



Florida Newborn Screening Guidelines 2012



www.floridanewbornscreening.com

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Introduction

Newborn screening is a highly successful public health program that identifies rare metabolic, endocrine, enzymatic, and other genetic disorders and assures early management and follow-up for those affected. Infant blood is collected at or near the time of birth on a filter paper card and transferred to the State Newborn Screening Laboratory in Jacksonville for testing. The specimens are handled in strict accordance with the guidelines set forth by the Federal Clinical Laboratory Improvement Act 1988 CLIA), the Federal Centers for Medicare and Medicaid Services, and the Florida Agency for Healthcare Administration (AHCA). Parents or legal guardians of infants have the option of not having their child tested and can opt out of the program.

Purpose

Newborn Screening (NBS) is a population-based, preventive public health program that is provided in every state in the United States and in many countries throughout the world. The intent of the Florida Newborn Screening Program is to screen, identify, diagnose, and manage newborns at risk for selected disorders that, without detection and treatment, can lead to permanent developmental and physical damage or death. The goal of newborn screening is to facilitate prevention of developmental impairments, delayed physical growth, severe illness, and death through early detection and intervention. With appropriate and timely treatment, newborns identified through the Newborn Screening Program will have the opportunity to grow and reach their potential.

Mission

The mission and primary goal of the Florida Newborn Screening Program is to ensure that all newborns screened receive appropriate, high-quality laboratory and follow-up services.

Goals

The goals of the Newborn Screening Program are:

- To ensure that all newborns born in Florida are screened with results processed within five days of birth.
- To ensure that all affected infants receive appropriate confirmatory testing, counseling, and initiation of treatment as soon as possible.
- To provide physician consultation with other healthcare providers regarding treatment options and patient care recommendations.
- To maintain a system of sound fiscal management to support the program.
- To provide an educational information program for the various healthcare providers that serve families.

The Florida Department of Health Newborn Screening Program is a comprehensive system with the following partners: Bureau of Laboratories Newborn Screening Laboratory in Jacksonville, Children Medical Services Newborn Screening Follow-up Program in Tallahassee and contracted referral centers throughout the State of Florida for genetic disorders, endocrine disorders, hemoglobinopathy disorders and cystic fibrosis. In addition, the Genetics and

Newborn Screening Advisory Council serves as an advisory body to the Florida Department of Health.

Limitations of Screening Tests

The purpose of newborn screening programs is to identify infants at risk using laboratory analyses and in need of more definitive testing or to identify carrier status for some genetic conditions. As with any laboratory screening test, both false positive and false negative results are possible. Screening test results alone are insufficient information upon which to base diagnoses or long-term treatment. It is imperative that healthcare providers remain watchful for any signs or symptoms of these disorders in their patients and follow up as needed. Confirmatory testing is performed to either verify or rule out a disorder.

Program History

1965 – The Newborn Screening Program began in Florida with the passage of Section 383.14 of the Florida Statutes (F.S.). This required the Florida Board of Health to promote the testing of all newborns for Phenylketonuria. At the time, 20% of testing was performed by hospitals and 80% was performed by the Department of Health Bureau of Laboratories in Jacksonville and Miami.

1978 - Congenital Hypothyroidism, Maple Syrup Urine Disease, and Galactosemia were added to the Newborn Screening panel. An Infant Screening Advisory Council (later renamed the Genetics and Newborn Screening Advisory Council) was established and regulations were set up with regard to follow-up, diagnosis and treatment of infants with abnormal results. In 1979/1980, the Bureau of Laboratories-Jacksonville was designated as the only site for testing all newborn screening specimens.

1984 - The program was expanded to identify infants at risk for hearing impairment and those with birth defects, with funds appropriated to establish a confidential computer registry for birth defects.

January 1985 - Maple Syrup Urine Disease was deleted from testing due to the lack of any detected case in 500,000 births but was added back to the MSMS panel in January 2006.

August 1988 - Testing for hemoglobinopathies was added to the Newborn Screening Program.

1993 - Funding of Newborn Screening services changed from general revenue to fees collected from hospitals and birthing centers. A fee of \$20.00 per live birth charged to facilities with over 60 births and up to 3,000 births was established through Florida Statutes 383.14.

March 1995 - Identifying infants at risk for hearing impairments and birth defects was discontinued

April 1995 - Testing for Congenital Adrenal Hyperplasia was added to the newborn screening panel.

October 1, 2000 - Hearing screening was mandated per Florida Statutes 383.145.

2002 - The Newborn Screening Task Force was created by House Bill 817 to evaluate the newborn screening program and make recommendations for improvement of the current infant screening program and consider an expansion to include those disorders recommended by the March of Dimes and the American College of Medical Genetics.

July 1, 2004 - The newborn screening fee was reduced to \$15 per live birth and the exemption and caps were removed. The Department of Health was given the authority to bill third party payers for newborn screening tests. The State Public Laboratory was given the authority to release the newborn screening results directly to the newborn's primary care physician or through Children's Medical Services.

October 1, 2005 - Biotinidase Deficiency was added to the newborn screening panel (for a total of nine disorders).

January 9, 2006 – Implementing tandem mass spectrometry added 25 new disorders to the panel including amino acid disorders, organic acid disorders, and fatty acid oxidation disorders (for a total of 34 disorders – including hearing).

September 17, 2007 - Cystic Fibrosis was added to the screening panel. Florida screens for a total of 35 disorders, which satisfied the recommendations by both the March of Dimes and the American College of Medical Genetics.

January 5, 2009 – Physicians were able to register and obtain newborn screening results through the Florida Newborn Screening Results (FNSR) website – <https://www.FNSR.net>. This online system allows access to newborn screening results for babies whose specimens were tested by the lab in the last six months.

January 28, 2011 – The Florida Department of Health Genetics and Newborn Screening Advisory Council recommended adding Severe Combined Immunodeficiency Disease (SCID) as the next additional disorder to the newborn screening panel.

January 20, 2012 – The Florida Department of Health Genetics and Newborn Screening Advisory Council recommended working with the Cardiac Subcommittee of Children’s Medical Services Advisory Council for recommendation of implementing Critical Congenital Heart Disease (CCHD) to the newborn screening panel.

Guidelines

Guidelines for this program were established in 1981 and expanded in 1984, 1992, 1999, and 2012. These guidelines set the standards for the Florida Newborn Screening Program.

Legal Authority

Sections 383.14 and 383.145, Florida Statutes, and Chapter 64C-7, Florida Administrative Code.

Registry

Newborn Screening law states that the Department of Health shall maintain a confidential registry of cases, including the information of importance for the purpose of follow-up services to prevent mental retardation, to correct or ameliorate physical handicaps, and for epidemiologic studies, if indicated.

Confidentiality

Newborn Screening is an activity described in its capacity as a public health authority as defined by the HIPAA Standards for Privacy of Individually Identifiable Health Information,

Final Rule (Privacy Rule). Pursuant to 45 CFR 164.512(b) of the Privacy Rule, covered entities such as primary care physicians, county health departments and other medical establishments may disclose, without individual authorization, protected health information (PHI) to public health authorities. **Public health entities are authorized by law to collect or receive such information for the purpose of preventing or controlling disease, injury, or disability including, but not limited to, the reporting of disease, injury, vital events such as birth or death, and for the purpose of conducting public health surveillance, public health investigations, and public health intervention.** Demographic information which identifies the patient and family is covered by the Department's rules on privacy and confidentiality. Questions regarding confidentiality may be answered by contacting the Newborn Screening Program at 850-245-4201 or the Bureau of Laboratories-Jacksonville at 904-791-1500.

Parental Refusal

Per Florida Statute 383.14, all newborns must have a specimen collected before discharge, unless a parent or guardian objects to the screening test. A written statement of such objection must be presented to the physician or other person whose duty it is to administer and report tests and screenings. A copy of the written objection should be filed in the newborn's medical record and be available upon request. Please do not submit these documents to the newborn screening laboratory or the follow-up program unless told to do so.

If newborn screening or hearing screening is refused, a specimen card should be sent to the Bureau of Laboratories-Jacksonville and the "Refused" bubble on the specimen card should be filled in along with the demographic information. Even with refused specimens, all fields should be completed.

Genetics and Newborn Screening Advisory Council

The Genetics and Newborn Screening Advisory Council serves as an advisory body to the Department of Health regarding medical and procedural aspects of the Newborn Screening Program. Recommendations for program changes are made by the Advisory Council to the Deputy Secretary for Children Medical Services.

Advisory Council members are appointed by the State Surgeon General of the Department of Health and are comprised of 15 members including the following:

- four representatives from four medical schools in the state
- the State Surgeon General or his or her designee
- the Children's Medical Services Deputy Secretary or designee
- the Agency for Persons with Disabilities Program Office
- three practicing pediatricians, at least one of whom must be a pediatric hematologist
- one representative from the Florida Hospital Association
- one individual with experience in newborn screening programs (e.g., March of Dimes)
- one individual representing audiologists
- two consumers

Members serve for a period of four years and terms are staggered, with possible reappointments. Meetings are held semi-annually or more frequently if necessary.

It is the purpose of the council to advise the Department about:

- conditions that should be included in the Florida Newborn Screening Program
- procedures for collecting and transmitting specimens and recording such results
- methods by which newborn screening can more effectively provide services to the children of Florida

Program Responsibilities

Birth Facilities:

- Designate a newborn screening coordinator or contact person to ensure efficient communication with the newborn screening laboratory in Jacksonville and the newborn screening follow-up program in Tallahassee. This person must track all newborns in their facility to ensure that all newborns obtain a valid newborn screening specimen.
- Order dried bloodspot specimen cards a month in advance before internal supplies run low.
- Provide hearing screening information on the dried blood specimen cards provided by the Bureau of Laboratories (or through eReports when accessible).
- Provide verbal and written information about the newborn screening program to the parent or guardian of all infants born in Florida.
- Initiate a birth record.
- **Legibly** complete **all** fields on the specimen card or the dried blood spots specimen card with accurate insurance information. Keep a copy for their records and remove all excess paper or debris before submitting the specimen card.
- Collect high quality and sufficient amounts of infant's blood specimen on a card which is not expired (see hour glass symbol).
- Unsatisfactory specimens and specimens over 14 days old will not be tested by the laboratory under any circumstances thus delaying a potentially fatal diagnosis. If a specimen is deemed unsatisfactory by the laboratory, it is the submitting entity's responsibility to ensure that a satisfactory specimen is collected per the lab report instructions to the submitting entity and resubmitted as soon as possible following first specimen collection guidelines.
- Document and track the collection of the 5 blood spots, the transmittal of the cards, receipt of the dried blood specimen card by the State Lab and the final laboratory results should be placed in the child's medical record.
- Provide the parent/guardian with written notification that a specimen must be collected by five days of age if the infant is discharged before 24 hours of age and/or before being on a protein diet for 24 hours. The clinical significance of certain lab results cannot be determined because the infant was less than 24 hours of age at the time of collection or

the infant was on protein feed less than 24 hours before collection of the specimen. Another specimen must be obtained immediately.

- It is the responsibility of the birth facility to follow up and ensure that a valid specimen is obtained. Information regarding the location where a repeat specimen is collected must be given to the parent prior to discharge or when the parent calls for instruction if the specimen is unsatisfactory or has borderline results.
- Provide the Newborn Screening Program with information on repeat screening tests and demographics information when contacted. Repeat specimens should include complete demographic information including mother's social security number, physician, birth facility, etc., and other identifying information.
- Forward a copy of the laboratory results and any follow-up information to the primary care provider if possible. (Primary care physicians can access the online system to check for newborn screening results at <https://www.fnsr.net>)
- Obtain a signed waiver from the parent that must be placed in the newborn's medical records if the parent/guardian refuses the newborn screening. These records may be reviewed upon request by the Florida Newborn Screening Program.
- If an infant is transferred to another facility within 24 hours of birth, document and inform the new facility that a blood specimen was not collected. The admitting facility will be responsible for obtaining a specimen.
- If copies of the original newborn screening laboratory report are needed by the submitting entity, submit the appropriate request form to the Newborn Screening Laboratory in Jacksonville via fax or mail. Obtain request forms through the laboratory. Alternatively, hospitals are encouraged to register in the Florida Newborn Screening Results (FNSR) web-based application at <https://www.fnsr.net> to access newborn screening results online as a laboratory user.
- It is the submitter's responsibility to designate a contact person and provide current contact information when requesting test results from the Newborn Screening Laboratory to avoid delays in processing.

Department of Health Bureau of Laboratories Newborn Screening Laboratory:

- Perform tests and report the results to the submitting entity using established procedures such as US Postal Service or Auto-Fax.
- Generate reports regarding screening tests, information errors on specimen cards and other information as needed.
- Advise CMS of laboratory-related problems and of recurring information errors on dried blood specimen cards.
- Respond to inquiries from submitting entities concerning laboratory results.
- Testing is performed 6 days per week, Monday-Saturday.

- In the event of activation of an emergency (hurricane etc), dried blood specimen cards are directed to the designated back-up laboratory for immediate processing as determined by the Bureau of Laboratories.
- Collect the newborn screening fee (\$15 per live birth) from the birthing facilities and bill third party payers for the newborn screening tests.
- Be the custodian for the DOH policy regarding specimen retention and storage.

-

Children's Medical Services

Newborn Screening Follow-Up Program:

- Notify Genetic, Endocrine, Hemoglobinopathy, Cystic Fibrosis and applicable Referral Centers of any infant identified with presumptive positive screening results on the day the result is released and the referral is created in the newborn screening data system except for Cystic Fibrosis and Sickle Cell referrals. These will **not** be referred on Saturdays, Sundays or Holidays.
- Monitor and track newborns with borderline and presumptive positive screening results to ensure repeat screenings, status confirmations, and request confirmatory testing results from the Referral Centers.
- Contact the family's primary care provider regarding borderline results (or family, if physician is unknown).
- Send sickle cell trait, unsatisfactory specimen, hearing test fails, and other letters generated by the data system to parents/guardians and physicians as appropriate.
- Respond to inquiries from physicians in obtaining results and assist in utilizing that information to avoid unnecessary repeat testing.
- Update case status as case reports are received and report to the national database as needed.
- Call parents regarding failed hearing screens to facilitate diagnostic hearing testing
- Notify hospital administration of recurring inaccurate or incomplete information on dried blood specimen cards, i.e., delay in submitting specimens, copies not being kept, and missing demographics and billing information.
- Develop, obtain, and provide educational materials for hospitals, healthcare professionals and parents or guardians about the newborn screening program and other pertinent information.
- Develop and provide reports to the public and users regarding the newborn screening program.

Specialty Referral Centers:

- Receive and accept all referrals from the Newborn Screening Follow-Up Program for newborns with presumptive positive screening results or newborns with multiple borderline results.
- Contact the primary care physician (or parent if physician is unknown) to coordinate immediate evaluation and/or treatment and confirmatory testing.
- Contact the parent.
 - If no address is available, contact the hospital/birth facility for assistance with locating the family.
 - If all contact attempts fail, contact the Newborn Screening Follow-Up Program for assistance.
 - If no phone number is available, local law enforcement may be sent to the address on record for assistance. Call the local police or sheriff and ask for the Watch Commander or equivalent to assist.
- Arrange for confirmatory testing or other medically necessary actions for newborns with presumptive positive screening results or multiple borderline results.
- Provide appropriate counseling and education to families regarding the course of treatment and diagnosis.
- Report any changes in address, location or contact information to CMS.
- Provide the Newborn Screening Follow-Up Program with a completed case report within ten (10) days of confirmation, including the name and address of the primary care physician and a copy of the laboratory confirmatory test results if required by contract.
- Contact the CMS Area Office for assistance when infants have no insurance to cover confirmatory testing.
- Refer all positively diagnosed newborns to the CMS Area Office for care coordination. CMS will determine financial eligibility.

Primary Care Providers:

- Ensure that all infants in their care have a valid newborn screening, including hearing, prior to leaving the birthing facility.
- Obtain the newborn screening report from the birth facility or obtain the newborn screening results through the Department of Health web based application -- Florida Newborn Screening Results (FNSR) <https://www.fnsr.net>
- Ensure all necessary repeat specimens for unsatisfactory specimens or borderline results have been obtained. **Do not collect a repeat specimen if the first specimen is valid and the newborn screening results are normal.**

- Provide updated information regarding changes in address, phone number, or contact person's email address in order to prevent erroneous delivery of newborn screening results.
- Assist the referral centers and the CMS Newborn Screening Follow-Up Program to locate a newborn and/or provide continuing care.

CMS Area Offices:

- Provide assistance to families of infants with presumptive positive newborn screening results if they have no Medicaid or private insurance for confirmatory testing funding.
- Babies identified with one of the newborn screening disorders are medically eligible for CMS services but must meet the financial eligibility requirements in order to receive care coordination services.

Completing the Specimen Card

It is important to fill out the specimen card completely, accurately, and legibly. All fields are required. It is also important that the submitting entities keep a copy of the card for their records. The information on the card provides information about the specimen collection that is necessary to interpret test results accurately. In addition, accurate and complete information allows the Newborn Screening Program to link additional test results with initial reports and to locate newborns in need of repeat testing or follow-up services. Linked specimens may change the actions necessary if the previous specimen was also borderline.

The fields on the specimen card are self explanatory and full completion is required; however, there are a few fields that are critical and could result in delays or cause erroneous test results that could be detrimental to a newborn's health if not accurately and/or fully completed.

Birth, feeding, and collection times: All times should be uniformly reported in military time.

Example:

2:16 a.m. = **0216**

1:15 p.m. = **1315**

It is important to have the correct date and time because some testing methodologies are impacted by the date and time of birth, feeding time and collection time.

Neonatal Intensive Care Unit (NICU) status: Indicate the location of a newborn if admitted to the Level II or Level III intensive care units as NICU.

Collection Facility: Write out the full name and address of the collection facility. *Do not use abbreviations.* Results of a specimen without a collection facility will be sent to the birth facility.

Address: Report the most recent address on file, ensuring that zip codes, cities and unit numbers are reported. It is vital that unit numbers (apartment, lot, etc.) be included. Always use United State Postal Service (USPS) approved abbreviations. Without accurate information, the USPS will not be able to deliver important information if a letter is sent to the parent regarding the test results.

Insurance: Medicaid or private insurance information should include the name of the insurance company, group number and the policy number. All Medicaid or Private Insurance fields should be completed, leaving no blanks.

Transfusion date: *If a newborn has not been transfused, do not report a date in this field.* Transfusions may require repeat testing and can invalidate certain test results.

Infant's name: If the newborn's name is known at the time of collection, it should be reported. If not known, the mother's last (preferably mother's name to avoid name change issues) name should be used and the first name listed as "baby girl" or "baby boy."

Multiple birth order: If a baby is one of a set of multiples (twins, triplets, etc.), it is imperative the birth order be reported on every specimen for every infant. Names alone are not enough to link multiple specimens in the Newborn Screening data system. **Do not list a birth order for single births.**

Medical Record Number (MR#): The newborn's medical record number must be accurately reported. Errors in the medical record numbers for the same patient will force the laboratory to declare the specimen as "Unsatisfactory due to information mismatch" further delaying the testing while waiting on a new specimen to be submitted.

Adoption: Fill in the bubble only if the baby is being adopted. The name and contact information of the legal guardian, adoption agency, or attorney at the time of collection should be listed instead of the biological mother's information. This avoids the issue of contacting the birth mother instead of the current parent/guardian. **In the event of abnormal results, the Newborn Screening Follow-Up Program relies on this contact information to provide prompt follow-up services to the correct people who know where the child is located.**

For infants in Foster Care: List the name of the agency/case worker and telephone number in the field for "mother" if the infant will not be going home with the mother. (If using the Department of Children and Families (DCF) case workers name, please add DCF beside the last name. For example, "Smith, DCF".)

Weeks of Gestation: List the infant's gestational age, NOT the current age.

Linking of specimens depends on the accuracy of the following fields on the specimen card:

Mother Last Name
Baby Last Name
Mother First Name
Baby DOB
Mother SSN
Medical Record Number
Mother Address
Birth Hospital

These fields need to match exactly in order for initial and subsequent specimens to be linked automatically in the data system, otherwise there might be a delay in notification of a presumptive positive result that has been converted due to two borderline specimens.

Please Remember...

- **Do not use abbreviations, hospital codes, or symbols.**
- **Demographic information should be taken from the medical record or directly from the parent/guardian.**
- **Complete, accurate, and legible patient information is critical for locating infants and rapid follow-up in the event that the newborn has abnormal results.**
- **Complete ALL sections of the specimen card.**

The Newborn Screening Follow-Up Program is available for questions or concerns regarding dried blood specimen card completion.

Call toll-free **(866) 804-9166** or **(850) 245-4201** for more information.

The Newborn Screening Laboratory customer service staff is also available to assist with this information at **(904) 791-1645**.

Specimen cards may be ordered via fax or mail using the form located at the following website:
http://www.doh.state.fl.us/Lab/PDF_Files/OrderForm_for_DH677.pdf

E-reports

The Florida Department of Health is currently developing a system that links data from the Vital Statistics Electronic Birth Registry System to the Newborn Screening Program's data system. The new system, called E-reports, will enable faster and more accurate data entry and will also allow certain providers to enter screening results remotely. E-reports is an innovative step towards more efficient and precise administration of public health data. It is projected for completion in the summer of 2012.

Newborn Screening Specimen Card

SUBMITTER OR PARENT COPY

REF 10534749 Rev. AS YYY-MM LOT WXXX XXXXXX IVD

1) Do not touch sample area

2) Do not use if damaged

IF NOT SUBMITTING BLOOD CHECK ONE

INFANT'S INFORMATION REFUSED INFORMATION ONLY E-VITALS COMPLETED ADOPTION DECEASED

Infant's Last Name _____ Birth _____ Time _____ (Military Form) _____

Hospital of Birth _____ DD _____ YY _____

Infant's First Name _____ Birth Weight (gms) _____ Weight (gms at Collection) _____

Infant's Medical Record Number _____

Infant's Gender: M F

WEEKS OF GESTATION _____

SELECT ALL THAT APPLY: WHITE BLACK AMERICAN INDIAN HISPANIC ASIAN/PACIFIC ISLANDER OTHER _____

WEEKS OF GESTATION _____

NICU _____

Transfused Date (mm/dd/yy) _____ Time (Military Form) _____

INSURANCE INFORMATION

Insured's Name (Last, First, Middle Initial) _____

Name of Insurance Co. _____

Insurance Group ID# _____

Insurance ID# _____

PRIVATE MEDICAID SELF-PAY

Insured SSN _____ Mother's Medicaid Number _____

HEARING SCREENING DARKEN CIRCLES THAT APPLY

RIGHT EAR PASS REFER LEFT EAR PASS REFER

OAE ABR OAE ABR

HEARING RISK STATUS: (DARKEN CIRCLES THAT APPLY)

FAMILY HISTORY PPHN ECWO BIRTH WEIGHT <1500 GRAMS

EXCHANGE TRANSFUSION FOR HYPERBILIRUBINEMIA (DARKEN ALL CIRCLES THAT APPLY)

PARENT/GUARDIAN REFUSED MISSED BIRTH DEFECT

BABY EXPIRED FACILITY TRANSFER NOT YET SCREENED (NICU)

HEARING SCREENING DARKEN CIRCLES THAT APPLY

HEARING SCREENING DATE: M M D D Y Y

STATE LAB USE ONLY

STATE LAB USE ONLY

WH181911 903 LOT WXXX XXXXXX

STATE LAB USE ONLY

STATE LAB USE ONLY

MOTHER'S INFORMATION

Mother's Last Name _____ Time (Military Form) _____

Mother's Address (include Apartment #) _____ City _____ State _____ Zip Code _____

Mother's Social Security Number _____ Mother's or Contractor's Telephone Number _____

PHYSICIAN'S INFORMATION

Physician's Last Name _____ First Name _____

Physician's Telephone No. _____

SUBMITTER INFORMATION

Name of Collection Facility _____ Laboratory ID# _____

Address _____ City _____ State _____ Zip Code _____

STATE OF FLORIDA-DEPT. OF HEALTH-BUREAU OF LABORATORIES 1217 PEARL STREET JACKSONVILLE FL 32202 (904) 791-1644 (904) 791-6447

ALL SPECIMENS COLLECTED ON INFANTS <24 HRS. OF AGE / <24 HRS. ON PROTEIN MUST BE REPEATED AND SENT TO THE STATE BUREAU OF LABORATORIES FOR RE-EVALUATION. THIS CARD IS NOT VALID FOR USE ON INFANTS COLLECTED ON THE NEWBORN SCREENING SPECIMEN COLLECTION CARD, DH 677, MM-YY. REPLACES ALL PREVIOUS EDITIONS. CONFORMS TO CLSI STANDARDS.

(Note: Specimens cards are frequently updated. Check the expiration date and lot number at the DOH website to ensure that you are not using an expired card.)

Specimen Collection Protocols

Newborn Screening Coordinators

Each birthing facility should designate one person and an alternate to be responsible for proper ordering, record keeping, collection of specimens, maintaining result reports, and submission of requests. These individuals should be knowledgeable of the entire collection process. Designated persons should inspect and approve all specimens prior to mailing to ensure that all information is included on the form and that a satisfactory sample is being submitted to the Bureau of Laboratories-Jacksonville. The Newborn Screening Coordinator should ensure that all specimen circles are properly filled to provide adequate samples for testing.

Normal, Term, or Well-Baby Nursery

VALID SPECIMEN: A specimen that is collected after the newborn is 24 hours of age and has been on protein feed for at least 24 hours. It is imperative to collect as close to this time as possible and submit the specimen as soon as possible to prevent delay in testing.

Specimens not meeting the requirements of a valid specimen will require a repeat specimen. The birthing facility must provide the newborn's parent or guardian with instructions to obtain a repeat specimen within **five** days of age. The family must be given the option to return to the birthing facility or arrangements must be made to ensure that a repeat specimen can be obtained at the newborn's physician's office or local County Health Department. It is the birthing facility's responsibility to ensure that the repeat specimen is obtained after discharge if the newborn is discharged early and a valid specimen is not obtained prior to discharge.

Transfusions

It is preferred that a specimen is collected prior to the first transfusion. If a baby was transfused prior to the first specimen, a repeat specimen must be collected three to four months after the last transfusion. Transfusions prior to blood collection can produce a false normal result or false partial test, or may invalidate the hemoglobinopathies and the galactosemia screening results.

Transported or Transferred Newborns

The birthing facility is not required to collect a blood specimen if the newborn is transferred to another facility *within 24 hours* of birth. The birthing facility is required to document and inform the new facility that a specimen was not collected and the hearing screening was not provided. For newborns transferred *after 24 hours* of birth, the specimen should be collected by the birthing facility and notification given to the facility to which the newborn is being transferred regarding the hearing test results if any. Regardless of time/transfer, the facility where the infant was born should be listed as the "hospital of birth" on the specimen card.

Home Births

Persons in attendance of a home birth are required to arrange for a specimen to be collected after 24 hours of age and 24 hours on protein feeding. Repeat specimens may be collected at a physician's office, county health department, clinic, midwife facility or hospital. The "Birth Facility" field on the specimen card should be reported as "Home."

Repeat Specimens

The most common reasons a repeat specimen may be required are:

- The initial specimen was collected too early - prior to 24 hours of age and/or 24 hours on protein feed.
- The initial/previous specimen was unsatisfactory for testing.
- The initial/previous specimen had borderline results and the recommendation indicated that a repeat must be obtained.
- The newborn was transfused prior to the collection of the initial specimen.

Critically ill and Premature Infants (NICU)

All newborns requiring neonatal intensive care services must have a screening specimen obtained (regardless of age and feeding status):

- Collect the first newborn screening upon admission to the Neonatal Intensive Care Unit (NICU) preferably prior to first transfusion and regardless of age before any other treatments are done (except respiratory).
- Collect the second newborn screening specimen at 48-72 hours of life, for infants initially tested prior to 24 hours of age at first screen, or less than 2000 grams at birth, or any infant with an abnormal first screen.
- Collect the third newborn screening specimen at 28 days of life, or discharge, whichever comes first for newborns less than 2000 grams at birth. For infants weighing > 2000 grams this third newborn screening is at the discretion of the Primary Care Provider.

Summary for Specimen Collection Timing

Infant Status	Time of Collection
Normal, Healthy	As soon as possible after 24 hours of age and 24 hours on protein feeding
Transfused	Prior to first transfusion OR 3 - 4 months after last transfusion
Premature, sick or extended stay	<ul style="list-style-type: none"> Collect the first newborn screening upon admission to the Neonatal Intensive Care Unit (NICU) preferably prior to first transfusion and regardless of age before any other treatments are done (except respiratory). Collect the second newborn screening specimen at 48-72 hours of life, for infants initially tested prior to 24 hours of age at first screen, or less than 2000 grams at birth, or any infant with an abnormal first screen. Collect the third newborn screening specimen at 28 days of life, or discharge, whichever comes first for newborns less than 2000 grams at birth. For infants weighing > 2000 grams this third newborn screening is at the discretion of the Primary Care Provider.

Clinical Signs or Family History

A number of clinical situations will modify the usual approach of obtaining a newborn screening specimen. The following are suggested guidelines for particular situations that may arise.

Infants who exhibit clinical signs and symptoms:

The newborn screening test, like any laboratory screening test, may have false positives and false negatives. If signs and symptoms of one of the newborn screening conditions are clinically evident, the physician should immediately consult referral center specialists for diagnostic/confirmatory testing and evaluation. It may be necessary to treat as if the infant has the condition before the newborn screening results are available.

Infants with affected siblings or close relatives:

Many of the conditions tested by newborn screening are genetic, so it is possible that multiple members of a family may be affected and known to the family. Physicians are also encouraged to contact the Newborn Screening Program if it is known whether infants are born to families with affected siblings or close relatives.

Prenatal diagnosis:

Prenatal diagnosis is available for some of these conditions; if the pregnant woman chooses to have testing prior to birth. This is usually done if a previous baby was born with the disorder.

For any infant with a positive family history, providers should contact appropriate consultant specialists, ideally before birth, or immediately after birth, to determine the proper diagnostic tests, timing, and the best strategy for patient care.

Unsatisfactory Specimens

According to Florida Statutes, if any specimen collected by a submitting entity is deemed unsatisfactory, or inconclusive, it is the submitting entity's responsibility to resubmit an additional specimen. A reasonable attempt must be made to locate these infants and document the steps taken.

A **reasonable attempt** is defined as a documented effort to locate the infant which includes:

- (1) a telephone call and/or letter to family;
- (2) notification to family by certified mail with return receipt; and
- (3) notification to physician of record (notifying the primary care physician does not absolve the birthing facility from their responsibility).

It is imperative that a repeat specimen must be obtained as soon as possible upon notification of unsatisfactory status.

The laboratory codes for unsatisfactory specimens as reported on the laboratory report are listed below.

Codes and explanations for unsatisfactory specimens

A	specimen is contaminated
B	specimen is quantity not sufficient (QNS) and damaged
C	QNS and separated
D	specimen is damaged
E	incomplete saturation
F	specimen is damaged and separated
G	specimen is QNS and has repetitive spots
H	supersaturated / layered
I	this specimen has insufficient data
J	information mismatch
K	specimen is damaged and has repetitive spots
L	specimen is QNS and has areas of clotted blood
M	specimen has areas of clotted blood
N	no blood spots
O	specimen is damaged and has areas of clotted blood
P	diluted / discolored
Q	specimen is quantity not sufficient (QNS)
R	specimen has overlapping or repetitive spots
S	specimen is separated with rings around blood spots
T	specimen is too old for valid results. Specimen was received by the laboratory more than 14 days after collection date.
U	specimen has repetitive spotting and areas of clotted blood
V	specimen is damaged, has repetitive spots and areas of clotted blood
W	specimen not completely dry when received at the laboratory
X	expired specimen card
Y	specimen has repetitive spots and separated
Z	other (please telephone the laboratory at 904-791-1647)

Whatman Neonatal Screening Charts

See charts on next three pages for proper specimen collection and handling procedures provided by Whatman.

Please note that on item 10 the poster shows a first class envelope, we encourage all submitting entities to send specimens priority overnight since the specimens need to be received in the lab by five days from collection. Heat may cause specimen to deteriorate and invalidate results.

Neonatal Screening

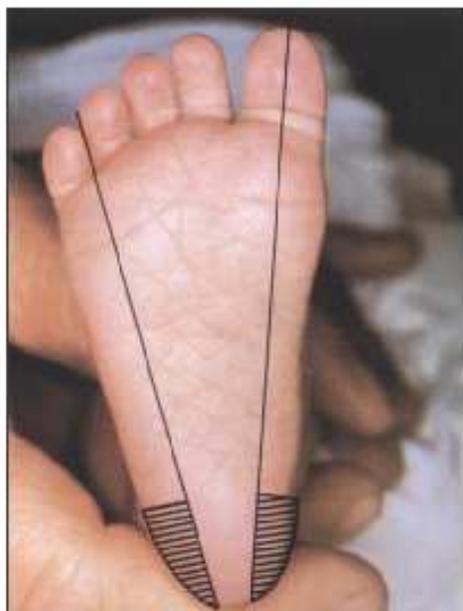
Blood Specimen Collection and Handling Procedure

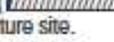


- 1 Equipment: sterile lancet with tip approximately 2.0 mm – sterile alcohol prep, sterile gauze pads, soft cloth, blood collection form, gloves.



- 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.

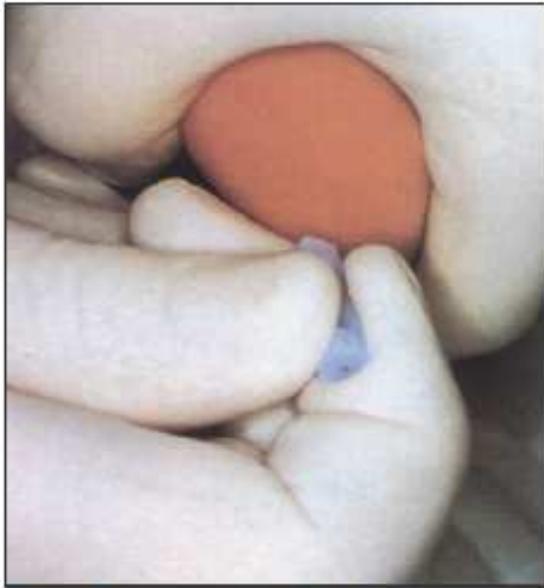


- 4 Warm site with soft cloth, moistened with warmwater up to 41°C, for three to five minutes.



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





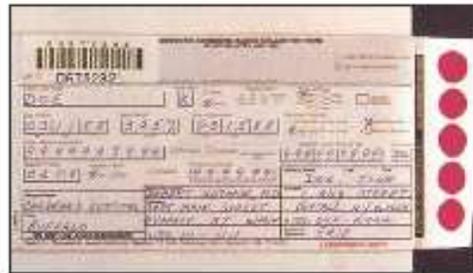
6 Puncture heel. Wipe away first blood drop with sterile gauze pad. Allow another **LARGE** blood drop to form.



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



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



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



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

Information provided by The New York State Department of Health.

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Simple Spot Check

Whatman®

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.

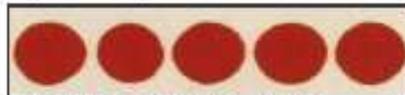
Invalid specimen:



1. Specimen quantity insufficient for testing.



2. Specimen appears scratched or abraded.



3. Specimen not dry before mailing.



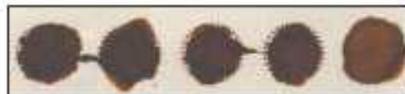
4. Specimen appears supersaturated.



5. Specimen appears diluted, discolored or contaminated.



6. Specimen exhibits serum rings.



7. Specimen appears clotted or layered.



8. No blood.

Possible causes:

- 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.
- Touching filter paper before or after blood specimen collection with gloved or ungloved hands, hand lotion, etc.
- Allowing filter paper to come in contact with gloved or ungloved hands or substances such as hand lotion or powder, either before or after blood specimen collection.
- Applying blood with a capillary tube or other device.
- Mailing specimen before drying for a minimum of four hours.
- Applying excess blood to filter paper, usually with a device.
- Applying blood to both sides of filter paper.
- Squeezing or "milking" of area surrounding the puncture site.
- Allowing filter paper to come in 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.
- Not wiping alcohol from puncture site before making skin puncture.
- Allowing filter paper to come in contact with alcohol, hand lotion, etc.
- Squeezing area surrounding puncture site excessively.
- Drying specimen improperly.
- Applying blood to filter paper with a capillary tube.
- Touching the same circle on filter paper to blood drop several times.
- Filling circle on both sides of filter paper.
- Failure to obtain blood specimen.

Information provided by The New York State Department of Health.

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Most common causes for unsatisfactory specimens

Unsatisfactory Code/Explanation	Possible Causes
Quantity of blood not sufficient for testing (QNS)	Filter paper circles incompletely filled or not saturated; not all circles filled.
Blood spots appear scratched or abraded	Blood applied with needle or capillary tube or other means (filter paper has been damaged or torn by device).
Blood spots wet	Specimen not properly dried for four hours before mailing.
Blood spots appear supersaturated	Excess blood applied. Blood applied to both sides of filter paper.
Blood spots appear diluted, discolored, or contaminated	Puncture site squeezed or “milked.” Exposure of blood spots to direct heat. Contamination of filter paper before or after specimen collection by gloved or ungloved hands, or by substances such as alcohol, formula, water, powder, antiseptic solutions, or hand lotion. Contamination during transit.
Blood spots exhibit “serum rings”	Alcohol not wiped off puncture site before skin puncture is made. Filter paper has come into contact with alcohol, water, hand lotion, etc. Puncture site squeezed excessively. Specimen dried improperly, possibly hung vertically. Blood applied to the filter paper with capillary tube.
Specimen has overlapping or repetitive spots	Same filter paper circle touched to a blood drop several times. Circle filled from both sides of the filter paper.
Blood will not elute from the blotter paper	Blood specimen has been heat-fixed. Blood specimen is too old (more than two weeks between collection and receipt by the screening laboratory).
No blood	Failure to obtain blood specimen.

Proper Specimen Handling

Do's...

- Do dry the specimen card in a horizontal position.
- Do dry the blood spots thoroughly at room temperature for at least 4 hours.
- Do keep specimen cards away from direct heat or sunlight.
- Do double-check that patient information section has been completely filled out before mailing.
- Do alternate the forms so that the dried blood spots do not come into contact with each other if mailing more than one specimen card in an envelope.
- Do check that a return address is present on the mailing envelope.
- Do mail specimen cards by overnight courier as soon as possible after it is thoroughly dry, within 24 hours of specimen collection.
- Do keep a list of specimens that were sent so that there is documentation.

Don'ts...

- Don't allow the bloodspots to come into contact with any other surface while drying.
- Don't refrigerate the specimen cards.
- Don't place the specimen cards in the envelope until completely dry.
- Don't place the specimen cards in outdoor mailboxes located on the street.
- Don't transport the specimen cards in plastic bags.
- Don't batch the specimen cards beyond the same day.
- Don't dry the specimen cards vertically.
- Don't submit copies of the card with the original specimen cards.
- Don't staple or tape the specimen cards together.

Specimen Transport Protocol

If lives are to be saved, it is critical that the Florida Newborn Screening Laboratory receives the newborn screening specimen card as soon as possible after collection. Ideally, specimens should be mailed or transported ***as soon as they are properly dried (four hours) and no later than 24 hours after collection.*** Some of the disorders that are included in Florida's screening panel can cause death or permanent, irreversible damage in the first days of life. To prevent this, diagnosis and treatment must occur rapidly. **Specimen cards should be sent by overnight courier or express mail service.** Significant degradation of hemoglobin and some analytes can occur in specimens older than one week and those exposed to high heat and humidity. Specimen cards should never be left in the heat. Please see storage and shipping recommendation on the back of the specimen card. If the specimen is received more than fourteen days after collection date, the specimen will not be tested.

The ideal shipping method for specimens is overnight courier or priority shipping. Specimens mailed out on Friday ***may be marked for Saturday delivery***, as the Bureau of Laboratories-Jacksonville is open to receive deliveries on Saturday and the Newborn Screening Laboratory processes dried blood specimen cards the same day. Taking advantage of Saturday delivery means faster turnaround time and being able to identify babies at risk for the disorders earlier. For some of the disorders, days - even hours - can make a difference.

In case of emergencies (e.g. hurricane, etc.), the Florida Department of Health will notify birthing facilities of alternative mailing addresses or procedures.

Specimen Retention and Storage Policy

Newborn screening is a highly successful public health program that identifies rare metabolic, endocrine, enzymatic, and other genetic disorders and assures early management and follow-up for those affected. Infant blood is collected at or near the time of birth on a filter paper card and submitted to the State Newborn Screening Laboratory in Jacksonville, Florida for testing. After testing, the DBS card is stored for a period of six months pending requests for follow up or additional testing to confirm screening test results, for use in internal quality assurance, and for instrumentation and/or methodology validation studies. The specimens are handled in strict accordance with the guidelines set forth by the Clinical Laboratory Improvement Act 1988 CLIA), Centers for Medicare and Medicaid Services (CMS)/Health and Human Services (HHS), and Agency for Healthcare Agency (AHCA). Parents or legal guardians of infants have the option of not having their child tested and can opt out of the program.

Based on a review of current practices among other United States Department of Health laboratories performing newborn screening, the Florida Department of Health Bureau of Laboratories has implemented a policy to retain newborn screening specimen cards for six months only. At the end of the six month period, these specimen cards containing dried blood spots will be destroyed according to appropriate biomedical waste procedures. Department of Health authority concerning the use and release of blood spots is enumerated in 383.14 F.S. which does not allow the release of blood spots for research.

Florida Newborn Screening Disorder Panel

Amino Acidemia Disorders

1. Phenylketonuria (PKU)
2. Maple Syrup Urine Disease (MSUD)
3. Homocystinuria (HCY)
4. Arginosuccinic Acidemia (ASA)
5. Citrullinemia (CIT)
6. Tyrosinemia Type II (TYR II)
7. Tyrosinemia Type I (TYR I)

Fatty Oxidation Disorders

8. Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
9. Very Long-chain acyl-CoA Dehydrogenase Deficiency (VLCAD)
10. Long-chain L-3-OH Acyl-CoA Dehydrogenase Deficiency (LCHAD)
11. Trifunctional Protein Deficiency (TFP)
12. Carnitine Membrane Transporter Deficiency (CUD)
13. Carnitine/Acylcarnitine Translocase Deficiency (CAT)
14. Carnitine Palmitoyl Transferase Deficiency I (CPT-I)
15. Carnitine Palmitoyl Transferase Deficiency II (CPT-II)
16. Short Chain Acyl-CoA Dehydrogenase Deficiency (SCAD)
17. Multiple Acyl-CoA Dehydrogenase Deficiency (MADD/GA-II)

Organic Acidemia Disorders

18. Isovaleric Acidemia (IVA)
19. Glutaric Acidemia Type I (GA I)
20. 3-OH 3-CH₃ Glutaric Aciduria

- (HMG)
21. Multiple Carboxylase Deficiency (MCD)
22. Methylmalonic Acidemia (mutase deficiency) (MUT)
23. Methylmalonic acidemia (Cbl A,B) (MMA)
24. 3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC)
25. Propionic Acidemia (PA) (PROP)
26. Mitochondrial Acetoacetyl-CoA Thiolase (beta-ketothiolase) Deficiency (SKAT) (BKT)

Endocrine Disorders

27. Congenital Adrenal Hyperplasia (CAH)
28. Congenital Hypothyroidism (CH)

Enzyme Disorders

29. Galactosemia (GALT)
30. Biotinidase Deficiency (BIOT)

Hemoglobinopathies

31. Hemoglobin SC Disease (FSC)
32. Sickle-Beta Thalassemia (FSA)
33. Sickle Cell Anemia (FS)

34. Cystic Fibrosis (CF)

35. Hearing Loss

Metabolic Conditions

These disorders, which are rare, fall into three different biochemical categories - amino acidemia disorders, organic acidemia disorders and fatty acid oxidation disorders. These disorders if left untreated may cause developmental delays, mental retardation, and death. Not all of the disorders identified through newborn screening and listed below are difficult to treat. Some require family education and oral medication. Some are treated with frequent feedings of normal food. Once the diagnosis is made, most of these disorders will also require lifelong management and follow up. Newborn Screening measures and identifies substances (amino acids and acylcarnitines) in a newborn's blood. Based upon the results of this test, an infant can be identified as having an increased risk of having an inherited metabolic disorder. Based upon this increased risk, the infant will need to have follow-up tests to confirm or rule out a diagnosis.

Amino Acidemia Disorders

Amino acid disorders are inherited metabolic conditions that affect the breakdown pathway of amino acids. This disruption leads to the accumulation of amino acids and their metabolites in the body.

Amino acid disorders are caused by absence or deficiency of enzyme activity at a specific step in the amino acid breakdown. These are autosomal recessive disorders in which the newborn is given one copy of the mutated gene from each parent. If untreated the baby could have poor feeding, vomiting, neurological symptoms, mental retardation, coma, and death.

- Phenylketonuria (PKU)
- Maple Syrup Urine Disease (MSUD)
- Homocystinuria (HCY)
- Arginosuccinic Acidemia (ASA)
- Citrullinemia (CIT)
- Tyrosinemia type II (TYR II)
- Tyrosinemia Type I (TYR I)

Treatment

A strict dietary management of the amino acid whose breakdown pathway is hindered must be followed for life. Medication may also be given to help dispose of toxic metabolites.

Fatty Acid Oxidation Disorders

Fatty acid oxidation disorders are inherited metabolic disorders leading to an accumulation of fatty acids and can interfere with energy supply after glycogen stores are depleted. Mitochondrial beta-oxidation of fatty acids is crucial to the body's ability to produce energy during fasting. In infants, a "fasting" state can be produced in as little as four hours. Failure to diagnose fatty acid disorders may result in excessive fat buildup in

the liver, heart and kidneys. If left untreated, low blood sugar, vomiting, seizures, lethargy, liver disease, developmental delay, coma, and death could occur. Many deaths due to fatty acid disorders have been misdiagnosed as Sudden Infant Death Syndrome (SIDS).

- Medium chain Acyl-CoA Dehydrogenase deficiency (MCAD)
- Very long-chain acyl- CoA Dehydrogenase deficiency (VLCAD)
- Long-chain L-3-OH Acyl-CoA Dehydrogenase deficiency (LCHAD)
- Trifunctional protein deficiency (TFP)
- Carnitine uptake deficiency (CUD)
- Carnitine/Acylcarnitine Translocase deficiency (CAT)
- Carnitine Palmitoyl Transferase deficiency type I (CPT-I)
- Carnitine Palmitoyl Transferase deficiency type II (CPT-II)
- Short chain Acyl-CoA Dehydrogenase deficiency (SCAD)
- Multiple Acyl-CoA Dehydrogenase deficiency (GA-II)

Treatment

Avoid fasting. Infants should be fed around the clock every 2-4 hours. If an illness occurs and is hospitalized, 10% dextrose IV should be started immediately to prevent hypoglycemia. Depending on the disorder, supplemental carnitine, a low fat diet, and home glucose monitoring may be prescribed.

Organic Acidemia Disorders

Organic Acidemia disorders are inherited disorders that cause a buildup of toxic organic acids due to the body's inability to break down certain amino acids and organic acids. Since the body cannot properly break down these amino acids, certain organic acids build up in the blood and urine. Most of these disorders have severe forms that present in the first week of life and constitute a neonatal emergency. Infants are generally well at birth, but the neonate can develop poor feeding, liver and kidney problems, irritability, lethargy, vomiting, severe metabolic ketoacidosis, mental retardation, and possibly death if left untreated

- Isovaleric Acidemia (IVA)
- Glutaric Acidemia type I (GA I)
- 3-OH 3-CH₃ Glutaric Aciduria (HMG)
- Multiple carboxylase deficiency (MCD)
- Methylmalonic Acidemia (mutase deficiency) MUT
- Methylmalonic Acidemia (MMA)
- 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC)
- Propionic Acidemia (PROP)
- Mitochondrial acetoacetyl-CoA thiolase deficiency (BKT)

Treatment

Any infant suspected of an organic acidemia disorder should be treated as a neonatal emergency. Treatments, which must be continued for life, consist of strict dietary amino acid restrictions and medications. They will need close supervision by a metabolic specialist and dietician.

Newborn Screening Disorder Testing Methodology:

Tandem mass spectrometry

Special Considerations

Low birth weight, Total Parenteral Nutrition TPN, carnitine, and other medications, i.e., antibiotics, can cause false positive results for MSMS disorders.

Subsequent abnormal specimens for term infants (more than 37 weeks gestational age) with abnormal borderline levels for Biotinidase Deficiency, Galactosemia, MS/MS disorders and babies on TPN will not be referred to the CMS Genetic Centers or need a repeat specimen if the initial specimen has normal results and is valid (collected after 24 hours of age and after the infant has been on protein feed for 24 hours). This policy does not apply to presumptive positive results if the newborn has had a previous normal specimen. All newborns with presumptive positive results should be referred to the CMS Genetic Centers.

Follow-Up Actions for Abnormal Screening Results - Metabolic Disorders

Determination	Follow-Up Action
Within Normal Limits	No further action needed.
Borderline Result	The primary care physician and/or parent is contacted for instructions to obtain a repeat specimen.
Second Borderline Result	The baby is referred to the Regional Genetics Center for evaluation and diagnostic testing.
Presumptive Positive	The baby is referred to the Regional Genetics Center for evaluation and confirmatory testing.

Endocrine Disorders

Congenital Adrenal Hyperplasia (CAH)

Congenital Adrenal Hyperplasia (CAH) is a group of autosomal recessive disorders in which there is a deficiency in one of the many enzymes needed to make cortisol. Cortisol is an adrenal gland hormone necessary to maintain blood sugar levels, maintain body fluids and electrolytes, and protect the body against stress.

As a result of inadequate production of cortisol, an infant is unable to maintain adequate energy supply and blood sugar levels to meet the stress of injury or illness. Lethargy and coma may progress to death. In some cases the production of aldosterone is also limited, which can result in dehydration due to sodium and water loss in the urine. Potassium also accumulates in the blood, causing irritability or lethargy, vomiting and muscle weakness, including cardiac muscle irritability. Additionally, an increase in the production of androgens, or virilizing hormones, can cause female infants to develop ambiguous genitalia, or if very severe, the infant may exhibit external genitalia resembling a normal male infant with undescended testes.

Unlike some other disorders in newborn screening, infants may be symptomatic at birth. In the salt-wasting form, an infant can have a crisis within the first five days to several months of life. CAH occurs in one out of every 12,000-15,000 births.

Newborn Screening Disorder Testing Methodology:

17 Hydroxyprogesterone (17-OHP) levels

Treatment

Treatment consists of hormone replacement therapy.

Special Considerations

Age and weight at time of collection are important as results are dependent upon this information.

Follow-Up Actions for Abnormal Screening Results for CAH

Initial Specimen Results - Determination	Recommended Action
Within Normal Limits	No further action needed.
CAH Early (<24 hours of age) – borderline results	Obtain a repeat specimen. Babies in the NICU should follow the NICU guidelines for specimen collection.
CAH Low birth weight (<2500 grams) - borderline results	Obtain a repeat specimen. Babies in the NICU should follow the NICU guidelines for specimen collection.
CAH > 24 hours & > 2500 grams - borderline results	Obtain repeat specimen. It is recommended that a specimen be obtained when the infant is two weeks of age to identify asymptomatic males.
CAH > 24 hours & > 2500 grams - repeat borderline results	<p>If the repeat specimen result determination is abnormal, take the following actions:</p> <p>If the repeat specimen is elevated, refer baby to the Regional Endocrine Center for evaluation and diagnostic testing.</p> <p>If the repeat specimen is still abnormal but decreasing in value, take following actions:</p> <ol style="list-style-type: none"> 1. If baby is in NICU, examine infant for ambiguous genitalia, hyponatremia and hyperkalemia and obtain repeat specimen. If symptoms are present or CAH is suspected, contact CMS for referral to the Regional Endocrine Center for evaluation and diagnostic testing. Babies in the NICU should follow the NICU guidelines for specimen collection. If third specimen has already been collected, collect specimen prior to discharge. 2. If baby is at home- obtain an immediate repeat specimen. Examine infant for ambiguous genitalia, hyponatremia and hyperkalemia and obtain repeat specimen. If symptoms are present or CAH is suspected, contact CMS for referral to the Regional Endocrine Center for evaluation and diagnostic testing.
Presumptive positive	Refer baby to the Regional Endocrine Center for evaluation and confirmatory testing.

Congenital Hypothyroidism (CH)

Congenital Hypothyroidism occurs when the thyroid gland fails to develop or function properly. The result is not enough thyroxine (thyroid hormone) being made by the thyroid gland. Adequate thyroxine is necessary for normal body growth and brain development.

Low birth weight, premature and sick newborns have been observed to have transient fluctuations in blood test results for congenital hypothyroidism. Standing physician orders and/or newborn intensive care nursery protocols are recommended because the newborn's condition dictates whether serum thyroid studies are warranted. Although cutoff values for borderline and abnormal hypothyroid results are the same for sick or premature neonates as for healthy full-term infants, follow-up of results is based on physician judgment in individual cases.

The screening test is not designed to detect late on-set hypothyroidism. This is a condition where the abnormality of the thyroid gland is usually not as severe, and thyroxine levels are high enough to pass the initial screen. However, with time the infant outgrows his/her ability to make adequate thyroxine. The physician must remain alert to clinical symptoms in older infants despite normal newborn screening results.

Infants with hypothyroidism frequently do not present with signs or symptoms in the newborn period. Five to ten percent with severe disease may present with prolonged neonatal jaundice, lethargy, poor muscle tone, feeding problems, constipation, coarse facial features, thick tongue, distended abdomen, umbilical hernia and hoarse cry.

Left untreated, congenital hypothyroidism may result in mental retardation, developmental delay and growth retardation. Congenital hypothyroidism occurs in one of every 3,500 births. It is twice as common in females as it is in males.

Newborn Screening Disorder Testing Methodology

T4 (thyroxine) and TSH (thyroid stimulating hormone) levels

Treatment

Treatment of congenital hypothyroidism is simple and effective. Thyroid hormone, in pill form, may be crushed and given with a small amount of formula. The dosage of medication is based on the child's weight and must be individualized and adjusted (by monitoring T4 and TSH levels) as the child grows. Pediatric endocrinology consultation should be obtained to determine recommendations for medication adjustment and follow-up for the child.

Special Considerations

Date and time of collection are important to note as results are interpreted differently once the infant is greater than 24 hours old. TSH levels are normally elevated in the first 24 hours of life.

Follow-Up Actions for Abnormal Screening Results - Congenital Hypothyroidism

Determination	Follow-Up Action
Within Normal Limits	No further action needed.
Low T4	The infant should have a repeat newborn screen at two to three weeks of age or have a free T4 and TSH collected at a local clinical laboratory to confirm results.
Borderline TSH	Physician discretion is strongly advised. The infant should have a repeat newborn screening specimen or serum TSH labs.
Second Borderline	If the second specimen is also a borderline result, the infant should be referred to the Endocrine Referral Center.
Date of Birth Hypothyroidism or Neonatal Hyperthyroxinemia	The Newborn Screening Laboratory report will recommend to the submitting entity or primary care physician to obtain a repeat newborn screening collection.
Presumptive Positive	The infant should be referred to the Endocrine Referral Center. The endocrinologist will notify the primary care physician and parent.

Endocrine Referral Center Responsibilities

The Endocrine Referral Center Specialist will consult with the infant's physician and family to arrange confirmatory testing and necessary follow-up treatment.

Enzyme Disorders

Galactosemia

Galactosemia is an inherited disorder which occurs when there is a deficiency of any of the three enzymes required to break down galactose (or milk sugar). The disorder results in elevated levels of galactose in the blood. Many infants with Galactosemia appear normal at birth but within a few days after milk feedings the symptoms of the disorder can become severe. Some of the early symptoms of Galactosemia include poor feeding, poor sucking reflex, vomiting, jaundice, irritability and seizures. If left untreated, Galactosemia can cause mental retardation, cataracts, failure to thrive and even death.

Newborn Screening Disorder Testing Methodology

Newborn Screening for Galactosemia in Florida is designed to detect classical Galactosemia due to a deficiency of the galactose – 1- phosphate uridyl transferase (GALT) enzyme. An assay measures the activity of the GALT enzyme in the red blood cells. The test associates the amount of GALT enzyme present with the amount of fluorescence appearing. Newborns with Classical Galactosemia may have no enzyme activity. Newborns with variant forms of Galactosemia may have decreased activity.

Treatment

Treatment for Galactosemia includes eliminating dietary galactose.

Special Considerations

Newborns who receive a blood transfusion prior to specimen collection may have false negative results for Galactosemia. A dried blood specimen must be collected before an infant receives a blood transfusion. In newborns who receive a transfusion prior to screening, a specimen should be obtained three to four months after the last transfusion.

The GALT enzyme is sensitive to heat. Therefore, specimens that are delayed in processing or exposed to heat may have false positive results.

GALT enzyme activity is dependent on the ingestion of lactose. Therefore, infants receiving lactose-free formula may have false negative results as well.

Follow-Up Actions for Abnormal Screening Results for Galactosemia

Determination	Action
GALT Partial	Infant should be referred to the Regional Genetics Center for follow-up and confirmatory testing
GALT PrePos	Infant should be referred to the Regional Genetics Center for follow-up and confirmatory testing

Biotinidase Deficiency

Biotinidase deficiency is an autosomal recessive disorder of biotin recycling. Biotin is an essential vitamin that is widely present in natural foods such as egg yolk, soybeans, and cereals. Its function is to activate certain enzymes, called “biotin-dependent enzymes.” These enzymes are important for breaking down some proteins, and producing certain fats and sugars. When biotinidase is not working properly, there is not enough biotin to perform its function. When this happens, harmful by-products build up in the body and may cause serious health problems.

Signs and symptoms may include seizures, skin rash, hair loss, hypotonia, ataxia, hearing loss, optic nerve atrophy, developmental delay and metabolic acidosis. Treatment is oral biotin on a daily basis. The occurrence of biotinidase deficiency (profound or partial) is one of every 60,000 births.

Newborn Screening Disorder Testing Methodology

The testing methodology for Biotinidase deficiency involves testing for Biotinidase enzyme activity. In September 2010, the Newborn Screening Laboratory changed their testing methodology.

Treatment

Treatment for Biotinidase deficiency involves daily oral biotin.

Special Considerations

- During specimen collection, it is imperative to allow alcohol to dry before initiating the heel stick and to waste the first blood drop as the interaction of alcohol and the blood specimen can produce a false presumptive positive result.
- Additionally, specimens should be kept away from direct light, heat, and moisture. Samples dried incompletely, exposed to moisture after drying, or exposed to heat may exhibit no biotinidase activity (false positive).

Follow-Up Actions for Abnormal Screening Results - Biotinidase Deficiency

Determination	Action
Normal Enzyme Activity	No further action needed
Partial Enzyme Activity (1 st borderline result)	Repeat specimen should be obtained.
Partial Enzyme Activity (2 nd borderline result)	Infant should be referred to the Regional Genetics Center for follow-up and confirmatory testing
Low or No Enzyme Activity	Infant should be referred to the Regional Genetics Center for follow-up and confirmatory testing

Hemoglobinopathies

Hemoglobinopathies, including sickle cell disease, are a group of inherited disorders caused by abnormal hemoglobin (Hgb) molecules in the red blood cells. These abnormal hemoglobin molecules are able to be detected at birth. The most common hemoglobinopathies detected in Newborn Screening are Sickle Cell Anemia (Hgb SS), Hemoglobin SC disease (Hgb SC), and Sickle Beta Thalassemia (Hgb SA). The most clinically significant abnormal hemoglobin condition is sickle cell anemia.

Infants with hemoglobinopathies appear normal at birth and typically develop symptoms during infancy or early childhood. Complications of hemoglobinopathies vary from mild to severe and include pneumococcal sepsis, severe musculoskeletal pain, gallstones, chronic anemia, jaundice, delayed growth, splenomegaly, stroke and acute chest syndrome.

Individuals with one abnormal hemoglobin chain and one normal hemoglobin chain are carriers - often referred to as "trait." Most hemoglobin carriers have few or no clinical symptoms. Carrier detection provides the opportunity to educate families, to test other family members and to offer genetic counseling to those with positive results. Genes for Hemoglobinopathies are most common in people of African, Mediterranean, Middle Eastern, Caribbean, Indian, and South American descent, but can appear in any race or ethnic group.

Newborn Screening Disorder Testing Methodology:

The Florida Newborn Screening Laboratory screens for the presence of hemoglobins in the blood: F, S, A, C, E, D, and BARTS.

Treatment

Infants with significant hemoglobinopathies should have a primary care provider and receive periodic evaluation in a comprehensive care setting. Treatments include managing acute illness, parent education and genetic counseling as needed. All newborns identified with Sickle Cell Anemia must start prophylactic penicillin before two months of age.

Follow-up Actions

A letter will be sent to the mother/guardian of an individual with hemoglobin trait. Presumptive positive babies are referred to a referral center.

Special Considerations

If a sample is collected after the newborn receives a transfusion, the results of the hemoglobinopathy test are invalid. A metabolic screen must be collected before an infant receives a blood transfusion. In newborns who receive a transfusion prior to the collection of the first metabolic screen, a specimen should be obtained 3 to 4 months after the last transfusion.

Follow-Up Actions for Abnormal Results - hemoglobinopathies

Determination	Possible Causes	Follow-Up Action
FS	Sickle Cell Disease	Infant should be referred to the regional hematology center. American Academy of Pediatrics (AAP) recommends starting prophylactic Penicillin or Erythromycin by two months of age.
FSC	Sickle Hemoglobin C Disease	Infant should be referred to the regional hematology center.
FSA	Sickle Beta Thalassemia	Infant should be referred to the regional hematology center.
F + Other	Other Hemoglobinopathy	Infant should be referred to the regional hematology center.
F	Premature Infant OR Homozygous Sickle Beta Zero Thalassemia; diabetes in mother	Infant should be referred to the regional hematology center.
FC	Hemoglobin C Disease	Infant should be referred to the regional hematology center.
FAS	Sickle Cell Trait	Letter sent to parents and physicians regarding results and any further recommendations.
FAC	Hemoglobin C Trait	Letter sent to parents and physicians regarding results and any further recommendations.
FA BARTS	Alpha Thalassemia Trait	Letter sent to parents and physicians regarding results and any further recommendations.
FA + Other	Other Hgb Trait	Letter sent to parents and physicians regarding results and any further recommendations.
FE	Hemoglobin E Disease	Infant should be referred to the regional hematology center.
FD	Hemoglobin D Disease	Infant should be referred to the regional hematology center.
FSE	Sickle Hemoglobin E Disease	Infant should be referred to the regional hematology center.
FSD	Sickle Hemoglobin D Disease	Infant should be referred to the regional hematology center.
FAE	Hemoglobin E Trait	Letter sent to parents and physicians regarding results and any further recommendations.
FAD	Hemoglobin D Trait	Letter sent to parents and physicians regarding results and any further recommendations.

Cystic Fibrosis (CF)

Cystic Fibrosis is the most common inherited fatal disease and primarily affects the lungs and digestive system. The gene for CF has been identified on chromosome 7 and codes for a protein that causes the absence or malfunction of a chloride channel on a cellular level. This causes decreased transport of chloride across cell membranes and leads to abnormal secretions in any organ that produces secretions, such as the lungs, intestines, pancreas, liver, reproductive organs and sweat glands. This leads to a multitude of problems including frequent lung infections and bronchiectasis with an excessive inflammatory response, obstruction of the pancreatic ducts leading to malabsorption, diabetes, cholestasis with possible consequences of liver failure and gall bladder disease, and absence of the vas deferens with male infertility. The malfunctioning chloride channels also provide the classic way to test for Cystic Fibrosis with a quantitative sweat test performed at an approved CF Center (and still the gold standard diagnostic test) with high salt content in the sweat of individuals with CF.

People with CF can have a variety of symptoms, including:

- very salty-tasting skin
- persistent coughing, often productive
- frequent lung and sinus infections
- wheezing or shortness of breath
- poor growth/weight gain
- meconium ileus (in newborns)
- frequent greasy, bulky stools or difficulty in bowel movements

If left untreated, cystic fibrosis will lead to impaired growth usually below the 10th percentile, severe chronic lung infections and an early death.

Newborn Screening Disorder Testing Methodology:

The newborn screening test for cystic fibrosis involves **two steps**:

- The dried blood specimens obtained from routine newborn screening is tested for levels of trypsinogen using the Immunoreactive Trypsinogen Test (IRT). Trypsinogen is an enzyme produced in the pancreas that is higher in infants with CF.
- The top four percent of specimens of each testing day with the highest trypsinogen levels are subjected to DNA mutation analysis.

Special Considerations

- IRT levels in some affected infants may decline into the normal range by ten days of age, and thus may not be picked up by repeat screenings.
- IRT levels may be falsely elevated in premature or sick infants.

(Current list at time of printing – may change at the discretion of the Genetics and Newborn Screening Advisory Council recommendation.)

CF Mutations Screened in Florida	
1078delT	G551D *
1717-1G>A *	G85E *
1898+1G>A *	N1303K *
2183AA>G	Q493X
2184delA *	R1162X *
2789+5G>A *	R117H *
3120+1G>A *	R334W *
3659delC *	R347H
3849+10kbC>T *	R347P *
3849+4A>G	R553X *
3876delA	R560T *
3905insT	S549N
394delTT	S549R A>C
621+1G>T *	S549R T>G
711+1G>T *	V520F
A455E *	W1282X *
D1152H	Y1092X C>A
deltaF508 *	Y1092X C>G
deltaI507 *	Y122X
E60X	IVS8-5T **
G542X *	IVS8-7T **
	IVS8-9T **

*American College Medical Genetics (ACMG) Panel

**If present with R117H and IVS8-5T

Follow-Up Actions for Abnormal Results for CF

Value	Determination	Follow-up Action
IRT < 96 th percentile	Considered normal	No further action needed
IRT > 96 th percentile and no mutations	Below 160ng/mL IRT	No further action needed
IRT > 96 th percentile and no mutation	Above 160ng/mL IRT	Collect repeat newborn screening specimen
IRT > 96 th percentile and 1 mutation	Possible CF	Refer for Sweat Test at CF Center
IRT > 96 th percentile and 2 mutations	Possible CF	Refer for Sweat Test at CF Center

Cystic Fibrosis Referrals

There are two reasons why a newborn may be referred to the Cystic Fibrosis Center:

1. **The trypsinogen level is elevated and there are two CF gene mutations found.** The child is presumed to have CF but a sweat test is needed to confirm the diagnosis. Genetic counseling for the parents is also recommended. There will be approximately 30-50 infants born in Florida each year that will have two CF gene mutations found through newborn screening.
2. **The initial trypsinogen level is elevated and there is one CF gene mutation found.** There is a 1 in 20 chance that the child has CF. A single gene mutation is not sufficient for CF, but additional CF gene mutations exist that the Newborn Screening Laboratory is not testing for since they are too rare. The possibility exists that the child could have a second CF gene mutation not identified through the newborn screening test. A sweat test is needed to determine whether or not the child has CF. Genetic counseling for the parents is also recommended and prenatal testing results would be considered by the CF Center. If a child has one CF gene mutation, then one of the parents is also a carrier of the CF gene mutation. The carrier status of the parents can only be determined by further genetic evaluation. There will be approximately 625 infants born in Florida each year that will have at least one CF gene mutation found through newborn screening.

If the primary care provider has concerns, i.e., the baby is not growing, is failing to thrive, or has a family history of CF, they may refer to the CF Center for further evaluation irrespective of newborn screening results.

If multiple specimens show an elevated IRT level (with no CF mutations), referral to the CF Center is discretionary by the Newborn Screening Follow-Up staff or primary care physician.

Hearing Screening

Suggested guidelines for newborn hearing screening and evaluation services in the State of Florida

Purpose

- To provide a description of a suggested quality hearing screening program for newborn nurseries.
- To provide a description of recommended procedures to audiologists performing follow up hearing screening or audiological assessment of infants identified during universal newborn hearing screening.

Description

Newborn Hearing Screening services are provided to identify newborns at risk of hearing impairment and to assure that follow-up audiometric screening, diagnosis, and referral to intervention is provided as indicated in accordance with Florida Statutes [391.301-304](#), [383.14](#) and [383.145](#).

Legislative Intent

To provide a statewide comprehensive and coordinated interdisciplinary program of early hearing screening and follow-up care for newborns identified as referring from the hearing screening process. The goal is to screen all newborns for hearing impairment in order to alleviate the adverse effects of hearing loss to speech and language development, academic performance, and cognitive development.

Definition of Targeted Hearing Loss

Congenital permanent bilateral, unilateral sensory or permanent conductive hearing loss of newborns in well-infant nurseries to include neural hearing loss (e.g., auditory neuropathy / auditory dyssynchrony) in infants admitted to the Newborn Intensive Care Unit (NICU) for more than five days. Identification of risk factors associated with delayed onset or progressive hearing loss.

Suggested Inpatient Community Hospital Guidelines

❖ **Definition**

An inpatient newborn hearing-screening program will be capable of providing newborn hearing screening testing to all newborns and infants during their birth admission or neonatal intensive care unit admission.

❖ **Standards for Hearing Screening**

It is recommended that:

- **The hospital designates one individual** (preferably an audiologist) to serve as the contact person for the newborn hearing screening program. The designated person is responsible for ensuring that persons who perform hearing screening are sufficiently trained, coordinating follow-up services, and managing all correspondence. This person will also serve as the liaison between the hospital, family, and Children’s Medical Services.

- **The hospital demonstrate training** criteria and competencies for newborn hearing screeners as determined by the hospital and document dates of training for each person who performs hearing screening. A CD/DVD called *Universal Newborn Hearing Screening Training Program* on screening techniques and necessary competencies is available to hospital newborn hearing screening programs from the Children’s Medical Services Newborn Screening Program by calling (850) 245-4201.

- **The hospital have protocols, policies, and procedures** available for inspection that provide operational details of the facility’s newborn hearing screening program including:
 - the staff training criteria
 - staff roles and responsibilities, including supervision of screening outcomes
 - referral and follow-up procedures
 - protocols for follow-up testing of babies who were discharged before receiving a hearing screening. Follow-up protocols may include return to the hospital for outpatient screening or referral to an audiologist.
 - procedure for reporting screening results in each individual child’s medical record
 - culturally and linguistically appropriate information for distribution to parents
 - documentation of final screening prior to discharge including:
 - screening outcome (pass or refer).
 - if a child is discharged from the hospital in “refer” status, the discharge documents will include a referral for follow up hearing testing.
 - if a child is discharged from the hospital in “refer” status,, an appointment will either be scheduled for follow up hearing testing as a hospital outpatient or the mother will be given information and resource materials to make an appointment for follow-up hearing testing. If there is no payer source for follow-up hearing testing contact (850) 245-4201 for information on potential funding sources.
 - the follow up appointment is to occur within 30 days of referral from hearing screening and diagnostic procedures are to be completed no later than three months of age.

- **Each hospital provides that all newborns receive a hearing screening prior to discharge.**
 - Each ear will be screened at each stage of screening.
 - In the well-baby nursery, newborns may be screened by either ABR or OAE techniques. If the child does not pass the initial screening and is available for re-test at a later time, then the child should be retested prior to discharge using either ABR or OAE. Hospitals are cautioned not to allow excessive rescreening on each test encounter (e.g., eight to ten attempts in a single encounter) to try to obtain a pass result after a valid refer has been recorded.
 - Newborn Intensive Care Unit: Infants admitted to Level II and/or Level III NICUs for more than five days will receive a screening of both ears with ABR. If the child does not pass the initial screening and is available for retest prior to discharge, then the retest should be completed using ABR.

- **Newborn hearing screening will be conducted by** a licensed audiologist, physician, or other appropriately supervised individual who has completed documented training specifically for newborn hearing screening.

- **The following risk factors should be considered for each newborn.** If present, the appropriate risk factor(s) should be checked on the Newborn Screening Specimen Card:
 - Family history of permanent childhood hearing loss (blood relative with permanent hearing loss in early childhood, e.g. grandparent, parent, aunt, uncle, first cousin, siblings).
 - Hyperbilirubinemia at a serum level requiring exchange transfusion.
 - Persistent pulmonary hypertension of the newborn associated with mechanical ventilation (PPHN).
 - Conditions requiring the use of extracorporeal membrane oxygenation (ECMO).
 - Birth weight < 1500 Grams.

- **As specified by law ([F.S. 383.145](#)), parents will sign a waiver only** if a hearing screening is refused. The signed document must be placed in the newborn's medical record. The hospital should give the parents who refuse information on appropriate developmental auditory, speech and language milestones such as what is included in the Children's Medical Services Newborn Screening Program brochure.

- **The hospital should inform parents,** in writing, of the results of the hearing screening, prior to hospital discharge. Screening results should be conveyed immediately to families so they understand the outcome and importance of follow-up when indicated. The parents should be given information on appropriate developmental auditory, speech and language milestones such as what is included in the Children's Medical Services

Newborn Screening Program brochure. For infants who are referred, a brochure offering information for the parents is available at no charge from the Children's Medical Services Newborn Screening Program. To request the *Does your baby need another hearing test?* brochure complete a request form at (a link will be inserted here once the form is posted on the website).

- **As specified by law (F.S. 383.145)**, the hospital shall ensure documentation of the hearing screening results in the newborn's medical record. It is recommended that the result of the final hearing screening prior to discharge and the presence of any hearing loss risk factors be recorded in a prominent place on the discharge summary.
- **The hospital should document the need for a hearing screening referral** as part of the discharge summary for the newborns that leave the hospital in a "refer" status (e.g., newborns who failed the in-hospital hearing screening or who were not screened prior to discharge), including the appointment time and place scheduled for hearing follow-up testing. The *Does your baby need another hearing test?* brochure has been designed to cite the follow-up hearing test appointment information. The brochures can be obtained at no cost by completing a request form at (a link will be inserted here once the form is posted on the website).
- **The newborn's hearing results shall be recorded** on the HEARING SCREENING INFORMATION section of the Newborn Screening Specimen Cards and subsequently submitted to the Department of Health Bureau of Laboratories at 1217 Pearl Street, Jacksonville, Florida 32202. If genetic screening is performed before the hearing screening has been completed, the appropriate box should be marked signifying that the hearing screening had not yet been done. If the hearing screening is completed at a time separate from the genetic screening, only the hearing section of the Specimen Card should be completed and submitted to the Department of Health Bureau of Laboratories-Jacksonville. Hospitals with access to the electronic birth registration system should enter hearing screening results into this system.
- **Infants readmitted within the first month of life** should receive a repeat hearing screening prior to discharge if there are conditions present that are associated with potential hearing loss (i.e., hyperbilirubinemia requiring exchange transfusion, culture results indicating sepsis or meningitis, and/or ototoxic medication exposure).

❖ **Audiological Screening Guidelines**

It is recommended that:

Determination of presence or absence of an Auditory Evoked Potential (e.g. ABR) and/or Otoacoustic Emissions (OAE) at a predetermined screening level to assess the need for further audiological evaluation.

- Newborn hearing screening services should be performed using FDA approved otoacoustic emissions or evoked potential testing that detects mild to profound hearing loss in newborns (ABR is to be used to screen infants with NICU stays of more than five days).
- Use of screening equipment should be in accordance with manufacturer's protocols and stated norms for newborn hearing screening purposes.
- The equipment should be calibrated in accordance with the manufacturer's recommendation and a log should be kept documenting the dates of calibration, repair or replacement of parts.
- For rescreening or a complete evaluation it is necessary to test both ears, even if only one ear caused a referral from newborn hearing screening.

❖ **Quality Indicators of Appropriate Referral Rate**

It is recommended that:

- A minimum of 96% of newborns born in the hospital should receive a hearing screening prior to discharge.
- A maximum outpatient referral rate of 4% of all newborns screened prior to discharge should be achieved.
- Following a two-stage screening, a minimum outpatient referral rate of 1% for ABR screening and 2% for OAE screening should be observed for all newborns screened prior to discharge (if the refer rate is worse than these percentages the hospital should train screening personnel on the importance of the two-stage (or repeat) screening process while avoiding excessive rescreening during an single encounter).

Suggested Practice Guidelines for Follow-up of Newborn Hearing Screening Referrals

❖ **Definition: Referral and outpatient follow-up will be necessary for:**

- Any newborn who did not receive a hearing screening prior to discharge, including home births.
- Any newborn that was discharged with a “refer” result during the inpatient stay.
- If conditions associated with delayed onset hearing loss are documented (refer to Appendix A for hearing loss risk factors).
- All infants with a risk factor for hearing loss (Appendix A), regardless of status on developmental milestones, should be referred for an audiological assessment at least once by 24 to 30 months of age. Children with risk factors that are highly associated with delayed-onset hearing loss, such as ECMO or CMV infection, should have more frequent audiological assessments.

❖ **Standards for follow-up of newborn hearing screening results for all referrals**

A pediatric audiologist or hospital based screening program will provide follow-up outpatient hearing testing. It is preferable when referring to an audiologist that an appointment be scheduled with a [CMS Approved Pediatric Audiologist](#). It is standard care for the follow-up screening or audiological evaluation to be completed within the first 30 days following discharge and for all diagnostic procedures to be completed prior to age three months.

- **At the time of discharge for newborns who are referred from newborn hearing screening** (including those who are discharged before hospital screening was completed):
 - The physician completing the discharge process will prepare a written referral for follow up hearing testing.
 - When possible, the appointment for follow up testing (hearing screening or audiological evaluation) will be scheduled prior to discharge and the appointment time and place included on the discharge summary to be given to parents.
 - The referral for follow-up screening should be to a pediatric audiologist or outpatient hospital program with FDA approved equipment for newborn hearing screening that uses a protocol instituting 30-35 dB HL newborn hearing screening criteria.
 - The state metabolic laboratory will prepare reports newborn screening results, including hearing, that are delivered to all birth hospitals to file in the medical record of all newborns. If a physician of record is known, hospitals will send this information to the newborn's physician (medical home) of record.
- **Newborns failing a subsequent screening procedure require audiological evaluation**, preferably by a [CMS Approved Pediatric Audiologist](#). It is standard of care for this evaluation to follow a protocol that renders information regarding the infant's auditory thresholds, status of auditory nerve and brainstem pathway, and determination of locus of hearing impairment if abnormal results are found. Refer to Appendix B for the recommended audiological evaluation and reporting protocols.

❖ **Standards for reporting hearing screening and follow-up results**

- **For newborns who (1) were discharged prior to screening completion, or (2) were readmitted in the first 30 days of life and rescreening is warranted the hearing (re)screening results shall be recorded on either:**
 - The Department of Health [Repeat Hearing Screen Form](#) and faxed to the Newborn Screening Program (850) 245-4049

OR

- The Dried Blood Newborn Screening Specimen Cards and submitted to the Department of Health Bureau of Laboratories at 1217 Pearl Street, Jacksonville, Florida 32202.

➤ **For newborns who failed the in-hospital screening:**

The hearing (re)screening performed in the hospital shall be recorded on either:

- The Department of Health [Repeat Hearing Screen Form](#) and faxed to the Newborn Screening Program (850) 245-4049 **or**

OR

- The Newborn Screening Specimen Cards and submitted to the Department of Health Bureau of Laboratories at 1217 Pearl Street, Jacksonville, Florida 32202.

The hearing (re)screening performed by an audiologist shall be recorded on either:

- The Department of Health [Repeat Hearing Screen Form](#) and faxed to the Newborn Screening Program (850) 245-4049

OR

- [Diagnostic Hearing Evaluation Form](#) and faxed to the Newborn Screening Program (850) 245-4049

➤ **Test results that confirm the presence of a hearing loss** for newborns or infants who receive a diagnostic audiological evaluation shall be recorded on [Diagnostic Hearing Evaluation Form](#).

➤ **Evaluation tools for outpatient screening of “refer” status follow-up**

- Refer to Appendix B for information on the rescreen/evaluation protocol to be used.
- Results / recommendation:
 - Parents should receive information about hearing, speech and language milestones and information regarding risk factors for progressive hearing loss.
 - For infants that exhibit a “pass” result after follow up hearing testing, the hearing test results shall be:
 - Sent in writing to the primary care physician (medical home)

- Reported to the Children’s Medical Services Newborn Screening Program via:
 - ✓ The Department of Health [Repeat Hearing Screen Form](#) and faxed to the Newborn Screening Program (850) 245-4049
 - OR**
 - ✓ The Newborn Screening Specimen Cards and submitted to the Department of Health Bureau of Laboratories at 1217 Pearl Street, Jacksonville, Florida 32202.
- For infants that continue to exhibit a “refer” result:
 - A referral will be made to an audiologist for a complete audiological evaluation.
 - The results of hearing testing should be sent in writing to the primary care physician (medical home) for further audiological and medical evaluations and referrals, as appropriate.
 - The hearing results indicating a continuation of “refer” status shall be reported to the Children’s Medical Services Newborn Screening Program via:
 - ✓ The Department of Health [Repeat Hearing Screen Form](#) and faxed to the Newborn Screening Program at (850) 245-4049
 - OR**
 - ✓ The Newborn Screening Specimen Cards and submitted to the Department of Health Bureau of Laboratories at 1217 Pearl Street, Jacksonville, Florida 32202.

➤ **Quality Indicators for rescreening rate:**
It is recommended that:

- Achieve 95% attendance for infants rescheduled for outpatient re-screening.
- Achieve 100% reporting of newborn hearing rescreen results to the Department of Health.

Considerations for Audiological Assessment of Infants (0-12 months) with Hearing Loss

❖ **Definition**

A licensed [CMS-Approved Pediatric Audiologist](#) with experience in assessing hearing in infants should conduct a diagnostic audiological evaluation. The audiologist must have the equipment necessary to complete all described evaluation procedures. The goal is

to determine the presence or absence of a hearing loss through the application of a battery of audiological tests culminating in the referral of children diagnosed with hearing loss to local Children's Medical Services Part C Early Steps for intervention services.

❖ **Audiologic Assessment Guidelines for Infants and Toddlers in Florida**

Appendix B is a summary of the recommended audiological evaluation and reporting protocols. Consult the [Guidelines for Infant Hearing Screening, Referral, Audiologic Assessment, Hearing Loss Management and Early Intervention document](#) for detail on recommended assessment and management procedures.

❖ **Criteria for significant hearing loss for eligibility for local Children's Medical Services Part C Early Steps**

- Evidence of a documented permanent hearing threshold level of (Re: ANSI 1996):
 - 25 dB or greater based on pure tone average of 500, 1000, and 2000 Hz unaided in the better ear (Air-bone gap not to exceed 10 dB HL)
 - Air conduction thresholds, unaided in the better ear, 25 dB or greater HL at two or more frequencies in the high frequency range (2000, 3000, 4000, 6000 Hz) in both ears with air-bone gaps no greater than 10 dB HL.
- Evidence of auditory dys-synchrony (auditory neuropathy) in both ears characterized by a unique constellation of behavioral and physiologic auditory test results.

❖ **Quality Indicators for referral to early intervention**

- Achieve 100% referral rate to the local Early Steps within two business days of diagnosis of infants with permanent or long term conductive hearing loss. Reporting of infants and toddlers with diagnosed hearing loss to the local Children's Medical Services Part C Early Steps is required by [Federal Law 34 CFR, § 303.321 d.2.](#)
- For infants that are confirmed to have hearing loss, parents should receive information about support services available in the form of:
 - "Serving Hearing Impaired Newborns Effectively (SHINE)" brochure (available free of charge from the Children's Medical Services Newborn Screening Program at (850) 245-4201).
 - "Florida's Resource Guide for Families of Young Children with Hearing Loss " (available free of charge from the Children's Medical Services Newborn Screening Program at (850) 245-4201).
- The outcome of the objective test measurements should be in agreement with parental report of sound awareness and alertness to environmental acoustic stimulation.
- Achieve a diagnosis with initiation of amplification fitting as appropriate by four months of age for 95% of infants with significant hearing loss.
- Completion of early intervention evaluation and initiation of family-centered early intervention services no later than six months of age.

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.

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:
 - a. ECMO** assisted ventilation
 - b. Exposure to ototoxic medications (gentamycin and tobramycin)
 - c. Exposure to loop diuretics (furosemide/Lasix)
 - d. Hyperbilirubinemia that requires exchange transfusion
4. In utero infections, such as CMV**, 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.
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**

Source: Year 2007 Position Statement: Principles and Guidelines for Early Hearing Detection and Intervention Programs, Joint Committee on Infant Hearing.

Florida Protocol for Follow-Up Hearing Evaluation and Reporting of Results

Evaluation and Practice Protocols

It is necessary for pediatric audiologists to abide by the evaluation and practice protocol below for infants referred from newborn hearing screening (per [Guidelines for Infant Hearing Screening, Referral, Audiologic Assessment, Hearing Loss Management and Early Intervention](#)). NOTE: It is recognized that there may be some situations in which professional judgment should supersede the below practice protocols.

- **IF** failed one hospital screening test:
THEN: screening or diagnostic OAE **OR** screening click AER
- **IF** failed two hospital screening tests:
THEN: diagnostic OAE testing (2000 Hz – 6000 Hz) **OR** air conduction click AER
IF diagnostic OAE results are abnormal or inconclusive **OR** air conduction click AER was not performed
THEN: high frequency tympanometry (600 Hz or 1000 Hz probe tone)
AND: air conduction click AER
- **IF** air conduction click AER is abnormal:
THEN: bone conduction click AER
AND: frequency specific tone bursts (500 Hz, 2000 Hz, 4000 Hz)
AND/OR: Auditory Steady State Response (ASSR)
- **IF** there is a suspicion of auditory dyssynchrony, confirm by observing **more than one** of the following:
 - a. AER wave 1 is present with the absence of later waveforms
 - b. Cochlear microphonic is verified in the complete absence of all waveforms
 - c. Middle ear muscle reflex is absent
 - d. AER response indicates moderate hearing loss but there is no observable response to sound stimulation in combination with absent middle ear muscle reflex
 - e. Presence of fluctuating hearing thresholds in the absence of any detectable middle ear abnormalities
 - f. Presence of normal OAEs and abnormal ABR results.
- **IF** the child is an audiological candidate for amplification:
THEN: obtain medical clearance for amplification fitting
AND: use probe microphone measures as a part of appropriate hearing aid fitting

Timelines and Reporting

It is intended that children referred from newborn hearing screening have their hearing status confirmed by three months of age. Therefore, the following activities must occur within the specified timelines, including reporting hearing results to the Department of Health, Children’s Medical Services Newborn Screening Program:

- A. Every effort shall be made to schedule infants referred from newborn hearing screening so that follow up screening or diagnostic procedures will be completed within 15 working days following the day of referral.

- B. Every effort shall be made to re-schedule missed appointments within ten working days of the missed appointment.
- C. Any no-show family will be contacted and rescheduled within 15 working days of the missed appointment. Fax the [Repeat Hearing Screen Form](#) (for missed screenings) or [Diagnostic Hearing Evaluation Form](#) (for missed diagnostic evaluations) to the CMS Newborn Screening Unit, indicating that the family did not show for the appointment so that follow up with the family can occur.
- D. Follow up evaluation of well babies with abnormal diagnostic OAE and/or click AER results should occur as soon as possible, preferably within the same appointment or as soon as possible after medical approval for continued evaluation is obtained as necessary.
- E. Completion of the diagnostic test battery shall not be delayed beyond three months of age for treatment of middle ear effusion. Bone conduction AER procedures should be used as a means to confirm hearing status if a continuing middle ear condition is present.
- F. Fax or otherwise transmit the confirmation of hearing status including diagnosis within two days of determination to the Children's Medical Services Newborn Screening Program using (a) the [Repeat Hearing Screen Form](#) if screening tools were used; or (b) the [Diagnostic Hearing Evaluation Form](#) if diagnostic tests were used.
- G. Children up to age 36 months with apparent late onset or late diagnosed hearing loss shall be referred to the Children's Medical Services Newborn Screening Program the [Diagnostic Hearing Evaluation Form](#) if the child was identified with hearing loss.
- H. Refer children under age 36 months who have confirmed hearing loss to the local Early Steps office within two days of confirmation. The audiologist shall not wait until complete audiometric information (frequency specific responses) has been obtained to refer the child to Early Steps.
- I. Strive to ensure that amplification (loaner or purchased) is fit to children with hearing loss within 30 calendar days of confirmation of hearing loss as appropriate to meet the needs of the child and family.

Resources

Contact Information

Children's Medical Services Newborn Screening Follow-Up Program

Mailing address:

4052 Bald Cypress Way, Bin A06
Tallahassee, FL 32399-1707

Physical address:

4025 Esplanade Way
Tallahassee, FL 32311

Phone numbers:

Main phone number: (850) 245-4201
Toll-free Metabolic Line: (866) 804-9166
Toll-free Hearing Line: (866) 289-2037
Secure Metabolic Fax Line: (850) 922-5385
Secure Hearing Fax Line: (850) 245-4049

Website address:

http://doh.state.fl.us/cms/Newborn_Screeningreen.html

Department of Health

Bureau of Laboratories

Physical address (For mailing specimens):

1217 Pearl Street
Jacksonville, FL 32202

Mailing address (For correspondence only—do not use for mailing specimens as it will delay sample processing):

PO Box 210
Jacksonville, FL 32231

Phone numbers:

Main phone number: (904) 791-1500
Customer service: (904) 791-1645, 791-1644, 791-1646, 791-1647
Fax number: (904) 791-1671

Children’s Medical Services Area Offices

Northwest Florida Region – Pensacola and Panama City Area Offices	
Pensacola—Escambia, Okaloosa, Santa Rosa, Walton Counties	Area Office 5192 Bayou Boulevard Pensacola, FL 32504 (800) 381-3685 (850) 484-5040 Fax: (850) 484-5189
Panama City—Bay, Calhoun, Gulf, Holmes, Jackson, Washington Counties	Area Office 230 N. Tyndall Parkway Panama City, FL 32404 (800) 299-4700 (850) 872-4700 Fax: (850) 872-4817
Tallahassee Big Bend Region – Tallahassee Area Office	
Tallahassee—Franklin, Gadsden, Jefferson, Leon, Liberty, Madison, Taylor, Wakulla Counties	Area Office 2390 Phillips Road Tallahassee, FL 32308 (800) 226-2604 (850) 487-2604 Fax: (850) 922-2123
North Central Florida Region – Gainesville Referral Center, and Gainesville, Ocala, Jacksonville and Daytona Area Offices	
Gainesville – Alachua, Bradford, Columbia, Dixie, Gilchrist, Hamilton, Lafayette, Levy, Putnam, Suwannee, Union	Area Office 1701 S.W. 16th Avenue, Bldg. B Gainesville, FL 32608 (800) 523-7545 (352) 334-1400 Fax: (352) 334-1476
Ocala – Citrus, Hernando, Lake, Marion, Sumter Counties	Area Office 1515 E. Silver Springs Blvd., Suite 215 Ocala, FL 34470 (888) 326-7485 (352) 369-2100 Fax: (352) 369-2134
Jacksonville – Baker, Clay, Duval, Nassau, St. Johns Counties	Area Office 910 North Jefferson St. Jacksonville, FL 32209-6810 (800) 340-8354 (904) 360-7070 Fax: (904) 798-4568
Tampa Bay Region – Tampa, St. Petersburg and Lakeland Area Offices	
Tampa – Hillsborough County	Area Office 13101 N. Bruce B. Downs Blvd. Tampa, FL 33612

	(813) 396-9743 Fax: (813) 396-9795
St. Petersburg – Pasco, Pinellas Counties	Area Office 3251 3rd Avenue, North, Suite 130 St. Petersburg, FL 33713 (800) 336-1612 (727) 893-2775 Fax: (727) 893-2484
Lakeland – Hardee, Highlands, Polk Counties	Area Office 4718 Old Highway 37 South Lakeland, FL 33813 (800) 741-2250 (863) 701-1151 Fax: (863) 701-1161

Central Florida Region – Orlando and Viera Area Offices	
Orlando – Orange, Osceola, Seminole Counties	Area Office 7000 Lake Ellenor Drive Orlando, FL 32809 (800) 226-6530 (407) 858-5555 Fax: (407) 856-6558
Viera – Brevard County	Area Office 2565 Judge Fran Jamieson Way Viera, FL 32940 (321) 639-5888 Fax: (321) 690-3887

Southeast Florida Region – Ft. Lauderdale, West Palm Beach and Ft. Pierce Area Offices	
Ft. Pierce – Indian River, Martin, Okeechobee, St. Lucie Counties	Area Office 1701 South 23 rd Street Ft. Pierce, FL 34950-4804 (800) 226-1354 (772) 467-6000 Fax: (772) 467-6092
West Palm Beach – Palm Beach County	Area Office 5101 Greenwood Avenue West Palm Beach, FL 33407 (877) 822-5203 (561) 881-5040 Fax: (561) 881-5075
Ft. Lauderdale – Broward County	Area Office Broward General Medical Center 1625 SE 3rd Avenue, Suite 415 Ft. Lauderdale, FL 33316 (800) 204-2182 (954) 713-3100 Fax: (954) 713-3179

Southwest Florida Region – Sarasota, Ft. Myers, and Naples Area Offices	
Sarasota – Charlotte, Desoto, Manatee, Sarasota Counties	Area Office 6055 Rand Blvd. Sarasota, FL 34238-5189 (800) 235-9717 (941) 361-6250 Fax: (941) 361-6272
Ft. Myers – Glades, Hendry, Lee Counties	Area Office 9800 South Healthpark Drive, Suite 405 Ft. Myers, FL 33908 (800) 226-3290 (239) 433-6723 Fax: (239) 433-9739 Primary Care
Naples – Collier County	Area Office 1665 Medical Boulevard Naples, FL 34110 (239) 513-7400 Fax: (239) 513-7435 (239) 261-8877 (Immokalee)

South Florida Region – Miami North and South and Marathon Area Offices	
Miami N – North Dade County	Area Office 155 S. Miami Ave, Suite 1000 Miami, FL 33130 (866) 831-9017 (305) 349-1330 Fax: (305) 349-1331
Miami S - South Dade County	Area Office 17615 S. W. 97th Avenue, Bldg. 1 Miami, FL 33157 (786) 624-5700 Fax: (786) 624-5790
Marathon - Monroe County	Area Office 10015 Overseas Highway 100 th Street Center Marathon, FL 33050 (800) 342-1898 (305) 289-2779 Fax: (305) 289-2781

Referral Center Contact Information

Disorder	Referral Center	Phone
Genetic Disorders: <ul style="list-style-type: none"> Galactosemia Biotinidase Deficiency MS/MS disorders 	University of Florida Genetic Center Gainesville	(352) 294-5050
	University of South Florida Genetic Center & All Children's Hospital Tampa/St. Petersburg	(727) 767-4237 Ext. 3720
	University of Miami Genetic Center Miami	(305) 243-6006
Endocrine Disorders: <ul style="list-style-type: none"> Congenital Hypothyroid Congenital Adrenal Hyperplasia 	University of Florida Endocrine Center Gainesville	(352) 334-1390
	University of South Florida Endocrine Center All Children's Hospital Tampa/St. Petersburg	(727) 767-3588
	University of Miami Endocrine Center Miami	(305) 243-2920
Hemoglobinopathy Disorders: <ul style="list-style-type: none"> Sickle Cell Anemia Sickle Beta Thalassemia Hemoglobin SC Disease, etc. 	Nemours Children's Clinic Pensacola	(850) 505-4708
	University of Florida Gainesville	(352) 265-0111 Ext. 49680
	Shands-Jacksonville Hospital & Nemours Children's Clinic Jacksonville	(904) 697-3789
	All Children's Hospital St. Petersburg	(727) 767-4221
	University of South Florida (Sickle Cell Referral only) Tampa	(813) 396-9725
	Orlando Regional Healthcare System Arnold Palmer Hosp. for Women and Children Orlando	(321) 843-9977
	Florida Hospital Orlando	(407) 303-2080
	University of Miami Miami	(305) 243-6924 press #1
	Memorial Regional Hospital Joe DiMaggio Children's Hospital Broward General Medical Center Hollywood/Ft. Lauderdale	(954) 713-3140
	Lee Memorial Health System Ft. Myers	(239) 343-5333

Disorder	Referral Center	Phone
Cystic Fibrosis	Nemours – Pensacola Pensacola	(850) 505-4785
	Nemours – Jacksonville Jacksonville	(904) 697-3408
	University of Florida Gainesville	(352) 273-8380
	Nemours – Orlando Orlando	(407) 650-7539
	University of South Florida Tampa	(813) 396-9774
	All Children’s Hospital St Petersburg	(727) 767-3995
	St Mary’s Medical Center West Palm Beach	(561) 840-6065
	Joe DiMaggio Children’s Hospital Hollywood	(954) 265-3665
	Miami Children’s Hospital Miami	(305) 662-8380 ext. 8
	University of Miami Miami	(305) 243-6162
	HealthPark / Lee Memorial Hospital Ft. Myers	(239) 466-1243

Additional Resources

Florida Newborn Screening Program:

http://doh.state.fl.us/cms/Newborn_Screeningcreen.html

Florida Newborn Screening Results:

<https://www.fnsr.net>

CMS Kids:

<http://www.cms-kids.com/>

Early Steps:

<http://www.cms-kids.com/earlysteps/index.html>

National Newborn Screening and Genetics Resource Center:

<http://genes-r-us.uthscsa.edu/index.htm>

Southeastern Regional Genetics Group (SERGG):

<http://www.sergginc.org/>

Centers for Disease Control and Prevention (CDC):

<http://www.cdc.gov/nceh/dls/newborn.htm>

March of Dimes (homepage):

<http://www.marchofdimes.com/home.asp>

Emory Newborn Screening

http://www.genetics.emory.edu/newborn_screening/

Early Steps Contact List

cms-kids.com/home/contact/earlysteps.pdf

AG Bell Florida

www.agbellflorida.org

Florida chapter of the Alexander Graham Bell Association for the Deaf and Hard of Hearing, which advocates independence through listening and talking.

Clearinghouse Information Center

Florida Department of Education

Room 628 Turlington Building

Tallahassee, FL 32399-0400

850-245-0477

www.fldoe.org/ease/clerhome.asp

Materials about children ages birth through 4 focusing on child development, early intervention, education, disabilities, developmental delays, health and related topics.

Children's Medical Services

www.cms-kids.org

Provides children with special health care needs a family centered, managed system of care.

CMS-Early Steps State Office

(800) 654-4440

http://cms-kids.com/families/early%5Fsteps/early_steps.html

Policy and public reporting information on Florida's early intervention system, Early Steps.

Florida Hands & Voices

(866) 422-0422

<http://www.flhandsandvoices.org>

Florida's chapter of the nationwide Hands & Voices, a non-profit organization dedicated to supporting families and their children who are deaf or hard of hearing, as well as the professionals who serve them.

Florida Kidcare

www.floridakidcare.org

Health insurance for Florida children from birth through age 18.

Florida Newborn Hearing Screening Program

(866) 289-2037

www.doh.state.fl.us/Cms/nbscreen-hearing.html

Florida's newborn hearing screening page with statewide data and required forms/procedures.

Florida Outreach Project

University of Florida

PO Box 100234

Gainesville, FL 32610

(352) 846-2757 V (352)846-2759 TTY

www.deafblind.ufl.edu

Assists families aged birth through 21 who have both hearing loss and vision disabilities by promoting the full inclusion and participation of persons with deaf-blindness as active members of their communities.

Florida School for the Deaf and Blind

(800) 344-3732

www.fldb.k12.fl.us

Florida's public boarding school for eligible students who are deaf or hard-of-hearing, or blind or visually impaired students in preschool through grade 12.

Florida Speech-Language-Hearing Association

www.flasha.org

Serves the needs of Florida professionals by providing support, opportunities for professional growth, and public awareness and advocacy of issues related to the highest quality care for the individuals they serve.

Florida's Alliance for Assistive Services and Technology

(850) 487-3278 or (888) 788-9216

www.faast.org

Provides hands on assistive technology demonstrations and training, financing for assistive technology purchases, assistive device lending programs, community outreach to rural and underserved groups, and advocacy and education on consumer choice.

Resource Materials and Technology Center for the Deaf and Hard of Hearing

207 N. San Marco Ave.
St. Augustine, FL 32084
(904)827-2666

www.fsdb.k12.fl.us/rmc

A specialized FDLRS center serving teachers of the deaf and hard of hearing throughout the state of Florida.

Assistance with Achieving Results in Education (AWARE) Project

1021 Delaware Ave.
Palm Harbor, FL 34683-3529
(888) 612-9273

<http://www.cflparents.org>

Florida's Technical Assistance ALLIANCE for Parent center, federally funded parent centers that provide a variety of services including one-on-one support and assistance, workshops, publications, and web sites. Serves 30 counties in central and northeast Florida