an audiologist for rescreening on infants who do not pass AABR.

DID NOT PASS RE-SCREEN IN ONE OR BOTH EARS

Schedule follow-up testing with a hearing professional within 2-3 weeks after discharge.

PASS WITH RISK FACTORS

Further testing recommended between 24-36 months of age.
NICU admission > 5 days is considered a risk factor.

Hearing Results should be sent electronically to the Alabama Newborn Screening Program each day. See the instructions for *Reporting Hearing Results Electronically*.

INSTRUCTIONS FOR ENTERING HEARING DEMOGRAPHIC INFORMATION INTO THE HEARING DEVICE & REPORTING HEARING RESULTS ELECTRONICALLY

INSTRUCTIONS 2020-03-31

ALABAMA NEWBORN SCREENING PROGRAM

Alabama Department of Public Health
Bureau of Clinical Laboratories
8140 AUM Drive, PO Box 244018
Montgomery, AL 36124-4018

Infant's Last Name		Infant's First Name		Medical Record #	
Date of Birth	Time of Birth (Military)	Birth Weight (gms) (Current WT. if > 1 mth.)		Multiple Birth Order	
Date of Collection	Time of Collection (Military)	<input type="checkbox"/> Male <input type="checkbox"/> Female <input type="checkbox"/> TPN		Last Transfusion MM DD YY	
<input type="checkbox"/> Home Birth	Infant's Age	<input type="checkbox"/> White <input type="checkbox"/> Black <input type="checkbox"/> Other <input type="checkbox"/> Asian <input type="checkbox"/> Hispanic		<input type="checkbox"/> First Test <input type="checkbox"/> Retest - Prior Unsat	
Mother's Last Name		Mother's First Name		Mother's Social Security Number	

Hearing demographic information must match the information highlighted in red on the blood spot form when entered into the hearing device.

Entering Hearing Demographic Information into the Hearing Device:

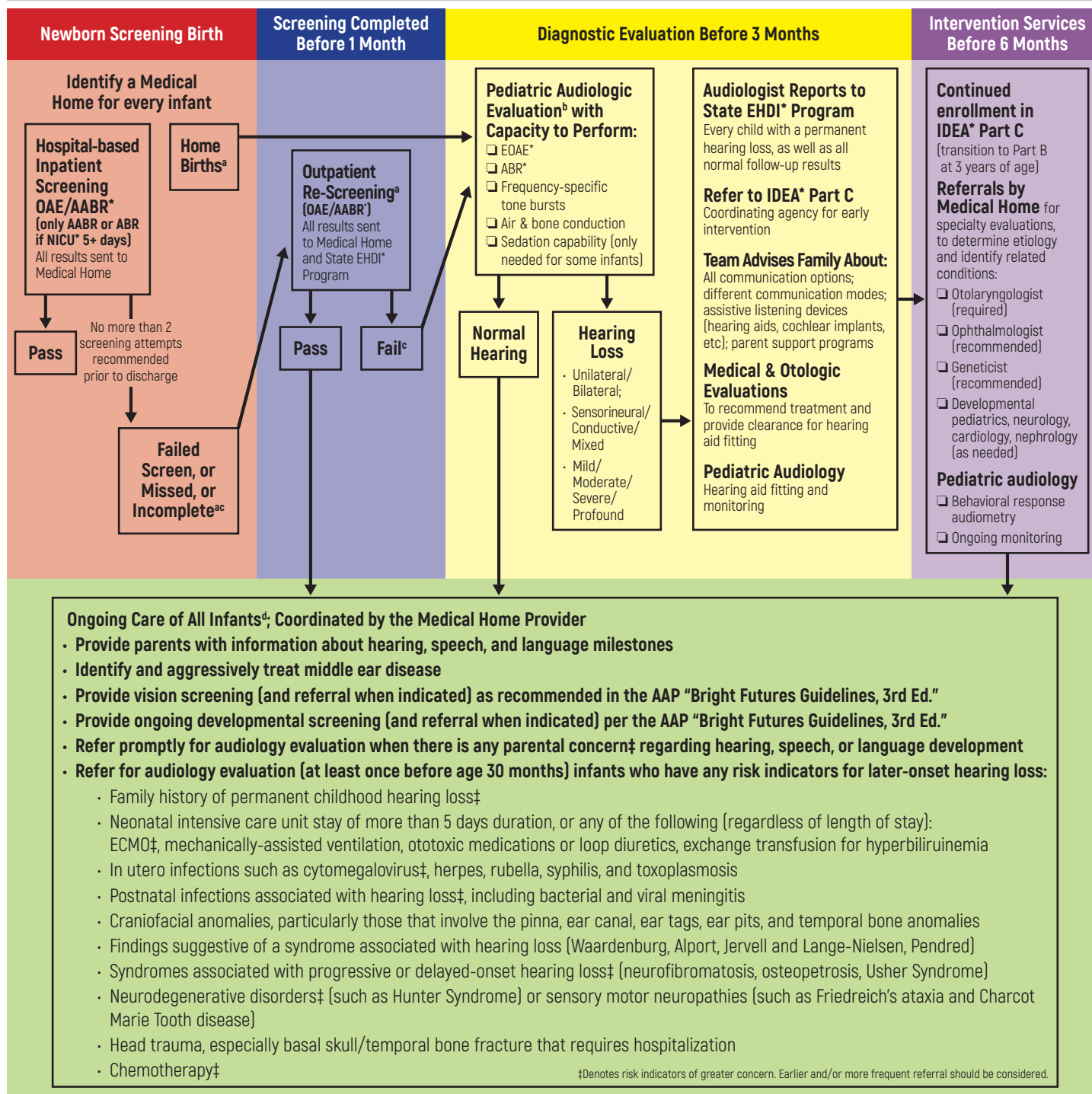
- ✓ Remove page two of the blood spot form after completing all demographic information.
- ✓ Enter the **same** information into the hearing device from this copy to ensure linkage of the hearing result to the blood spot record and laboratory report.
- ✓ Ensure that the infant's name is entered the same to include hyphens if the name is hyphenated.
- ✓ Ensure that only the unique portion of the medical record number is documented on both the blood spot form and in the hearing device with no preceding letters or zeros (ex: D00654321).
- ✓ Ensure that the infant's, not the mother's, date of birth, medical record number, and gender is used and matches on both the blood spot form and hearing device.
- ✓ Ensure that numbers are legible and do not resemble any other numbers or letters.

Reporting Hearing Results Electronically:

- ✓ Ensure the following format is used when naming files, **##H####_YYYYMMDD**.
- ✓ If more than one file is submitted daily, add "A", "B", etc. to the name to differentiate the files (ex: 45H58010_20190114A, 45H58010_20190114B).
- ✓ Each type of hearing device has a specific format in which results must be uploaded via the File Transfer Protocol (FTP). The data system cannot receive hearing results in any other format.
Written instructions for specific hearing equipment is available upon request.
- ✓ It is recommended that hospital Information Technology (IT) support is available. If updates to system security or software upgrades are completed in your hospital then core FTP software will need to be reinstalled as this may impact the ability to electronically send hearing results to the state.
- ✓ It is recommended hospitals report hearing results to the state every day an infant is screened.

If the parent refuses the blood spot screen or hearing screen, please fax a newborn screening refusal form to (334) 206-3791.

AMERICAN ACADEMY OF PEDIATRICS EARLY HEARING DETECTION & INTERVENTION (EHDI) GUIDELINES FOR PEDIATRIC PROVIDERS



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

*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.

Re-screen Newborn Hearing Results Form

ALABAMA NEWBORN HEARING PROGRAM

PHONE 334.206.2944 FAX 334.206.3791



Hearing re-screen should be completed before one month of age

NEWBORN'S NAME		DATE OF BIRTH	
HOSPITAL OF BIRTH		HOSPITAL ID NUMBER	
MOTHER'S OR GUARDIAN'S NAME (as noted per hospital records)		HOME PHONE NUMBER	
HOME ADDRESS			
PRIMARY CARE PHYSICIAN		PHYSICIAN PHONE NUMBER	
ADDRESS			
BIRTH	HEARING SCREEN PERFORMED AT BIRTH FACILITY OR HOME BIRTH	Inpatient Screen Date: _____ Right Ear: <input type="checkbox"/> Pass <input type="checkbox"/> Refer <input type="checkbox"/> Not Tested Left Ear: <input type="checkbox"/> Pass <input type="checkbox"/> Refer <input type="checkbox"/> Not Tested Method: <input type="checkbox"/> AABR <input type="checkbox"/> OAE <input type="checkbox"/> TEOAE <input type="checkbox"/> DPOAE	
		Infants who fail initial OAE screen may 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 <input type="checkbox"/> Outpatient <input type="checkbox"/>	DATE SCREENED: _____ Both ears should be tested even if only one ear did not pass the initial screen. Right Ear: <input type="checkbox"/> Pass <input type="checkbox"/> Refer <input type="checkbox"/> Not Tested Left Ear: <input type="checkbox"/> Pass <input type="checkbox"/> Refer <input type="checkbox"/> Not Tested Method: <input type="checkbox"/> AABR <input type="checkbox"/> OAE <input type="checkbox"/> TEOAE <input type="checkbox"/> DPOAE *Date referred for diagnostic evaluation: _____	
		RISK FACTORS FOR DELAYED HEARING LOSS: <input type="checkbox"/> NICU admission <input type="checkbox"/> Received ototoxic medications <input type="checkbox"/> Transfused <input type="checkbox"/> Other _____ If any risk factors present, refer for an audiology assessment by 24 to 30 months of age.	
TEST SITE NAME		PHONE	FAX
ADDRESS			

COMMENTS/FOLLOW-UP PLAN :

The completed form should be returned as soon as the hearing re-screen/initial diagnostic audiological evaluation is completed. Fax to the Newborn Hearing Screening Program at 334-206-3791 .

*If refer, infant should have diagnostic testing by three months of age per the Joint Committee on Infant Hearing.

NBS.Hearing Re-Screen Reporting Form.2018

Diagnostic Hearing Evaluation Form

ALABAMA NEWBORN HEARING PROGRAM

PHONE 334.206.2944 FAX 334.206.3791

Diagnostic testing should be completed before three months of age



NEWBORN'S NAME	DATE OF BIRTH
HOSPITAL OF BIRTH	HOSPITAL ID NUMBER
MOTHER'S OR GUARDIAN'S NAME (as noted per hospital records)	HOME PHONE NUMBER
ADDRESS	

TEST SITE

Audiology Provider Name	Phone	Fax
-------------------------	-------	-----

Address

Before 3 Months	Pediatric Diagnostic Audiology Evaluation	DIAGNOSTIC TEST DATE _____ METHOD: <input type="checkbox"/> ABR <input type="checkbox"/> AABR <input type="checkbox"/> OAE <input type="checkbox"/> TEOAE <input type="checkbox"/> DPOAE <input type="checkbox"/> Normal Hearing <input type="checkbox"/> Hearing Loss Confirmed (Please Complete Section Below)	Please select all that apply. Both ears should be tested at each visit.
	Before 6 Months	Enrollment in Early Intervention Date of Referral to EI _____ Enrollment Date _____ Medical Referral: <input type="checkbox"/> Otolaryngologist <input type="checkbox"/> Geneticist <input type="checkbox"/> Ophthalmologist <input type="checkbox"/> Pediatrician <input type="checkbox"/> Other (specify) _____ Additional Audiology Services _____	

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

*Includes fluid in the middle ear, ear infection, poor eustachian tube function, hole in eardrum, earwax, swimmer's ear, foreign body in the ear canal, and malformation of the outer ear, ear canal, or middle ear per the American Speech-Language Hearing Association.

COMMENTS/FOLLOW UP (please add other descriptors associated with hearing loss):

The completed form should be returned as soon as the hearing re-screen/initial diagnostic audiological evaluation is completed. Fax to the Newborn Hearing Screening Program at 334-206-3791.

NBS.Hearing Diagnostic Reporting Form.2018

CRS NEWBORN HEARING ASSESSMENT CLINICS

CHILDREN'S REHABILITATION SERVICE HEARING SERVICES

Communication is a critical skill that connects people. Through its Hearing Services, CRS gives children and youth a chance for success at home, at school, and in the community.

Newborn Hearing Screening

for follow-up to hearing screening done at the birthing center

Early detection of hearing loss is vital because it can help assure that your baby acquires the skills that are essential to success throughout life. Testing is the only way to know if an infant has a hearing loss. If your child is referred to CRS for a follow-up newborn hearing screening, a CRS hearing specialist (called an audiologist) will test your child, explain the results, and tell you if more tests are necessary.

Hearing Aid Clinic

- Hearing aid evaluation and prescription
- Hearing aid and ear mold ordering
- Hearing aid orientation and fitting
- Hearing aid follow-up checks
- Hearing aid maintenance and repairs
- Ear mold replacement
- Explanation of educational options
- Consultation or technical assistance for local schools
- Referral for speech-language therapy
- Education and support for the child and family

Hearing Assessment Clinic

- Evaluation by a pediatric audiologist
- Referral to Hearing Clinic and/or Hearing Aid Clinic as necessary

Hearing Clinic

- Evaluation by a pediatric audiologist
- Examination by an ear, nose, and throat (ENT) physician
- Referral for specialized testing or services (i.e., CT scans, MRI, lab work, hearing impaired education, etc.)

If you think your child might benefit from CRS Hearing Services, contact the CRS office in your area.

CRS OFFICE LOCATIONS

STATE OFFICE

602 S. Lawrence St., Montgomery, 36104
334-293-7500, 1-800-846-3697

ANDALUSIA

1082 Village Square Drive, Suite 2, 36420
334-222-5558, 1-800-723-8064

ANNISTON

1910 Coleman Road, 36207
256-240-8801, 1-800-289-9533

BIRMINGHAM

Homewood CRS
234 Goodwin Crest Drive, 35209
Community Office: 205-290-4550,
1-888-430-7423

Birmingham TCH (The Children's Hospital)
1600 Seventh Ave. South, 35233
205-939-5900, 1-800-285-9318

DOTHAN

795 Ross Clark Circle NE, 36303
334-699-6600, 1-800-677-9123

GADSDEN

1100 George Wallace Drive, 35903
256-547-8653, 1-800-289-1353

HUNTSVILLE

3000 Johnson Road, 35805
256-650-1701, 1-800-283-9352

JACKSON

1506 College Ave., 36545
251-246-4025, 1-800-283-8140

MOBILE

1610 Center St., Suite A, 36604
251-432-4560, 1-800-879-8163

MONTGOMERY

602 S. Lawrence St., 36104
334-293-7500, 1-800-568-9034

MUSCLE SHOALS

1450 E. Avalon Ave., 35661
256-381-1212, 1-800-285-9924

OPELIKA

516 W. Thomason Circle, 36801
334-749-8339, 1-800-568-8428

SELMA

2906 Citizens Parkway, 36701
334-872-8422, 1-800-967-6876

TUSCALOOSA

1110 Dr. Edward Hilliard Drive, 35401
205-759-1279, 1-800-723-0490

EHDI Program Update

CDC's Progress in Detecting Infant Hearing Loss

CDC's Early Hearing Detection and Intervention (EHDI) has made clear progress in supporting the early identification of deaf and hard of hearing (DHH) infants.

The earlier children with hearing loss are identified and start getting intervention, the more likely they will reach their full potential.



Hearing Professionals use These Important 1-3-6 Benchmarks



1

Before one month of age: Hearing Screening



3

Before three months of age: Hearing evaluation



6

Before six months of age: Early Intervention

Hearing screening is the first hearing service to determine if a baby has hearing loss.

Hearing evaluation is a comprehensive test to determine the severity of hearing loss.

Identifying hearing loss early is important

- Hearing loss is one of the most common birth defects.
- Each year 12,000 infants are born deaf or hard of hearing (DHH).
- When left undetected, a hearing loss can delay a child's speech and language development, as well as his or her thinking, learning, and social skills.
- Newborn hearing screening and intervention programs can save nearly \$200 million in additional education costs annually¹.

How CDC is helping to make progress

- CDC is responsible for collecting and analyzing EHDI data from across the United States.
- The CDC EHDI program provides technical assistance to all states and territories to help support the early identification of DHH infants.
- CDC funds the development and use of systems and data tools that help states and territories ensure DHH children receive essential services:
 - Hearing screening
 - Hearing evaluation
 - Early intervention
- Nearly all newborns are screened for hearing loss, usually before leaving the hospital.

National Center on Birth Defects and Developmental Disabilities
Division of Human Development and Disability



CS262933

ALABAMA NEWBORN HEARING PROVIDER DIRECTORY



The Early Hearing Detection & Intervention – Pediatric Audiology Links to Services (EHDI-PALS) is a web-based link to information, resources, and services for children with hearing loss. It includes a directory of facilities that offer pediatric audiology services to young children who are younger than five years of age.

For an updated list of Alabama audiology providers please visit the EHDIPALS site at the following link: www.ehdipals.org.

Are you a provider interested in listing your facility in the EHDI-PALS directory?
If so, enter your information at: www.ehdipals.org/EP_AudiologicalServiceProviders.aspx

1-3-6 NEWBORN HEARING SCREENING CHECKLIST

Patient Name:		Patient DOB:	Date of Visit:
1 INITIAL SCREENING (by no later than 1 month of age)			
Has the child had a newborn hearing screening?	Yes	No ⇒	Schedule initial screening
Did you obtain the test results from the screening hospital or state EHDl program?	Yes	No ⇒	Contact the hospital or state EHDl program
Are the results recorded in the patient's chart?	Yes	No ⇒	Record test results in patient chart
Did the child pass the newborn hearing screening?	Yes	No ⇒	Schedule rescreening appointment
Have the results been reported to the state EHDl program?	Yes	No ⇒	Confirm results have been reported to state EHDl program within 48 hours of receiving them
Have results been discussed with family?	Yes	No ⇒	<input type="checkbox"/> For a child who passed, stress the importance of ongoing surveillance and risk factors* <input type="checkbox"/> For a child who did not pass, discuss the need for follow-up and assist in arranging a rescreening
Has a rescreening occurred (if the initial screen resulted in "did not pass" or if otherwise necessary)?	Yes	No ⇒	Schedule rescreening appointment
RESCREENING (by no later than 1 month of age)			
Where will the rescreening be performed?	<input type="checkbox"/> Hospital: <input type="checkbox"/> Office <input type="checkbox"/> Other (specify): Location: _____ Date: _____		
✓ If hospital/outpatient center, when is the rescreening appointment? ✓ If conducted in office: <ul style="list-style-type: none"> Determine what screening equipment was used at the hospital. Follow the AAP office rescreening guidelines. 			
Did the child pass the rescreening?	Yes	No ⇒	Send child to audiologist with pediatric expertise for diagnostic evaluation.
Are the results recorded in the patient chart?	Yes	No ⇒	Record results in patient chart.
Have the results been discussed with the family?	Yes	No ⇒	<input type="checkbox"/> For a child who passed, stress the importance of ongoing surveillance and risk factors* <input type="checkbox"/> For a child who did not pass, discuss the need for follow-up and assist in arranging an audiologic evaluation
Have the results been reported?	Yes	No ⇒	Confirm results have been reported to state EHDl program within 48 hours of receipt
3 DIAGNOSTIC EVALUATION (by no later than 3 months of age)			
If the child did not pass the rescreening, was he/she referred to an audiologist with expertise in pediatrics?	Yes Provider: _____ Date of Visit: _____	No ⇒ Refer to audiologist with expertise in pediatrics	
Were the results of the diagnostic test normal?	Yes	No ⇒	Discuss early intervention (EI) and need for comprehensive plan
Have the results been discussed with the family?	Yes	No ⇒	<input type="checkbox"/> For a child who passed, stress the importance of ongoing surveillance and risk factors* <input type="checkbox"/> For a child who did not pass, discuss EI and need for comprehensive plan
Have the results been reported?	Yes	No ⇒	Confirm results have been reported back to state EHDl program within 48 hours of receipt
6 EARLY INTERVENTION (by no later than 6 months of age)			
If the child was diagnosed with a hearing loss, was he/she referred for early intervention and multidisciplinary evaluation?	Yes Date of visit: _____	No ⇒ Provide referral for EI, ophthalmology, and otolaryngology and offer referral for genetics	
ONGOING SURVEILLANCE AND SCREENING			
Continue to perform ongoing surveillance and screening for late-onset hearing loss, particularly children with risk factors.			

* JCIH Risk Factors



GLOSSARY OF TERMS FOR NEWBORN HEARING SCREENING

The American Academy of Pediatrics (AAP) Early Hearing Detection and Intervention (EHD) Loss to Follow-up/Documentation (LTF/D) Workgroup has compiled a glossary of terms important to newborn hearing screening and resources related to LTF/D.

TERM	DEFINITION
Newborn hearing screening (NBHS)	Hearing screening performed shortly after birth, typically performed in hospitals prior to discharge involving the use of OAEs or AABR.
Otoacoustic emissions (OAEs)	This test measures a response produced by the cochlea (outer hair cells) when a sound is presented to the ear. To conduct the test, a tiny probe is placed just inside the baby's ear canal and a soft click is presented, a tiny microphone measures the response produced by the baby's ear. The test is quick (about 5 to 10 minutes) and painless and may be performed while the baby is sleeping or lying still. Thus, OAEs reflect the status of the peripheral auditory system extending to the cochlear outer hair cells.
Automated auditory brainstem response (AABR)	This screening test measures how the hearing nerve responds to sound. Clicks are presented to the ear through a probe or soft earphones, and the neural response is measured through 3 electrodes placed on the baby's head. AABR measurements reflect the status of the peripheral auditory system, the eighth nerve, and the brainstem auditory pathway.
Outpatient rescreening	Hospital screening protocols vary and often include an outpatient screening stage. The specific technology used to conduct the outpatient screening should be based on the knowledge of how the inpatient screening was conducted. For example, when a baby fails an inpatient AABR screening, the outpatient screening must be conducted using AABR; if an OAE screening is used, auditory neuropathy will be missed. The outpatient screening may be completed at the birth hospital or by another provider, such as an audiologist, or physician.
Lost to follow-up	For infant who did not pass newborn hearing screening, "lost to follow-up" refers to a failure to receive the next step of treatment, be it rescreening or comprehensive audiologic evaluation.
Lost to documentation	Failure to report the results from hearing screening, rescreening, diagnostic services, and/or treatment services that are needed for comprehensive surveillance and monitoring by EHD and the medical home
Lost to treatment	Failure for a child with an identified hearing loss to receive needed therapeutic services and failure for families to receive needed information to support decisions regarding treatment options.
Medical home	A model for providing high-quality primary care that addresses and integrates health promotion, acute care, and chronic condition management in a planned, coordinated, and family-centered manner.
Late-onset hearing loss	A hearing loss that is not present at birth and the newborn hearing screening, which would result in a "pass."
Auditory neuropathy	Children with auditory neuropathy have evidence of normal cochlear function but show impairment in the function of the auditory nerve. Functional hearing can often be quite impaired, and diagnosis and treatment can be confusing and complicated.
Risk factors	Risk factors are indicators used (1) for the identification of infants who should receive audiologic evaluation but who live in geographic locations (eg, developing nations, remote areas) where universal hearing screening is not yet available; (2) to help identify infants who pass the neonatal screening but are at risk of developing delayed-onset hearing loss and, therefore, should receive ongoing medical, speech and language, and audiologic surveillance; and (3) to identify infants who may have passed neonatal screening but have mild forms of permanent hearing loss.



<p>JCIH 11 risk indicators</p>	<p>The Joint Commission on Infant Hearing (JCIH) lists 11 risk indicators associated with permanent congenital, delayed-onset, or progressive hearing loss in childhood (risk indicators that are marked with a "*" are of greater concern for delayed-onset hearing loss.)</p> <ol style="list-style-type: none"> 1. Caregiver concern* regarding hearing, speech, language, or developmental delay. 2. Family history* of permanent childhood hearing loss. 3. Neonatal intensive care of more than 5 days or any of the following regardless of length of stay: ECMO,* assisted ventilation, exposure to ototoxic medications (gentamicin and tobramycin) or loop diuretics (furosemide/Lasix), and hyperbilirubinemia that requires exchange transfusion. 4. In utero infections, such as cytomegalovirus,* herpes, rubella, syphilis, and toxoplasmosis. 5. Craniofacial anomalies, including those that involve the pinna, ear canal, ear tags, ear pits, and temporal bone anomalies. 6. Physical findings, such as white forelock, that are associated with a syndrome known to include a sensorineural or permanent conductive hearing loss. 7. Syndromes associated with hearing loss or progressive or late-onset hearing loss, such as neurofibromatosis, osteopetrosis, and Usher syndrome; other frequently identified syndromes include Waardenburg, Alport, Pendred, and Jervell and Lange-Nielson syndrome. 8. Neurodegenerative disorders,* such as Hunter syndrome, or sensory motor neuropathies, such as Friedreich ataxia and Charcot-Marie-Tooth syndrome. 9. Culture-positive postnatal infections associated with sensorineural hearing loss,* including confirmed bacterial and viral (especially herpes viruses and varicella) meningitis. 10. Head trauma, especially basal skull/temporal bone fracture* that requires hospitalization. 11. Chemotherapy*
---------------------------------------	---

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. **This is also a reportable diagnosis of hearing loss.**
2. Sensorineural hearing loss – occurs when there is damage to the inner ear. This may be caused by disease, birth injury, toxic drugs, or genetic syndromes.

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

- slight hearing loss
- mild hearing loss
- moderate hearing loss
- moderately severe 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. Automated Auditory Brainstem Response (AABR) – 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 screening method 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 AABR.**
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 initial hearing screening at birth, then no more than one other attempt to re-screen should be completed on the day of discharge. If the baby does not pass on discharge, an appointment should be made with an audiologist for a re-screen (second tier screen) and notify the primary care physician of appointment so he or she can send a referral.

SECTION 4 – PULSE OXIMETRY SCREENING

Critical Congenital Heart Disease (CCHD)	52
Pulse Oximetry Screening Equipment	53
Pulse Oximetry Training	54-61
Knowledge Assessment	58-59
Competency Checklist	60
Training Log	61
Pulse Oximetry Screening	62
Pulse Oximetry Screening Algorithm	63
Pulse Oximetry Reporting Form	64

CRITICAL CONGENITAL HEART DISEASE (CCHD)

INTRODUCTION

In September 2011, U.S. Department of Health and Human Services (HHS) Secretary Kathleen Sebelius approved adding Critical Congenital Heart Disease (CCHD) to the Recommended Uniform Screening Panel (RUSP). This recommendation was endorsed by the Alabama Chapter of the American Academy of Pediatrics. Donald E. Williamson, M.D., Alabama's State Health Officer, supported implementation of screening for CCHD in Alabama's birthing facilities. The Newborn Screening Program convened a CCHD Work Group that met on November 30, 2011, and again on December 13, 2011, to create a protocol for pulse oximetry screening on well infants in Alabama's fifty-three birthing facilities with a goal to implement by April 2012.

According to the Centers for Disease Control and Prevention (CDC), congenital heart defects account for 24% of infant deaths due to birth defects. In the United States, about 4,800 (or 11.6 per 10,000) babies born every year have CCHDs. In Alabama, approximately seventy infants are expected to be diagnosed with a CCHD each year. Babies with a CCHD are at significant risk for death or disability if their CCHD is not diagnosed and treated soon after birth. Pulse oximetry, which is a test to determine the amount of oxygen in the blood, is the recommended screening method to detect CCHDs in newborns.

There are seven defects classified as CCHD:

- Hypoplastic left heart syndrome
- Pulmonary atresia (with intact septum)
- Tetralogy of Fallot
- Total anomalous pulmonary venous return
- Transposition of the great arteries
- Tricuspid atresia
- Truncus arteriosus

This manual serves as a guide to assist each birthing facility to establish its own policy and procedures to implement a Critical Congenital Heart Disease Screening Program (CHDSP). These policies and procedures should establish clear, complete, and concise evidence-based policy and address the components listed below:

- Equipment
- Training
- Screening
- Education

It is recommended that each facility designate a program coordinator to facilitate the planning and implementation of the screening program, including the establishment of an interdisciplinary team. Members of this team should participate in the planning process and should represent hospital executives, physicians, nurses, and ancillary staff.

PULSE OX SCREENING TRAINING

SECTION 1 - EQUIPMENT

Each birthing facility will be responsible for selecting and securing pulse oximeter equipment for screening newborns for CCHD, if appropriate equipment is not already available. Such equipment must be compliant with national standards and adhere to the following:

- Must be motion-tolerant and report functional oxygen saturation.
- Must be validated in low-perfusion conditions.
- Must have been cleared by the FDA for use in newborns.
- Must have 2% root, mean-square accuracy.
- Must be calibrated regularly based on manufacturer guidelines.



PULSE OX SCREENING TRAINING

SECTION 2 - TRAINING

Training should be performed by qualified personnel who have participated in the planning process (e.g., unit nurse manager or assistant nurse manager, nurse educator, the program coordinator, or a registered nurse). This training should be hands-on and competency based. The training of personnel should include:

- Overview of screening protocol
- Education on the use, care, maintenance, and trouble-shooting of screening equipment
- A review of general nursery policies and procedures
- Education on the differences between adult and pediatric oximeter probes
- An explanation on the importance of adequate circulation
- The effects of hypothermia and phototherapy on pulse oximetry screening
- Facility resources for pediatric echocardiogram and referral sources when not available in house

PULSE OX SCREENING TRAINING

IN-SERVICE EDUCATION PROGRAM COMPONENTS

The following is an overview of educational tools and components that may be used to educate staff who will be directly involved in screening implementation. Educational tools discussed are included.

1. PowerPoint Presentation:

- a. Provides attendees with education on background, significance, and need for screening.
- b. Provides attendees with education on Congenital Heart Disease Screening Program (CHDSP) screening methods and guidelines.

2. Education for Providers:

- a. Provides attendees with educational tool, "Congenital Heart Disease Screening Program: Education for Providers," which includes an overview of pulse oximetry, congenital heart disease, and pulse oximetry screening for critical congenital heart disease.

3. Pulse Oximetry Demonstration:

- a. Provide attendees with a demonstration of correct and safe use of pulse oximetry equipment in obtaining an accurate infant reading by in-service facilitator or representative from pulse oximeter manufacturer.
- b. Provide attendees with an opportunity to practice performing pulse ox screening on a doll.
- c. Provide attendees with the opportunity to ask questions regarding correct and safe methods for performing pulse ox screening.
- d. Provide attendees with the "Performing Pulse Oximetry (Pulse Ox) with the Infant Patient: Education for Providers" and "Pulse Ox Placement" educational tools.

4. Knowledge Assessment Quiz:

- a. Allow time for attendees to complete the "Knowledge Assessment Quiz."
- b. Review the correct answer for each question.
- c. Allow time for remediation of questions answered incorrectly.
- d. Allow time for attendees to re-take quiz, if necessary.

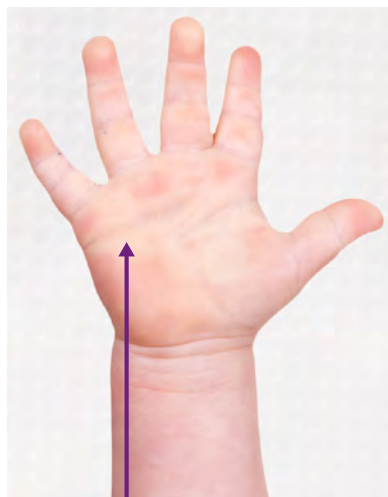
5. Competency Checklist:

- a. Allow adequate time for completion of competency checklist.
- b. Provide each attendee with a copy of the complete competency checklist to forward to his or her manager.

PULSE OX SCREENING TRAINING

PULSE OX PROBE PLACEMENT EDUCATION

1. Select application site on the outside, fleshy area of the infant's hand or foot.



RH Application Site



Foot Application Site

2. Place the photodetector portion of the probe on the fleshy portion of the outside of the infant's hand or foot.
3. Place the light emitter portion of the probe on the top of the hand or foot. Place the photodetector directly opposite of light emitter, on the bottom of the hand or foot.
4. Remember: The photodetector and emitter must be directly opposite each other in order to obtain an accurate reading.
5. Secure the probe to the infant's hand or foot using the adhesive or foam tape recommended by the vendor. It is not recommended to use tape to secure probe placement.
6. Some vendors use visual images such as a star or bar to specify which side of the probe should be placed on top of the hand or foot. You may choose to use a helpful statement such as, "Raise the bar" to help you to remember proper probe placement.



PULSE OX SCREENING TRAINING

PERFORMING PULSE OXIMETRY (PULSE OX) WITH THE INFANT PATIENT: EDUCATION FOR PROVIDERS

Pulse Ox – Dos

1. If you are using disposable pulse ox probes, use a new, clean probe for each infant. If you are using reusable pulse ox probes, clean the probe with recommended disinfectant solution between each infant. Dirty probes can decrease the accuracy of your reading and can transmit infection. A disposable wrap should be used to secure the probe to the site.
2. The best sites for performing pulse ox on infants are around the palm and the foot. An infant pulse ox probe (not an adult pulse ox clip) should always be used for infants.
3. When placing the sensor on the infant's skin, there should not be gaps between the sensor and the infant's skin. The sides of the probe should be directly opposite of each other.
4. Nail polish dyes and substances with dark pigmentation (such as dried blood) can affect the pulse ox reading. Assure that the skin is clean and dry before placing the probe on the infant. Skin color and jaundice do not affect the pulse ox reading.
5. Movement, shivering and crying can affect the accuracy of the pulse ox reading. Ensure that the infant is calm and warm during the reading. Swaddle the infant and encourage family involvement to promote comfort while obtaining the reading. If possible conduct screening while the infant is awake.
6. Pulse oximeters have different confidence indicators to ensure that the pulse ox reading is accurate. Determine the confidence indicators for the pulse oximetry equipment that you are using.
7. If an infant requires pulse ox monitoring for an extended amount of time, assess the site where the probe is placed at least every two hours. Monitor for signs of irritation and burning of the skin.

Pulse Ox – Don'ts

1. Never use an adult pulse ox clip when obtaining a pulse ox reading for an infant. Using an adult clip on an infant will give you an inaccurate reading.
2. Blood flow is needed to obtain an accurate pulse ox reading. Never attempt to obtain a pulse ox reading on the same extremity that you have an automatic blood pressure cuff.
3. Bright or infrared light, including bilirubin lamps and surgical lights, can affect the accuracy of the reading. Ensure that the infant is not placed in bright or infrared light while pulse ox is being performed. You may cover the pulse ox probe with a blanket to ensure that extraneous light does not affect the accuracy of your reading.
4. Do not use tape to apply the pulse ox probe to the infant's skin.

Pulse Ox - Caution!

1. The pulse is needed to determine the oximetry reading. Pulse ox is not accurate if the patient is coding or is having a cardiac arrhythmia. Remember: No pulse, no oximetry!
2. Pulse ox readings are not instantaneous. The oximetry reading that is displayed on the monitor is an average of readings over the past few seconds.

PULSE OX SCREENING TRAINING

KNOWLEDGE ASSESSMENT

1. The following can affect the accuracy of the pulse oximetry (pulse ox) reading:
 - a. Movement
 - b. Cold extremities or shivering
 - c. Crying
 - d. Bilirubin lamps and surgical lights
 - e. All of the above
2. One clean, disposable pulse ox probe can be used on up to five patients.
 - a. True
 - b. False
3. All of the following can affect the accuracy of the pulse ox reading except:
 - a. Placing the pulse ox probe on the same extremity that you are taking the blood pressure
 - b. Performing the pulse ox test while the infant is crying
 - c. Using a clip on the finger of an infant
 - d. Infant skin color or jaundice
4. Pulse ox screening will detect all forms of CHD
 - a. True
 - b. False
5. The screening guidelines state that pulse ox should be performed on:
 - a. The right hand
 - b. One foot
 - c. Both a and b
 - d. Neither a or b
6. Pulse ox screening should be performed when the infant is what age?:
 - a. Less than 8 hours
 - b. Between 8 hours and 18 hours
 - c. Greater than 24 hours
 - d. Less than 24 hours
7. An infant's pulse ox readings should be reported to the physician or nurse practitioner caring for the infant if:
 - a. Pulse ox readings are greater than 94% for both right hand and one foot and there is a difference of 4 or more between the two on three measures each separated by one hour
 - b. Pulse ox readings are less than 95% for both right hand and one foot or there is a difference of 4 between the two on three measures each separated by one hour
 - c. Pulse ox reading is less than 90% for either or both the right hand and one foot
 - d. All of the above
8. Pulse ox screening results can be shared with individuals that are not directly involved in the patient's care:
 - a. True
 - b. False

PULSE OX SCREENING TRAINING

KNOWLEDGE ASSESSMENT ANSWERS

1. The following can affect the accuracy of the pulse oximetry (pulse ox) reading:
 - a. Movement
 - b. Cold extremities or shivering
 - c. Crying
 - d. Bilirubin lamps and surgical lights
 - e. All of the above
2. One clean, disposable pulse ox probe can be used on up to five patients.
 - a. True
 - b. False
3. All of the following can affect the accuracy of the pulse ox reading except:
 - a. Placing the pulse ox probe on the same extremity that you are taking the blood pressure
 - b. Performing the pulse ox test while the infant is crying
 - c. Using a clip on the finger of an infant
 - d. Infant skin color or jaundice
4. Pulse ox screening will detect all forms of CHD
 - a. True
 - b. False
5. The screening guidelines state that pulse ox should be performed on:
 - a. The right hand
 - b. One foot
 - c. Both a and b
 - d. Neither a or b
6. Pulse ox screening should be performed when the infant is what age?:
 - a. Less than 8 hours
 - b. Between 8 hours and 18 hours
 - c. Greater than 24 hours
 - d. Less than 24 hours
7. An infant's pulse ox readings should be reported to the physician or nurse practitioner caring for the infant if:
 - a. Pulse ox readings are greater than 94% for both right hand and one foot and there is a difference of 4 or more between the two on three measures each separated by one hour
 - b. Pulse ox readings are less than 95% for both right hand and one foot or there is a difference of 4 between the two on three measures each separated by one hour
 - c. Pulse ox reading is less than 90% for either or both the right hand and one foot
 - d. All of the above
8. Pulse ox screening results can be shared with individuals that are not directly involved in the patient's care:
 - a. True
 - b. False

PULSE OX SCREENING TRAINING

COMPETENCY CHECKLIST

• Competency Title: Congenital Heart Disease Screening Process

• Competency Criteria includes the following:

1. Completion of the in-service education.
2. Accomplishment of 90 percent or more on the knowledge assessment quiz with remediation as necessary.
3. Appropriate application of pulse oximetry.
4. Accurate reading and documentation of the pulse oximetry readings.

• Competency Statement: Proficiently perform the required activities defined in research protocol.

Validation Criteria: A. Discussion (D) C. Written Test (T)
B. Verbal Feedback (VF) D. Return Demonstration (RD)

Directions for completing evaluation form: Evaluator, please circle the appropriate method of validation, initial each line and place signature in the appropriate place at the end of the document.

Name: _____ Job Title: _____

Competency	Date	Method of Validation	Supervisor Initials	Comments
Explains screening eligibility guidelines for pulse oximetry screening		D VF T		
Identifies safe and correct methods for performing pulse oximetry		D VF T RD		
Describes methods to ensure that pulse oximetry reading is accurate		D VF T RD		
Explains screening methods and guidelines for pulse oximetry screening		D VF T		
Discuss HIPAA confidentiality standards		D VF T		

Name: _____ Date: _____

Supervisor Name (Printed): _____

Supervisor Signature: _____

PULSE OX SCREENING TRAINING

TRAINING LOG *(For the records of unit managers or nursing educators)*

Employee Name and Title	Date	Completion of Competency Checklist		Manager Initials
		Yes	No	

**Each employee responsible for performing pulse oximetry screening methods should complete the competency checklist prior to participation.*

Unit: _____

Supervisor Name (Printed): _____

Manager Signature: _____

PULSE OX SCREENING SCREENING

SECTION 3 - SCREENING

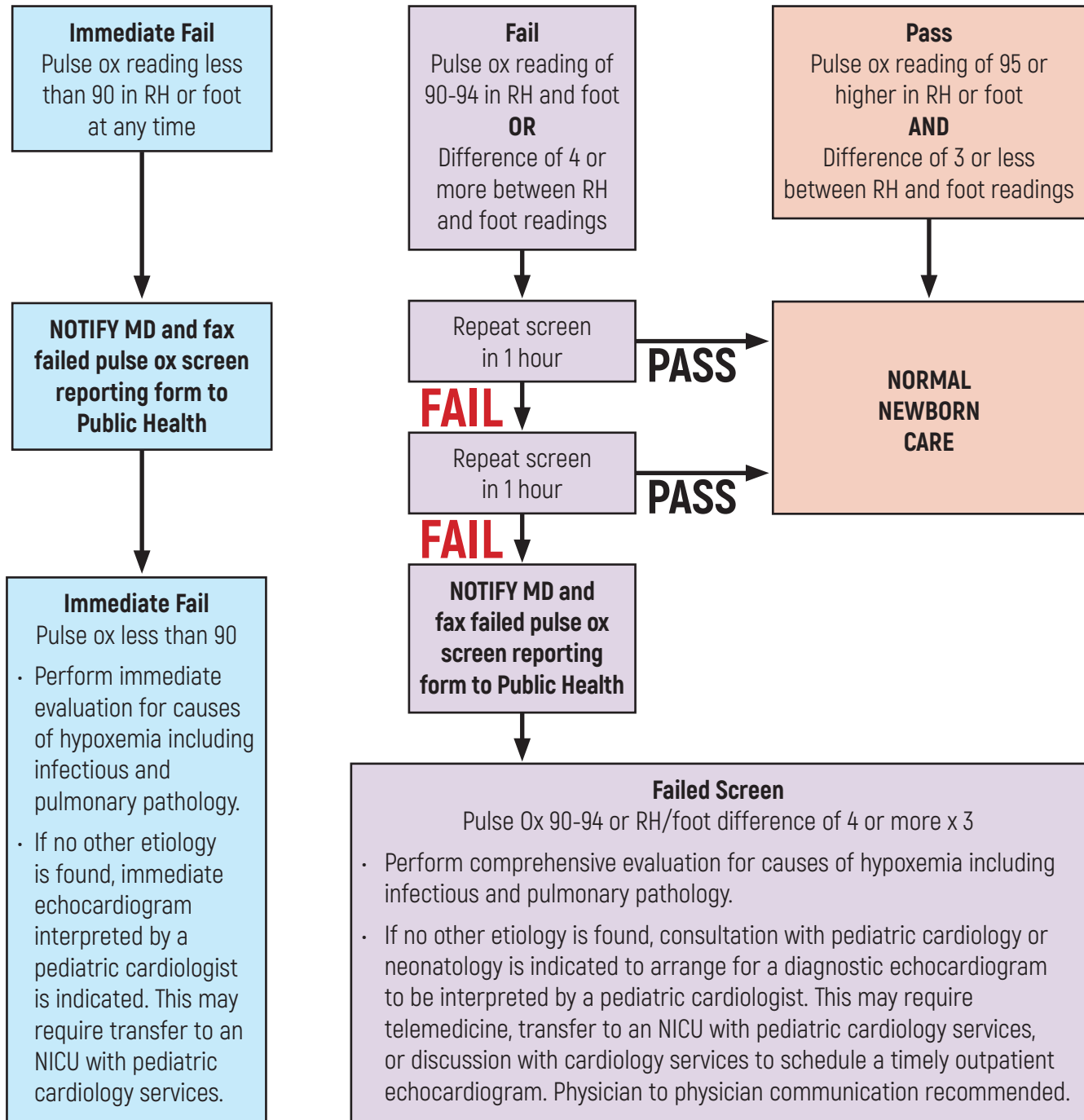
Supplies for screening

- Pulse Oximeters
 1. At least one motion-tolerant pulse oximeter to be used for screening
 2. One motion-tolerant pulse oximeter for back-up
- Infant Disposable or Reusable Pulse Ox Sensors
 1. If using disposable sensors, one disposable sensor for every infant screened
 2. If using reusable sensors, one reusable sensor for each pulse oximeter. Also consider additional reusable sensors for back-up
 - a. Disinfecting agent recommended by pulse oximetry equipment manufacturer
 3. One disposable wrap per infant screened to secure sensor to hand or foot
- Rolling Cart for Supplies
- Data Collection Forms
 1. One for every infant screened
- Dedicated individual to perform screening
- Red Heart-Shaped Stickers
 1. One red heart-shaped sticker for every infant who has been screened (optional)
- Blankets for warming the infant and blocking extraneous light
- A parent for comforting infant during screening (optional)

PULSE OX SCREENING ALGORITHM

Screen

Obtain pulse oximetry reading on right hand (RH) and either foot (in parallel or direct sequence) at 24-48 hours of age (infant should be on room air, warm and quiet, with screening sites clean and dry)



• 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>

PULSE OX SCREENING REPORTING FORM

FAILED 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 (right hand – 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 (right hand – 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 (right hand – foot) _____ ☐ Fail

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

Transferred: _____

Provider referred to: _____

Screeener's First Initial/Last Name: _____ Date: ____ / ____ / ____

SECTION 5 – RESOURCES

Public Health Districts	66
Alabama County Health Departments	67
Alabama Early Intervention System (AEIS)	68-69
Child Find Referral Form	70
Children’s Rehabilitation Service (CRS)	72-73
Alabama Community-Based Sickle Cell Organizations	74

PUBLIC HEALTH DISTRICTS

EAST CENTRAL DISTRICT

Richard Burleson, District Administrator
3060 Mobile Highway
Montgomery, AL 36108
(334) 293-6400
Connie King, Assistant District Administrator
1850 Crawford Rd.
Phenix City, AL 36867
(334) 297-0251

JEFFERSON COUNTY

Mark E. Wilson, M.D., County Health Officer
David Hicks, D.O., M.P.H., Deputy Health Officer
1400 Sixth Ave. S.
Birmingham, AL 35233
(205) 933-9110

MOBILE COUNTY

Bernard H. Eichold, II, M.D.
County Health Officer
Susan Stiegler, Assistant Health Officer
251 N. Bayou St.
Mobile, AL 36603
(251) 690-8827

NORTHEASTERN DISTRICT

Karen Landers, M.D., District Medical Officer
Mary Gomillion, District Administrator
Mark Johnson, Assistant District Administrator
709 E. Broad St.
Gadsden, AL 35903
(256) 547-6311

NORTHERN DISTRICT

Karen Landers, M.D., District Medical Officer
1000 S. Jackson Hwy.
Sheffield, AL 35660
(256) 383-1231
Judy Smith, District Administrator
Michael Glenn, Assistant District Administrator
3821 Highway 31 South
Decatur, AL 35603
(256) 340-2113

SOUTHEASTERN DISTRICT

Corey Kirkland, District Administrator
1781 E. Cottonwood Rd.
Dothan, AL 36301
(334) 792-9070

SOUTHWESTERN DISTRICT

Chad Kent, District Administrator
Suzanne Terrell, Assistant District Administrator
1115 Azalea Place
Brewton, AL 36426
(251) 947-1645
303 Industrial Drive
Linden, AL 36748
(334) 295-1000

WEST CENTRAL DISTRICT

Stacey Adams, District Administrator
2350 Hargrove Rd., E.
Tuscaloosa, AL 35405
(205) 554-4500



ALABAMA COUNTY HEALTH DEPARTMENTS

East Central District

Autauga	334-361-3743
Bullock	334-738-3030
Chambers	334-756-0758
Elmore	334-567-1171
Lee	334-745-5765
Lowndes	334-548-2564
Macon	334-727-1800
Montgomery	334-293-6400
Russell	334-297-0251
Tallapoosa	256-329-0531

Jefferson

Central Health	205-933-9110
Eastern Health	205-591-5180
Morris Health	205-933-4242
Western Health	205-715-6121

Mobile County

Keeler Clinic	251-690-8158
Semmes Clinic	251-445-0582
Citronelle Clinic	251-866-9126
Eight Mile Clinic	251-456-1399
Dauphin Island	251-445-3450
Newburn Clinic	251-405-4524
North Mobile	251-829-9884
Southwest Mobile	251-666-7413

Northeastern District

Blount	205-274-2120
Calhoun	256-237-7523

Cherokee	256-927-3132
Clay	256-396-6421
Cleburne	256-463-2296
DeKalb	256-845-1931
Etowah	256-547-6311
Randolph	334-863-8981
St. Clair	205-338-3357
Shelby	205-664-2470
Talladega	256-362-2593

Northern District

Colbert	256-383-1231
Cullman	256-734-1030
Franklin	256-332-2700
Jackson	256-259-4161
Lauderdale	256-764-7453
Lawrence	256-974-1141
Limestone	256-232-3200
Madison	256-539-3711
Marion	205-921-3118
Marshall	256-582-3174
Morgan	256-353-7021
Winston	205-489-2101

Southeastern District

Barbour	334-687-4808
Butler	334-382-3154
Coffee	334-347-9574
Covington	334-222-1175
Crenshaw	334-335-2471

Dale	334-774-5146
Geneva	334-684-2256
Henry	334-585-2660
Houston	334-678-2800
Pike	334-566-2860

Southwestern District

Baldwin	251-947-1910
Choctaw	205-459-4026
Clarke	251-275-3772
Conecuh	251-578-1952
Dallas	334-874-2550
Escambia	251-867-5765
Marengo	334-295-4205
Monroe	251-575-3109
Washington	251-847-2245
Wilcox	334-682-4515

West Central District

Bibb	205-926-9702
Chilton	205-755-1287
Fayette	205-932-5260
Greene	205-372-9361
Hale	334-624-3018
Lamar	205-695-9195
Perry	334-683-6153
Pickens	205-367-8157
Sumter	205-652-7972
Tuscaloosa	205-562-6900
Walker	205-221-9775

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
- well-baby clinics
- hospital follow-up clinics
- physicians
- pediatricians' offices
- community health services
- developmental disabilities programs
- prenatal/postnatal facilities
- child day care centers
- home child day care programs
- Head Start programs
- local educational agencies
- outpatient clinics
- public health facilities
- Medicaid programs
- hospitals
- social service 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; the language spoken by the family; the areas of development that are of concern to the parents and professional; the name of the child's primary care physician; and acceptance/refusal of the referral to AEIS, from the family.

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.

ALABAMA'S EARLY INTERVENTION SYSTEM FOR INFANTS AND TODDLERS WITH DISABILITIES

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.



Alabama's Early Intervention System

Child Find Referral Form

To make a referral by phone: 1-800-543-3098

Mail to: ADRS/EI, 602 S. Lawrence St., Montgomery, AL 36104 or Fax to: Child Find Fax # (334) 293-7393

or send via email to: REHAB--Childfind@rehab.alabama.gov

For more info, please visit: <http://rehab.alabama.gov/individuals-and-families/early-intervention>

Please print clearly and complete all blanks - no stamps or labels

INFANT/TODDLER INFORMATION

1. SSN# (if available): _____ 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 (if different from above): _____
- City/State/Zip: _____ 18. County: _____
19. Primary contact #: () _____ 20. Alternate contact #: () _____
- Alternate contact #: () _____ Work Phone #: () _____ Ext #: _____
- Primary Contact Email address: _____

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 # (leave blank if unknown): _____
- Date Mailed/Faxed to Child Find: _____ Sender's Name/Phone #: _____

PHYSICIAN/CRNP USE ONLY

28. I certify that the child named above has a confirmed diagnosis of _____
29. Printed Name of Physician/CRNP: _____ 30. Phone #: _____
31. Signature of Physician/CRNP: _____ Today's date: _____

STATE OFFICE USE ONLY

- New Case ID#: _____ SS# or T#: _____
- Referral taken by: _____ Date taken: _____ Received by: ☐ phone ☐ email ☐ fax Processed by: _____ Official referral/entry date: _____
- ☐ ATTACHMENT: _____ ☐ Signed release of information

Revised 01/2019

Alabama's Early Intervention System (AEIS) – Child Find Referral Info Sheet

IMPORTANT NOTE: Question #'s 2 through 7 and 11 through 27 are required information

INCOMPLETE REFERRALS WILL NOT BE ACCEPTED (FILL IN ALL REQUIRED BLANKS)

1. Please provide the SS# if available, however, if the number is unavailable we can assign a pseudo number in order to process the referral.
5. Please answer either yes or no. We cannot process the referral without this information.
6. Enter the primary race that the family identifies. If the child is of multiple races, check all boxes that apply.
American Indian or Alaska Native – A person having origins in any of the original peoples of North and South America (including Central America) and who maintains tribal affiliation or community attachment. (Does not include persons of Hispanic/Latino ethnicity)
Asian – A person having origins in any of the original peoples of the Far East, Southeast Asia, or Indian subcontinent. This includes for example, Cambodia, China, India, Japan, Korea, Malaysia, Pakistan, the Philippine Islands, Thailand, and Vietnam. (Does not include persons of Hispanic/Latino ethnicity)
Black or African American – A person having origins in any of the Black racial groups of Africa. (Does not include persons of Hispanic/Latino ethnicity)
Hispanic or Latino – A person Cuban, Mexican, Puerto Rican, South or Central American, or other Spanish culture or origin, regardless of race.
Native Hawaiian or Other Pacific Islander – A person having origins in any of the original peoples of Hawaii, Guam, Samoa, or other Pacific Islands. (Does not include persons of Hispanic/Latino ethnicity)
White – A person having origins in any of the original peoples of Europe, the Middle East, or North Africa. (Does not include persons of Hispanic/Latino ethnicity)
Two or More Races – A person having origins in two or more of the six race categories listed immediately above. (Does not include persons of Hispanic/Latino ethnicity)
7. If the family is multi-lingual and English is one of the languages spoken, please enter English. If English is not spoken in the home, please enter the language spoken so that an interpreter can be obtained, if needed.
8. Not required, but please enter if available.
11. Enter the first and last name of the primary caregiver of which the child lives with.
12. How is this person that the child lives with related to the child? (mother, father, aunt, foster parent, etc.)
13. Is the person named the child's primary caregiver?
14. Does the child live with the person named?
15. Enter the address where correspondence for this child should be sent.
17. Where does the family live (if different from mailing address)? This determines which program will serve the child.
19. Provide all available contact information for the family.
21. The name of the person making this referral.
22. The organization affiliated with the person making the referral or description of who that person is (for example, Children's Hospital, ABC Therapy Company, DPS, grandfather).
23. -25. Demographic and contact information for the referral source.
27. Who told the family about Early Intervention? Please choose one of the following:
Agency, APC Parenting Kit, Audiologist, Certified Registered Nurse Practitioner, Child Care, Developmental Follow Up Clinic, Doctor, Early Head Start, EI Program, EI Recipient's family, Head Start, Healthy Child Care Alabama, High Risk Clinic, Hospital, Hurricane Katrina Evatee, Interpreter, Media, Military, Nurse-Family Partnership, Other, PA Materials, Parent Assistance Line (PAL), Parent (Previously Received EI Services), Receiving Service in Other State, Relative/Friend, School System, Self, Social Media (Facebook, Twitter, Etc.), Social Worker, SSA, Therapist, Web Site
In additional information, please enter any other information that may be useful in helping us serve this child. Please enter when this referral was sent to Child Find and who sent it along with their phone number so that we can call if there are any questions.
28. This section can only be completed by a physician or nurse practitioner who is making the referral. In order to expedite eligibility determination, a physician/nurse practitioner can provide documentation of any diagnoses the child may have. We must have the physician/nurse practitioner's name and signature along with the diagnosis.

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: 334-745-7579 or 1-800-568-8428 Counties: Chambers, Lee, Macon, Randolph, Russell, Tallapoosa
Colbert County – Muscle Shoals CRS 714 State Street, Muscle Shoals, AL 35661 Phone: 256-381-4047 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 720 Alabama Avenue, Selma, AL 36701 Phone: 334-877-2900 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 1400 James I. Harrison, Jr. Parkway East, Suite 100 Tuscaloosa, AL 35405 Phone: 205-562-1802 or 1-800-723-0490 Counties: Bibb, Fayette, Greene, Hale, Lamar, Pickens, Sumter, Tuscaloosa

CHILDREN'S REHABILITATION SERVICE MAP



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 these families.

Organization	Contacts	Address & Phone	Counties
Sickle Cell Disease Association of America Central Alabama Chapter Service Area I	Claudette Stallworth <u>Executive Director</u> Ms. Sharon Lewis	3813 Avenue I Ensley Birmingham, AL 35218 205-780-2355 Fax: 205-780-2368 www.sicklecellbham.org	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	<u>Executive Director</u> Jennifer Harris Sherman	3011 5th Street Northport, AL 35476 205-758-1761 Fax: 205-758-1781	Bibb, Fayette, Green, Hale, Lamar, Marion, Pickens, Sumter, Tuscaloosa, Winston
Sickle Cell Foundation of Greater Montgomery, Inc. Service Area IV	Monique Hopkins <u>Executive Director</u> Monica Vandiver	3180 US Highway 80 West P.O. Box 9278 Montgomery, AL 36087 334-286-9122 Fax: 334-286-4804 www.riverregionsicklecell.com	Autauga, Butler, Chambers, Chilton, Coffee, Coosa, Crenshaw, Dallas, Elmore, Lowndes, Montgomery, Tallapoosa, Wilcox
Southeast Alabama Sickle Cell Association Service Area V	Janie Cowan <u>Executive Director</u> Shelby B. Powell	P.O. Box 1079 Tuskegee, AL 36087 334-727-6120 www.seasca.com	Barbour, Bullock, Dale, Geneva, Henry, Houston, Lee, Macon, Marengo, Perry, Pike, Russell
Sickle Cell Disease Association of America Mobile Chapter, Inc. Service Area VI	<u>Executive Director</u> Keava Boswell Jones	P.O. Box 40696 1453 Springhill Avenue Mobile, AL 36604 251-432-0301 www.scdamobile.org	Baldwin, Choctaw, Clarke, Conecuh, Covington, Escambia, Mobile, Monroe, Washington
North Alabama Sickle Cell Foundation, Inc. Service Area VII	<u>Executive Director</u> Pamela Thompson	P.O. Box 813 Huntsville, AL 35804 256-536-2723 1-800-636-2723 Fax: 256-536-2714 www.sicklecellna.org	Colbert, DeKalb, Franklin, Jackson, Lauderdale, Lawrence, Limestone, Madison, Marshall, Morgan

APPENDIX

Alabama Newborn Screening Timeline.....	76
Newborn Screening Disorder Descriptions.....	77-80
Alabama Newborn Screening Confirmed Disorders.....	81
Alabama Newborn Screening Public Health Law	82
Alabama State Board of Health Administrative Code	83-88
American College of Medical Genetics ACT Sheets	89

ALABAMA NEWBORN SCREENING TIMELINE

1964	PKU	10/2006	Organic Acid Disorders: Glutaric Acidemia (GA-1) Isovaleric Acidemia (IVA) Multiple carboxylase (MCD) 3-Hydroxy 3-methylglutaric Aciduria (HMG)
1978	Congenital Hypothyroidism		
1987	Hemoglobinopathies		
1992	Galactosemia	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)
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)		Organic Acid Disorders: 3-Methylcrotonyl-CoA carboxylase (3-MCC) Beta ketothiolase (BKT) Carnitine palmitoyltransferase 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)
		10/2018	Severe Combined Immunodeficiency (SCID)
		*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 corticosteroids
- occurs 1 in 15,000 newborns
- masculinization of genitals, infertility in males and females, shock and/or death if untreated
- treatment may include steroids 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 ileus), 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 cystathionine, 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/lowfat diet, nutritional supplements, and avoidance of fasting

Maple Syrup Urine Disease (MSUD)

- amino acid disorder affecting branched 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

Severe Combined Immunodeficiency (SCID)

- group of very rare inherited disorders resulting in a defect in the immune system
- occurs 1 in 50,000 newborns
- may lead to deadly infections if untreated
- most effective treatment is a bone marrow transplant within first few months of life

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)

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

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

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

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

ALABAMA NBS CONFIRMED DISORDERS

Disorders	2012	2013	2014	2015	2016	2017
Biotinidase Deficiency	0	0	0	2	0	0
Classical Galactosemia	3	1	2	4	3	3
Cystic Fibrosis	14	12	19	14	18	11
Hearing Loss	60	54	42	39	103	102
Critical Congenital Heart Disease	3	2	2	1	3	0
Endocrine Disorders						
Congenital Hypothyroidism	32	36	37	28	42	35
Congenital Adrenal Hyperplasia	4	2	4	5	4	6
Hemoglobinopathy						
Sickle Cell Disease	52	53	58	42	47	63
Sickle Cell Trait	1866	1817	1851	1479	1861	1917
Amino Acid Disorders						
Phenylketonuria	2	4	4	1	2	2
Homocystinuria	0	0	0	0	0	0
Maple Syrup Urine Disease (MSUD)	0	1	0	0	0	0
Citrullinemia	0	0	0	0	0	0
Tyrosinemia	0	0	0	0	0	0
Fatty Acid Disorders						
Carnitine Uptake Defect	1	1	1	0	1	0
Medium chain Acyl-CoA Dehydrogenase Deficiency (MCAD)	4	3	3	4	2	7
Long chain Acyl-CoA Dehydrogenase Deficiency (LCAD)	0	1	0	0	0	0
Very long chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)	1	1	4	2	3	1
Trifunctional Protein Deficiency	0	0	0	0	0	0
Organic Acid Disorders						
Glutaric Acidemia	0	0	0	0	0	1
Isovaleric Acidemia	0	0	1	2	0	0
Propionic Acidemia	0	0	1	0	0	1
Methylmalonic Acidemia (MMA)	1	1	1	0	0	0
3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC)	2	0	0	0	0	0
2-Methylbutyryl-CoA Dehydrogenase Deficiency	1	0	1	0	0	0

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
420-10-1-.02	Definitions
420-10-1-.03	Designation Of Additional Heritable Diseases
420-10-1-.04	Reporting And Notification
420-10-1-.05	Counseling And Management
420-10-1-.06	Fees

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, severe combined immunodeficiency, 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. **Amended:** Filed January 19, 2017; effective March 5, 2017.

420-10-1-.02 Definitions.

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

ALABAMA STATE BOARD OF HEALTH

ADMINISTRATIVE CODE

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

(c) **Hemoglobinopathy** - 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.

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

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

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

(k) **Organic acid disorders** B 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.

(n) **Severe Combined Immunodeficiency (SCID) and Related T-Cell Lymphocyte Deficiencies** - a group of rare inherited immune disorders in which T lymphocytes are either absent or compromised.

ALABAMA STATE BOARD OF HEALTH ADMINISTRATIVE CODE

(o) **Licensed Midwife** – a practitioner who holds a certified professional midwife credential and is licensed by the Alabama State Board of Midwifery to practice midwifery.

Authors: Thomas M. Miller, M.D.; William J. Callan, Ph.D.; Lucinda G. Ashley, R.N. – B.C.; Rachael N. Montgomery, B.S.N., R.N.

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: Filed December 17, 2007; effective January 21, 2008.

Amended: Filed May 17, 2013; effective June 21, 2013. **Amended:** Filed January 19, 2017; effective March 5, 2017. **Amended:** Filed July 19, 2018; effective September 2, 2018.

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, hemoglobinopathy, biotinidase deficiency, cystic fibrosis, aminoacidopathies, fatty acid oxidation disorders, organic acidurias and acidemias, CCHD, SCID 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: Filed December 17, 2007; effective January 21, 2008.

Amended: Filed May 17, 2013; effective June 21, 2013. **Amended:** Filed January 19, 2017; effective March 5, 2017.

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, CCHD, SCID, and other heritable disease testing to the submitting health care provider. Test results on

ALABAMA STATE BOARD OF HEALTH ADMINISTRATIVE CODE

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 Secure Remote Viewer 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.

(6) A licensed midwife must refer all newborns in his or her care to a licensed physician within 24 hours of age to perform Newborn Screening Tests which include: 1) bloodspot specimen tests; 2) newborn hearing screening tests; and 3) pulse oximetry screening tests. The licensed midwife must instruct the client regarding the requirements of the administration of these newborn health screening tests by the Alabama Department of Public Health.

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

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: Filed December 17, 2007; effective January 21, 2008.

ALABAMA STATE BOARD OF HEALTH

ADMINISTRATIVE CODE

Amended: Filed May 17, 2013; effective June 21, 2013. **Amended:** Filed January 19, 2017; effective March 5, 2017. **Amended:** Filed July 19, 2018; effective September 2, 2018.

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, CCHD, SCID, and other heritable diseases to notify them of positive test results and to 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.

(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: Filed December 17, 2007; effective January 21, 2008.

Amended: Filed May 17, 2013; effective June 21, 2013. **Amended:** Filed January 19, 2017; effective March 5, 2017.

420-10-1-.06 Fees. The Board shall assess and collect newborn

ALABAMA STATE BOARD OF HEALTH ADMINISTRATIVE CODE

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 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.

ACT SHEETS

The following ACT Sheets were developed by the American College of Medical Genetics and have been updated to include information specific to the Alabama Newborn Screening Program. These ACT sheets, as well as some not included here, can be found 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.

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
Newborn Screening Contact: Alicia Roberts, R.D.
Phone: 205-996-6983
1530 3rd Avenue South, Kaul 241
Birmingham, AL 35294

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

© American College of Medical Genetics, 2010 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957)



American College of Medical Genetics
Medical Genetics: Translating Genes Into Health®

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-OHP (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.
Preterm (<2500 grams) collected ≤ 24 hours of age	CAH > 150	2. Evaluate infant and consult with pediatric endocrinologist if considered appropriate.
Any Age and Birthweight	2 borderline CAH (45-65)	1. Diagnostic testing to include 17-OHP is recommended.

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

© American College of Medical Genetics and Genomics, 2012 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957)

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	<ol style="list-style-type: none"> 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	

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

© American College of Medical Genetics and Genomics, 2012 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957)



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:
<ul style="list-style-type: none"> • 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
<ul style="list-style-type: none"> • Ultrahigh IRT > 170 • Collected > 24 hours of age • No mutations of any type reported 	Diagnostic sweat test indicated
<ul style="list-style-type: none"> • 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
<p>*Disease-Causing Mutation – a difference in the genetic code that leads to an abnormal protein</p> <p>**Benign variant – change in the DNA code that has been studied and does not cause disease</p>	

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

© American College of Medical Genetics and Genomics, 2012 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957)

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 $\geq 38.5^{\circ}$ 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
251-405-5121

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

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

© American College of Medical Genetics and Genomics, 2012 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957)



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 $\geq 38.5^{\circ}\text{C}$ (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:

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
251-405-5121

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

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

© American College of Medical Genetics and Genomics, 2012 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957)



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 $\geq 38.5^{\circ}\text{C}$ (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
Felicia Wilson, M.D.
Pediatric Hematology/Oncology
1504 Springhill Avenue, Suite 5230
Mobile, AL 36604
251-405-5121

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

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

© American College of Medical Genetics and Genomics, 2012 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957)



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 B⁰), 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
251-405-5121

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

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

© American College of Medical Genetics and Genomics, 2012 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957)



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 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.

Additional Information:

[Hemoglobin Disorders \(Grady Comprehensive Sickle Cell Center\)](#)

[Thalassemias](#)

[Genetics Home Reference](#)

[Sickle Cell Disease in Children and Adolescents: Diagnosis, Guidelines for Comprehensive Care, and Care Paths and Protocols for Management of Acute and Chronic Complications](#)

Referral (local, state, regional and national):

[Testing](#)

[Clinical Services](#)

[Thalassemia Care Center Directory](#)

[Thalassemia Treatment Centers Directory](#)

[Find Genetic Services](#)

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

© American College of Medical Genetics and Genomics, 2012 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957)

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
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).**

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

© American College of Medical Genetics, 2010 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957)



American College of Medical Genetics
Medical Genetics: Translating Genes Into Health®

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.

MaryEllen Whigham, R.N.

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 otoacoustic 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.

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

© American College of Medical Genetics and Genomics, 2012 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957)



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
1530 3rd Avenue South, Kaul 241
Birmingham, AL 35294
205-996-6983

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

© American College of Medical Genetics and Genomics, 2012 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957)



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
1530 3rd Avenue South, Kaul 241
Birmingham, AL 35294
205-996-6983

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

© American College of Medical Genetics and Genomics, 2012 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957)

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:

University of Alabama at Birmingham
Department of Genetics
KAUL 210B
1530 3rd Avenue South
Birmingham, AL 35294-0024
Phone: 205-996-6983
Fax: 205-975-6390

Alabama Newborn Screening Program
334-206-5556

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.

Recommended 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.

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

© American College of Medical Genetics and Genomics, 2012 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957)

CARNITINE SUPPLEMENTATION PROTOCOL

CARNITINE SUPPLEMENTATION FOR A 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 7-10 days** after carnitine supplementation is discontinued.
3. May need to initiate metabolic evaluation if abnormal levels persists when infant on full feeding.

Maria Descartes, M.D.
Professor Genetics, Pediatrics, and Neurology
University of Alabama at Birmingham

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:

University of Alabama at Birmingham
Department of Genetics
KAUL 210B
1530 3rd Avenue South
Birmingham, AL 35294-0024
Phone: 205-996-6983
Fax: 205-975-6390

Alabama Newborn Screening Program
1-866-928-6755

**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 tyrosine levels.*

Alert Value = tyrosine 445.75 or greater
Recommended 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)

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians are also advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

VITAMIN C PROTOCOL

PROTOCOL FOR VITAMIN C (ASORBIC 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:

1-866-928-6755

Maria Descartes, M.D.

Professor Genetics, Pediatrics, and Neurology

University of Alabama at Birmingham

Newborn Screening ACT Sheet Severe Combined Immunodeficiency (SCID) and Conditions Associated with T Cell Lymphopenia

Condition Description: Severe Combined Immunodeficiency (SCID) includes a group of rare but serious, and potentially fatal, inherited immune disorders in which T lymphocytes fail to develop and B lymphocytes are either absent or compromised. Impairment of both B and T cells leads to the term “combined.” Untreated patients develop life-threatening infections due to bacteria, viruses and fungi. The screening test for T cell receptor excision circles (TRECs), a byproduct of normal T cell development, identifies SCID as well as certain related conditions with low T cells. For example DiGeorge Syndrome with impaired thymus development may cause low T cells and low TRECs.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact the family to inform them of the newborn screening result. Point out that additional tests are required to determine whether the baby actually has an immune deficiency.
- Avoid exposing patient to illness pending completion of testing.
- If the infant has any signs of illness, refer to a pediatric hospital right away for evaluation, administration of immunoglobulin and antibiotics.
- If the infant requires transfusion of any blood product, be sure that only leukoreduced, irradiated products that are negative for cytomegalovirus (CMV) are used.
- DO NOT give live attenuated rotavirus vaccine, which could cause serious diarrhea in a baby with SCID. This vaccine is to be given only after an immunology specialist confirms that the baby’s immune system is normal.
- Consult with a specialist in pediatric immunodeficiency diseases (consult with a pediatric allergist/immunologist and/or infectious diseases specialist) who will assist with further testing.
- Provide the family with basic information about SCID and T cell lymphopenia (see resource list) and offer or arrange genetic counseling.
- Report confirmatory findings to newborn screening program.

Diagnostic Evaluation: Confirmatory studies include absolute lymphocyte counts, determination of the presence/absence of T and B lymphocytes and assessment of their function and molecular genetic testing. The specialist will:

- Order diagnostic tests, likely to include: CBC with differential and lymphocyte subset enumeration.
- Coordinate further testing, antibody levels, lymphocyte proliferation to mitogens, and molecular genetic testing as deemed appropriate.
- Offer disease/genetic counseling

Clinical Considerations: Immunoglobulin infusions and prophylactic antibiotics are essential to protect against infections. Diarrhea, failure to thrive, otitis media, serious infections (pneumonia, meningitis and/or sepsis), and opportunistic infections commonly occur starting by 2-4 months of life in individuals with SCID. Oral thrush may be seen. Bone marrow hematopoietic cell transplantation may be curative, and outcomes are best if this is performed within the first 3 months of life or before infections occur. Enzyme replacement and experimental gene therapy are available for some SCID genotypes. The most common form of SCID is XSCID (X-linked SCID), occurring only in males. However, autosomal recessive forms of SCID affect both males and females. Specific gene diagnosis is important for directing therapy as well as providing genetic counseling.

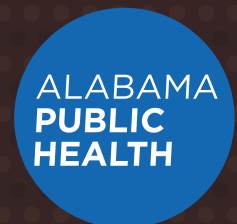
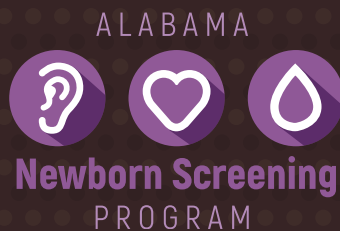
Local Referral Site:

Prescott Atkinson, M.D.
Professor and Director
UAB Division of Pediatric Allergy & Immunology
(205) 638-9072

Suthida Kankirawatana, M.D.
Assistant Professor
UAB Division of Pediatric Allergy & Immunology
(205) 638-9072

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians are also advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.





Alabama Department of Public Health Newborn Screening Program
www.alabamapublichealth.gov/newbornscreening • 1-866-928-6755