FLORIDA NEWBORN SCREENING

2022 Protocols

March 15, 2022

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Florida HEALTH

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Introduction

Newborn screening (NBS) is a highly successful population-based public health program which identifies certain genetic, endocrine, hemoglobinopathy, immunology, and metabolic conditions and provides for early identification, management, and follow-up for those affected. NBS programs are in place in every state and in many countries throughout the world. Many of the conditions are rare and may not be diagnosed before the onset of symptoms, which may, in some cases, be too late to prevent health issues.

In Florida, infant blood is collected at or near the time of birth on a filter paper card and submitted to the Bureau of Public Health Laboratories (BPHL)-Jacksonville for testing. Specimens are handled in strict accordance with 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 Health Care Administration (AHCA). Screening is not mandatory, but strongly encouraged. Parents/guardians may opt out of screening by providing a signed declination form for documentation and maintained in the infant's medical record.

Purpose

The purpose of the Florida Department of Health (DOH) NBS Program is to facilitate prevention of developmental impairments, delayed physical growth, severe illness, and death through early detection and intervention. With appropriate and timely treatment, infants identified through the NBS Program have the opportunity to grow and reach optimal potential. The Florida NBS Program promotes newborn screening for all newborns in Florida.

Goals

The goals of the NBS Program are:

- To maintain the quality of screening tests used in the BPHL-Jacksonville.
- To provide the required materials needed to collect NBS specimens.
- To ensure the availability of timely NBS results.
- To promote education regarding prevention and management of conditions screened by the NBS Program.
- To ensure all affected infants receive appropriate confirmatory testing, counseling, and initiation of treatment after identification.

The NBS Program is a comprehensive system with the following partners: BPHL-Jacksonville within the Division of Disease Control and Health Protection, NBS Follow-up Program and NBS Hearing Program within the Division of Children's Medical Services (CMS) in Tallahassee, Genetics and Newborn Screening Advisory Council (GNSAC), referral centers, birthing facilities, and healthcare providers.

Limitations of Screening Tests

The NBS Program identifies infants at risk for conditions included on the Florida NBS panel using laboratory analyses. Infants in need of more definitive testing are referred to an

appropriate referral center for confirmatory testing. 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 treatments. It is imperative health care providers remain watchful for any signs or symptoms of conditions in patients and follow up as needed. Confirmatory testing should always be performed to either confirm or rule out a diagnosis.

Program History

1965 - The NBS Program began in Florida with the passage of section 383.14, Florida Statutes (F.S.). This required the Florida Board of Health to promote the testing of all newborns for Phenylketonuria (PKU). At the time, 20% of testing was performed by hospitals and 80% was performed by state laboratories in Jacksonville and Miami.

1978 - Congenital Hypothyroidism (CH), Maple Syrup Urine Disease (MSUD), and Galactosemia were added to the NBS panel. An Infant Screening Advisory Council (later renamed the Genetics and Newborn Screening Advisory Council [GNSAC]) was established and regulations were set up regarding follow-up, diagnosis, and treatment of infants with abnormal results.

1979/1980 - BPHL-Jacksonville was designated as the only newborn screening testing site.

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

January 1985 - MSUD was removed due to the lack of any detected cases in 500,000 births.

August 1988 - Hemoglobinopathies were added to the NBS panel.

1993 - Funding of NBS services changed from general revenue to fees collected from hospitals and birthing centers. A fee of $20.00 per live birth was charged to facilities with over 60 births and up to 3,000 births was established in section 383.14, F.S.

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

April 1995 - Congenital Adrenal Hyperplasia (CAH) was added to the NBS panel.

October 1, 2000 - Newborn hearing screening was mandated per section 383.145, F.S.

May 2002 - The NBS Task Force was created by House Bill 817 to evaluate the NBS Program, make recommendations for improvements, and consider expansions to the panel to include conditions recommended by March of Dimes and the American College of Medical Genetics (ACMG).

July 1, 2004 - The NBS fee was reduced to $15 per live birth and the exemption and caps were removed. DOH was given authority to bill third party payers for NBS testing. BPHL-Jacksonville and CMS were given authority to release NBS results to primary care providers.

October 1, 2005 - Biotinidase Deficiency was added to the NBS panel.

January 9, 2006 - Implementation of tandem mass spectrometry (MSMS) added 25 new conditions to the NBS panel including amino acidemias, organic acidemias, and fatty acid oxidation disorders. MSUD, which had been removed in 1985, was included in this expansion. The NBS Program now screened for 34 conditions.

January 26, 2007 - The GNSAC recommended changing the minimum age at collection from 48 hours to 24 hours. The need for a protein feeding at least 24 hours prior to collection was

kept.

September 17, 2007 - Cystic Fibrosis (CF) was added to the NBS panel. Florida now screened for a total of 35 conditions, which satisfied recommendations by March of Dimes and ACMG.

January 5, 2009 - The Florida Newborn Screening Results (FNSR) website went live, allowing providers to create an account and access NBS results online.

January 28, 2011 - The GNSAC recommended the addition of Severe Combined Immunodeficiency Disease (SCID) to the NBS panel.

January 20, 2012 - The GNSAC recommended working with the Cardiac Subcommittee of the CMS Advisory Council for recommendation of implementing Critical Congenital Heart Disease (CCHD) to the scope of the NBS Program.

October 1, 2012 - SCID was added to the NBS panel, which now included 36 conditions.

December 2013 - Statewide implementation of CCHD reporting began. The addition of CCHD and a change to count secondary conditions resulted in the Florida NBS Program screening for 31 core conditions and 22 secondary conditions, for a total of 53 conditions. Fifty of the conditions were on the national Recommended Uniform Screening Panel (RUSP).

February 6, 2015 - The GNSAC voted to remove the protein feeding requirement for NBS specimen collection.

February 2016 - The CF DNA testing panel was expanded to a total of 60 CF variants.

February 19, 2016 - The GNSAC recommended the addition of X-linked Adrenoleukodystrophy (X-ALD) to the NBS panel.

July 1, 2017 - Timelines for the consideration of addition of new conditions connected to the RUSP were instituted by a statutory mandate. The GNSAC must consider whether a condition should be included on Florida's NBS panel within one year of being added to the RUSP. If recommended for addition, the NBS Program must begin screening for the condition within 18 months, pending availability of an appropriate screening method for the state.

May 1, 2018 - X-ALD was added to the NBS panel, which now included 54 conditions.

August 24, 2018 - The GNSAC voted to add Pompe Disease and Mucopolysaccharidosis I (MPS I) to the NBS panel.

February 15, 2019 - The GNSAC voted to add Spinal Muscular Atrophy (SMA) to the NBS panel.

July 9, 2019 - A fully redesigned FNSR site was launched, allowing for a more user-friendly experience for providers to access NBS results.

February 3, 2020 - Pompe Disease and MPS I were added to the NBS panel, for a total of 56 conditions.

April 27, 2020 - SMA was added to the NBS panel, for a total of 57 conditions.

November 15, 2021 - The CF DNA testing panel was expanded to a total of 74 CF variants.

Protocols

Protocols for this program were established in 1981 and expanded in 1984, 1992, 1999, 2012, and 2022. These protocols set the standards for the Florida NBS Program.

Legal Authority

Sections 383.14, 383.145, and 383.15, F.S., and Chapter 64C-7, Florida Administrative Code (F.A.C.).

Registry

Section 383.14(3)(d), F.S. states DOH shall maintain a confidential registry of cases, including the information of importance for the purposes of follow-up services to prevent intellectual disability, to correct or ameliorate physical disabilities, and for epidemiologic studies, if indicated.

Confidentiality

NBS is an activity described in its capacity as a public health authority as defined by the Health Insurance Portability and Accountability Act (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 DOH's rules on privacy and confidentiality. Questions regarding confidentiality may be answered by contacting the NBS Follow-up Program at (850) 245-4201 or the BPHL-Jacksonville at (904) 791-1500.

Parental Refusal

Section 383.145, F.S., requires that all newborns must have a specimen collected before discharge from care, unless a parent or guardian signs a written statement of refusal. A copy of this refusal should be filed in the newborn's medical record and be available to the NBS Program upon request.

If any portion of the newborn screen is refused, a specimen card should be sent to BPHL-Jacksonville with the appropriate "Refused" bubble filled in on the specimen card. All

demographic fields must be completed on all specimen cards submitted, including those marked as refusal, to allow for proper linking of multiple specimens.

Quality Improvement Benchmarks

Blood spot collection:

- Less than 1 percent of specimens received by BPHL-Jacksonville are unsatisfactory for testing.
- At least 80 percent of specimens should be received at BPHL-Jacksonville no later than three days after collection. To achieve this, specimens should be shipped within 24 hours of collection to the BPHL-Jacksonville via overnight delivery.

CCHD screening:

- At least 90 percent of specimens submitted must have appropriate CCHD screening data included on the specimen card.

Hearing screening:

- At least 95 percent of all infants are screened by one month of age at the hospital or birthing facility (age correction for preterm infants is acceptable).
- Less than 4 percent of all infants do not pass the initial (two high-quality hearing screens) hospital-based screenings and require subsequent outpatient rescreening.
- Less than 4 percent of all infants do not pass the initial hearing screening and fail any subsequent rescreening prior to referral for outpatient comprehensive audiological evaluation.
- All infants receive a newborn hearing screening prior to discharge from care.
- All hearing screen results are reported to the state Early Hearing Detection and Intervention (EHDI) Program.

Responsibilities

GNSAC

The GNSAC serves as an advisory body to DOH regarding medical and procedural aspects of the Florida NBS Program. Recommendations for program changes are made by the GNSAC to the DOH.

GNSAC members are appointed by the State Surgeon General of DOH. The GNSAC comprises of 15 members including the following:

- The State Surgeon General or his/her designee.
- The CMS Deputy Secretary or his/her designee.
- One representative from 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 NBS programs (e.g. the March of Dimes).
- One individual representing audiologists.
- Four representatives from medical schools in the state.
- Two consumers.

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

GNSAC's purpose is to advise DOH regarding:

- Conditions which should be added to Florida's screening panel.
- Procedures for collecting and transmitting specimens and recording such results.
- Methods by which NBS may more effectively provide services to the children of Florida.

BPHL-Jacksonville

- Perform tests and release results to the submitting entity using established procedures, such as United States Postal Service (USPS), Auto-Fax, or electronic delivery.

- Generate reports regarding screening tests, information errors on specimen cards, and other information as needed.

- Advise the NBS Follow-up Program of laboratory-related problems and recurring information errors on specimen cards.

- Respond to inquiries from submitting entities concerning laboratory results.

- Perform testing six days per week, Monday–Saturday, except for designated state holidays.

- In the event of activation of an emergency (i.e. hurricane, etc.), specimen cards will be directed to the designated back-up laboratory for immediate processing as determined by the BPHL-Jacksonville.

- Collect the newborn screening fee ($15 per live birth) from birthing facilities and bill third-party payers for newborn screening testing.

- Be the custodian for the DOH policy regarding specimen retention and storage.

NBS Follow-up Program

- Notify an appropriate contracted DOH Referral Center of infants identified with a presumptive positive screening result as established by internal protocols, Monday–Friday, excluding state holiday closures. All presumptive positive results identified as Time Critical will be referred on Saturdays.

- Monitor and track infants with borderline and presumptive positive screening results. Ensure repeat screenings, status confirmations, confirmatory testing results, and diagnostic case reports are obtained from Referral Centers.

- Contact the infant's primary care provider regarding borderline results and recommendation for repeat testing. Contact the family if the primary care provider is unknown.

- Mail hemoglobinopathy trait, unsatisfactory specimen, referred hearing, borderline thyroid, and other letters generated by the data system to parents/guardians and providers on record for notification of NBS outcomes.

- Respond to inquiries from providers related to obtaining screening results and assist in utilizing information to avoid unnecessary repeat testing.

- Update case status as case reports are received from Referral Centers and report to state and national databases as requested.

- Assist parents/guardians with facilitating diagnostic hearing testing.

- Notify submitter administration of recurring inaccurate or incomplete information on specimen cards.

- Notify submitting entities, via the website (www.floridanewbornscreening.com), of statistics regarding timeliness of shipping specimens to the BPHL-Jacksonville and unsatisfactory collection rates.

- Develop and provide educational materials for birthing facilities, health care providers, and parents/guardians about the NBS Program and other pertinent information.

- Provide on-site technical assistance training to birthing and collection facilities.

- Provide reports to the public regarding the NBS Program, as requested.

- Assist with registering users for www.FNSR.net access.

DOH Contracted Referral Centers (Genetics, Endocrine, Hematology, SCID, & CF)

- Receive and accept all referrals from the NBS Follow-up Program for infants with presumptive positive screening results or infants with multiple borderline results as established in the NBS Follow-up Program internal protocols.

- Contact the primary care provider (or parent if provider is unknown) to coordinate immediate evaluation, confirmatory testing, and treatment, if necessary.

- Contact the parent/guardian.

    - If no address is available, contact the collection facility for assistance with locating the family.

    - If all contact attempts fail, contact the NBS Follow-up Program for assistance.

    - If handling a time critical condition and there is no response to contact attempts, local law enforcement may be sent to the address on record for assistance. Call local law enforcement and ask for the Watch Commander or equivalent to assist.

- Arrange for confirmatory testing and other medically necessary actions for infants referred.

- Provide appropriate counseling and education to families regarding the course of treatment and diagnosis.

- Report changes in address, location, or contact information to NBS Follow-up Program.

- Provide the NBS Follow-up Program with a completed case report within 15 days of confirmation/diagnosis, including the name and address of the primary care provider and a copy of the confirmatory laboratory results.

Birth and Collection Facilities

All birth and collection facilities are mandated to:

- Collect high quality specimens. (See Specimen Collection section for more information.)

- Complete all demographic portions of every specimen submitted, including insurance information. (See Specimen Collection section for more information.)

- Report final hearing screening results or NICU not screened (until baby is medically ready for a hearing screening) on the specimen card, web portal, or through eReports.

- Report CCHD screening information on the specimen card or through the web portal.

- Document collection of the specimen in the infant's medical record, as well as any specimen collection log maintained by the facility.

- Ship specimens within 24 hours of collection via overnight delivery, to be received by the BPHL-Jacksonville no later than three days after collection.

- Document collection and monitor shipment of specimens, as well as the receipt of the shipped specimen by BPHL-Jacksonville. It is recommended that a photocopy be made of every specimen included in a shipment to be kept with shipping documentation to serve as a manifest for the facility's records.

- Designate an NBS coordinator or contact person to ensure efficient communication with the BPHL-Jacksonville and the NBS Follow-up Program. This person must ensure all infants obtain a valid newborn screen per protocols.

- Ensure a valid specimen is obtained. A valid specimen is one which is collected after 24 hours of age and is satisfactory for testing.

- Follow up on unsatisfactory specimens. Unsatisfactory specimens will not be tested by BPHL-Jacksonville, thus delaying a potentially fatal diagnosis.
  NOTE: The submitting entity must ensure a satisfactory specimen is collected and resubmitted as soon as possible following first specimen collection protocols.

- Designate a contact person and provide current contact information when requesting test results from BPHL-Jacksonville to avoid delays in processing and reporting.

- Monitor specimen cards for supply levels and expiration dates to ensure valid cards are utilized. Large facilities will receive an automatic shipment of specimen cards on a quarterly basis.

- Provide verbal and written information about the NBS Program to parents/guardians of all infants born in Florida.

- Provide the BPHL-Jacksonville and NBS Follow-up Program with information when contacted.

- Include all NBS results in the infant's medical record.

- Forward a copy of the NBS results and any follow-up information to the primary care provider.

Non-Hospital Birth Providers

All responsibilities listed above for birth and collection facilities also apply to non-hospital birth providers.

All non-hospital birth providers are additionally mandated to:

- Arrange for the collection of NBS blood specimens and completion of CCHD screening as soon after 24 hours of age as possible.

- Instruct parents or caregivers on the importance of having the hearing screening performed and provide information to assist with obtaining any needed referrals within three months after the child's birth.

Primary Care Providers

All Primary Care Providers are mandated to:

- Ensure all infants in the practice have a valid newborn screen, including hearing and CCHD results, in the medical record.

- 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 screen results are normal.**

- Contact the NBS Follow-up Program if unable to locate results, or if there is a question about results.

- Provide information regarding changes in address, phone/fax number, or contact person to the NBS Follow-up Program to prevent inaccurate or failed delivery of NBS results.

- Assist contracted DOH Referral Centers and the NBS Follow-up Program with locating an infant and/or providing continuing care.

Specimen Collection

Completing the Specimen Card

Verify the expiration date and integrity of the card before completing demographics and collecting a specimen.

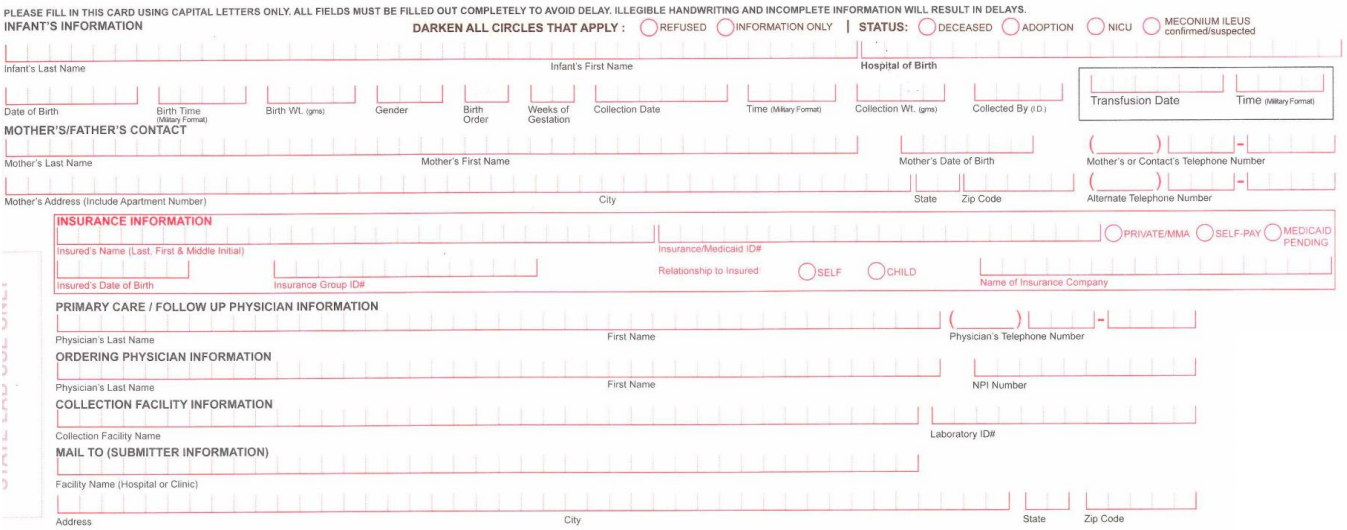
It is important to fill out the specimen card completely, accurately, and legibly.

All fields are required on every specimen card as the information provided is necessary to accurately interpret screening results. Information is also imperative for linking multiple records together to determine the proper care plan for an infant.

CRITICAL FIELDS:

BPHL-Jacksonville **cannot report results** if any of the following fields are missing:

- Infant's Name
- Date and Time of Birth
- Date and Time of Collection
- Birth and Collection Weights



Circles Along the Top: Bubble in all that apply.

Refused: Bubble in and complete the demographics if the parent/guardian refuses NBS collection. Submit the specimen card to BPHL-Jacksonville.

Information Only: Bubble in and complete demographics if using the specimen card to report information (i.e. hearing or CCHD screening results) but no blood is collected. Submit to BPHL-Jacksonville.

Deceased: Bubble in and complete demographics if the infant is deceased. Submit to BPHL-Jacksonville.

Adoption: Bubble in "Adoption" only if the infant is being adopted. (See Mother's/Father's Contact Information section below.)

NICU: Bubble in if the infant is in the NICU (or any ICU) at the time of collection.

Meconium Ileus (confirmed/suspected): Bubble in only if it applies to the infant. This information will trigger additional CF testing.

Infant's Name: Use the naming convention selected by the collecting facility for compliance with The Joint Commission. The infant's legal name may also be used.

Hospital of Birth: Write out the complete name of the birth facility, **no abbreviations**. If born at home, list "Home Birth."

Dates of Birth and Collection: Enter in MM-DD-YYYY format. **Accuracy is critical.**

Birth and Collection Times: All times should be reported in military time. **Accuracy is critical.**

Example:
2:16 AM = **0216**
2:16 PM = **1416**

Correct dates and times are imperative as some results are affected by age at collection.

Birth and Collection Weights: Report weights in **grams only**. **Accuracy is critical.** Weights can affect the interpretation of results and the care plan for the infant.

Gender: Enter M for male, F for female, or U for unknown.

Birth Order: If an infant is one of a set of multiples (twins, triplets, etc.), it is imperative the birth order be reported on every specimen. **Do not list a birth order for singletons.** Listing a birth order affects timely reporting of results due to the need to verify accuracy.

Weeks of Gestation: List the infant's gestational age at birth, not adjusted age.

Transfusion Date and Time: If an infant has **not** been transfused, leave this field blank. Transfusions may require repeat testing and invalidate certain test results.

Collected By: Complete this field per collection facility's protocol (i.e. initials, lab number, etc.).

Mother's/Father's Contact Information: Complete all sections accurately. This information should be verified by the parent/guardian as information may be different than what is in the medical record. **Accuracy is critical as this information is used to contact families in the event of an abnormal result.**

Last and First Name: Ensure accurate spelling.
Mother's Date of Birth: Ensure accuracy.
Address: Report the current address, ensuring ZIP code, city, and unit number (i.e. apartment or lot number). Only use abbreviations approved by USPS.
Contact and Alternate Phone Numbers: Include area codes. Alternate number may be an additional parent or family member. Phone numbers are used if follow-up is needed.

****Adopted infants:** Provide adoptive parent's information, if known. List the name and contact information of the attorney/adoption agency if the adoptive parent's information is unknown. Ensure the "Adoption" bubble is marked.

Please note, if a specimen has been marked as "Adoption," results will not upload to www.FNSR.net. Contact the NBS Follow-up Program for assistance obtaining results.

****Infants in foster care:** Provide the name of the agency/case worker and phone number in the "Mother's/Father's Contact" field. If using the Florida Department of Children and Families

(DCF) case worker's name, include DCF beside the last name. For example, "Smith, DCF."

Insurance: Medicaid or private insurance information should include the name of the insurance company, group number, and policy number. All Medicaid or private insurance fields should be completed. A printed face sheet may be attached to the demographics portion of the specimen card to provide insurance information only. If submitting a face sheet, write "See face sheet" in the insurance section.

Primary Care/Follow-Up Physician Information: List the name and phone number of the provider who will be seeing the infant after discharge. If the parent/guardian is unsure of the name of the provider, list the clinic/facility name.

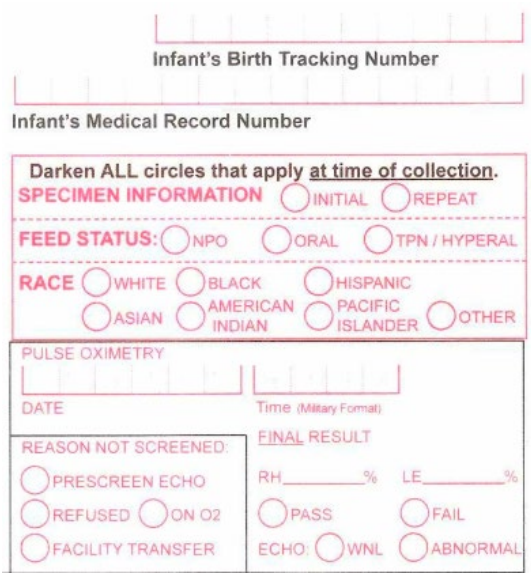
Ordering Physician Information: List the provider who ordered the newborn screen.

NPI Number: List the National Provider Identifier (NPI) of the provider who ordered the newborn screen.

Collection Facility Information: Write out the full name of the facility, **no abbreviations**. If collected in the home for a home birth, list the name of the midwife or home birth service.

Laboratory ID#: This is a facility-specific number. Complete per collection facility protocol.

Mail-To (Submitter Information): Location where results should be sent.
Facility Name: Write out the full name of the facility, **no abbreviations**.
Address: Provide full address. Use only USPS approved abbreviations.



Infant's Birth Tracking Number: List the birth tracking number. The tracking number is available from Vital Statistics/Birth Records in each facility. This information allows for linking of infant's records and directs follow-up efforts.

Infant's Medical Record Number: The infant's medical record number must be accurately reported. Errors in the medical record numbers may cause the BPHL-Jacksonville to deem a specimen "Unsatisfactory Due to Information Mismatch."

Specimen Information: Fill in as many bubbles as are appropriate.

Initial or Repeat: Fill in "Initial" for the infant's first specimen, "Repeat" for all subsequent specimens.

NPO, Oral, TPN/Hyperal: *Feed status is very important.* TPN/Hyperal can affect results and impact the plan of care for the infant.

Race: Fill in as many bubbles as are appropriate.

Pulse Oximetry: Report CCHD screening result here. ***Report only the final screening result.***

Date: MM/DD/YYYY format

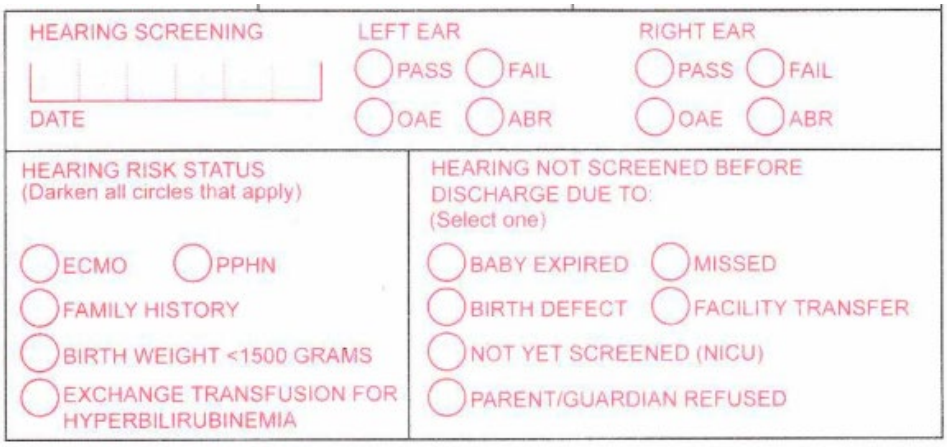
Time: Military format

RH% and LE%: Oxygen saturation results must be reported if reporting a Pass or Fail.

Pass/Fail: Refer to the CCHD screening algorithm for determination of Pass or Fail.

Echo: If infant fails CCHD screening, an echocardiogram (echo) is indicated per CCHD algorithm. Fill in the appropriate bubble with echo outcome.

Reason Not Screened: Select appropriate reason.



Hearing Screening:

Date: MM/DD/YYYY format.

Left Ear and Right Ear: Pass/Fail and OAE/ABR must be completed for both ears.

Hearing Risk Status: Fill in as many bubbles as are appropriate.

Hearing Not Screened Before Discharge: Fill in as many bubbles as are appropriate.

Specimen Collection Protocols

Term or Well Nursery

A valid specimen is defined as one that is collected after the infant is 24 hours of age and is satisfactory for testing.

If an infant is discharged to home prior to 24 hours of age, a specimen must be collected as close to discharge as possible. The birth facility must provide the parent/guardian with a written notification that a repeat specimen must be collected by five days of age. The clinical significance of certain laboratory results cannot be determined if the infant was less than 24 hours of age at the time of collection.

Specimens not meeting the requirements of a valid specimen will require a repeat specimen. The family must be given the option to return to the birth facility or arrangements must be made to ensure a repeat specimen can be obtained at the infant's physician's office. **It is the birth facility's responsibility to ensure the repeat specimen is obtained.**

Transfusions

A newborn screening specimen should be collected prior to the first transfusion. If an infant was transfused prior to the first specimen, a repeat specimen must be collected no sooner than120 days after the last transfusion. Transfusions prior to blood collection invalidate screening results for hemoglobinopathies, biotinidase, and galactosemia.

NOTE: The screening results for hemoglobinopathies, biotinidase, and galactosemia are valid prior to transfusion, regardless of age at collection. If a satisfactory specimen was collected prior to transfusion, a repeat specimen no sooner than 120 days after transfusion is not necessary.

Transported or Transferred Infants

If an infant is transferring to another facility, collect a specimen prior to transfer, regardless of age. If specimen collection before transfer is not possible, document and inform the receiving facility that a specimen was not collected. The receiving facility is responsible for obtaining a specimen upon admission. Regardless of time of transfer, the facility where the infant was born should be listed as the hospital of birth on the specimen card.

Critically Ill and Premature Infants (ICU)

All infants requiring neonatal intensive care unit (NICU) or additional intensive care unit (i.e. CVICU, PICU) services must use "Newborn Screening Guidelines for Preterm, Low Birth Weight, and Sick Infants" as put forth by the Clinical and Laboratory Standards Institute (CLSI). Specimens must be collected:

- **Upon admission** to the NICU/ICU, regardless of age and ideally before the first transfusion and/or other treatments (such as parenteral feeding), to serve as a baseline for certain conditions.
- **At 48–72 hours of life**, for infants initially tested prior to 24 hours of age at first screen,

or any infant with an abnormal first screen.

- **At 28 days of life, or before discharge, whichever comes first,** for infants < 34 weeks gestational age, or weighing < 2,000 grams at birth. This third specimen is at the discretion of the ordering physician for infants weighing ≥ 2,000 grams.
- Results of previous specimens should be reviewed prior to discharge.

Repeat Specimens

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

- The initial specimen was collected prior to 24 hours of age.
- The initial/previous specimen was unsatisfactory for testing.
- The initial/previous specimen had borderline results and the recommendation indicated a repeat must be obtained.
- The infant was transfused prior to the collection of the initial specimen. A repeat should be collected 120 days after the last transfusion.

Clinical Signs or Family History

A number of clinical situations will modify the traditional approach for obtaining a newborn screen. The following are suggested guidelines for situations which may arise.

Infants who exhibit clinical signs and symptoms:
The newborn screen, as with any laboratory screening test, may have false positives and false negatives. If signs and symptoms of a newborn screening condition are clinically evident, the provider should immediately consult Referral Center specialists for confirmatory testing and evaluation. Due to the rapid onset of some conditions, it may be necessary to treat as if the infant has the condition before the newborn screen or confirmatory results are available.

Infants with affected siblings or close relatives:
Many of the conditions screened by the NBS Program are genetic, so it is possible that multiple members of a family may be affected and known to the family. Providers are encouraged to contact the NBS Follow-up Program if it is known when infants are born to families with affected siblings or close relatives with a condition on the panel.

Prenatal diagnosis:
Prenatal diagnosis is available for some conditions if the mother chooses testing prior to birth. Prenatal testing is usually done if a previous child was born with a condition or there is a family history of disease.

For any infant with a positive family history or prenatal diagnosis, providers should contact appropriate specialists, ideally before birth, or immediately after birth. These steps will determine proper confirmatory tests, timing, and the best strategy for patient care.

Specimen Collection Timing Summary

Infant Status	Time of Collection
Term, Healthy	No earlier than 24 hours of age, but as soon as possible afterward.
Transfused	Prior to first transfusion. If a satisfactory specimen was not collected prior to transfusion, a specimen should be collected no sooner than 120 days after the last transfusion.
Premature, Low Birth Weight, or Sick	• Collect the first specimen upon admission to the NICU/ICU, regardless of age and prior to initiation of any treatments. • Collect the second specimen at 48–72 hours of life, for infants initially tested prior to 24 hours of age or any infant with an abnormal first screen. • Collect the third specimen at 28 days of life or prior to discharge, whichever comes first for infants < 34 weeks gestational age or < 2,000 grams at birth. For infants weighing > 2,000 grams at birth, a third specimen is at the discretion of the provider. Review all screening results prior to discharge.
NOTE: Additional specimens may be requested by the NBS Follow-up Program based on screening results.	

Unsatisfactory Specimens

Chapter 64C-7.002, F.A.C. requires the submitting entity ensure a satisfactory newborn screen has been collected. A reasonable attempt must be made to locate infants.

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 of the family by certified mail with return receipt.
(3) Notification of the provider on record. Please note: Notifying the provider does not absolve the birthing facility of responsibility. Original collector must ensure a repeat collected elsewhere is satisfactory.

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

Common Causes of Unsatisfactory Specimens

Unsatisfactory Types	Possible Causes
Quantity not sufficient (QNS)	Filter paper circles incompletely filled; not all circles filled.
Incomplete saturation	Blood did not soak through the filter paper.
Blood spots appear scratched or abraded	Blood applied with needle, capillary tube, or other means (filter paper has been damaged or torn by device).
Blood spots wet	Specimen not properly dried for three–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 came 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	Multiple blood drops applied to same filter paper circle. Circle filled from both sides of the filter paper.
No blood	Failure to obtain blood specimen.

Unsatisfactory Specimen Codes

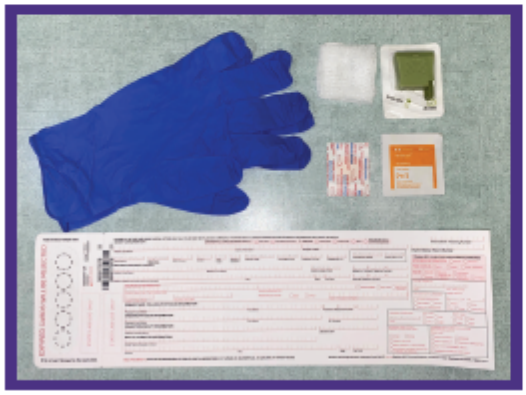
A	Specimen is contaminated
B	Specimen is quantity not sufficient (QNS) and damaged
C	Specimen is QNS and separated
D	Specimen is damaged
E	Incomplete saturation
F	Specimen is damaged and separated
G	Specimen is QNS and has repetitive spots
H	Supersaturated / layered
I	Specimen has insufficient data
J	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 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)

NBS Posters

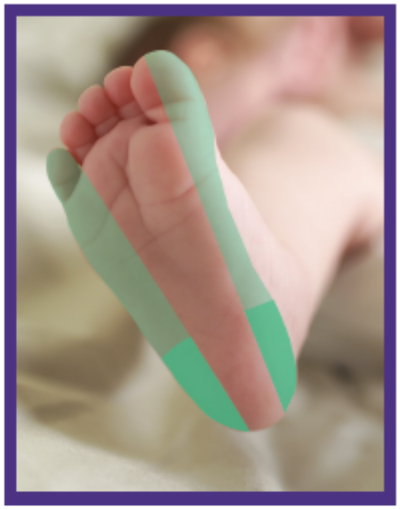
Please review the visual aids on the following pages for proper specimen collection and handling procedures.

FLORIDA NEWBORN SCREENING

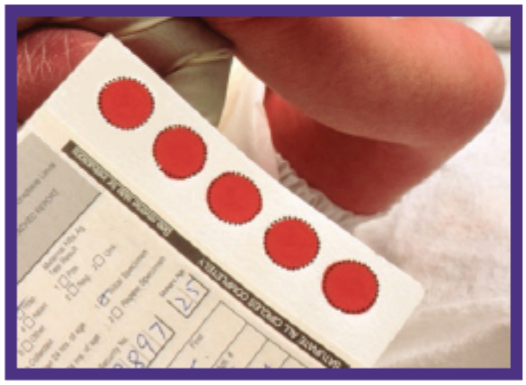
BLOOD SPECIMEN COLLECTION & HANDLING PROCEDURES



- Equipment needed for blood collection procedure: sterile lancet, alcohol prep, gauze pads, specimen card, bandage, and gloves.
- Complete ALL information on the specimen card. Take care not to handle the filter paper collection area.
- Tips to increase blood flow include:
  - Hold foot in a dependent position.
  - Use the warming method approved by the collection facility.
- The opaque green area along the bottom sides of the heel illustrated below indicates suitable areas for puncture site.



- Clean site with alcohol prep and wipe dry with gauze.
- Puncture the heel with lancet and wipe away the first blood drop.
- Allow another LARGE blood drop to form and lightly touch to the filter paper.
- The blood drop should soak through the filter paper and fill the collection circle. Only apply blood to one side of the filter paper.
- Fill all collection circles in the same manner as above.
- Gentle pressure may be applied to the foot to encourage continued blood flow.



- If blood flow is diminished, clean a new site and repeat steps to obtain more blood.
- Specimens must dry horizontally for 3-4 hours on a drying rack before packaging.
- Mail completed card to the Bureau of Public Health Laboratories (BPHL) - Jacksonville within 24 hours of collection, via overnight shipping method.
- Shipping address: 1215 N Pearl St., Jacksonville, FL 32202

Florida HEALTH

Simple spot check

Valid specimen:

Allow a sufficient quantity of blood to soak through to completely fill the pre-printed 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.

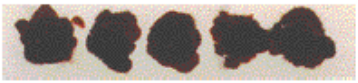
EBF

Invalid specimen and possible causes:

1. Specimen quality insufficient for testing.

Possible causes:

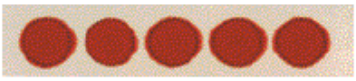
- Removing filter paper before blood has completely filled circle or before blood has soaked through to second side.
- Applying blood to filter paper with a capillary tube.
- Allowing filter paper to come into contact with gloved or ungloved hands or substances such as hand lotion or powder, either before or after blood specimen collection.



2. Specimen appears scratched or abraded.

Possible causes:

- Applying blood with a capillary tube or other device.



3. Specimen not dry before mailing.

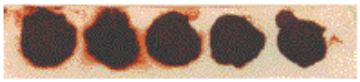
Possible causes:

- Mailing specimen before drying for a minimun of four hours.

4. Specimen appears supersaturated.

Possible causes:

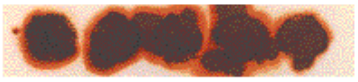
- Applying excess blood to filter paper, usually with a device.
- Applying blood to both sides of filter paper.



5. Specimen appears diluted, discolored or contaminated.

Possible causes:

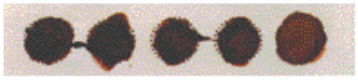
- Squeezing or "milking" of area surrounding the puncture site.
- Allowing filter paper to come into contact with gloved or ungloved hands or substances such as alcohol, formula, antiseptic solutions, water, hand lotion or powder, etc., either before or after blood specimen collection.
- Exposing blood spots to direct heat.



6. Specimen exhibits serum rings.

Possible causes:

- Not wiping alcohol from puncture site before making skin puncture.
- Allowing filter paper to come into contact with alcohol, hand lotion, etc.
- Squeezing area surrounding puncture site excessively.
- Drying specimen improperly.
- Applying blood to filter paper with a capillary tube.



7. Specimen appears clotted or layered.

Possible causes:

- Touching the same circle on filter paper to blood drop several times.
- Filling circle on both sides of filter paper.

8. No blood.

Possible causes:

- Failure to obtain blood specimen.

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

XXXXXXXX 10/2015

Proper Specimen Handling

DO

- Do dry the specimen card in a horizontal position.
- Do alternate the cards so the blood spots do not come into contact with each other.
- Do dry the blood spots at room temperature for 3–4 hours.
- Do keep specimen cards away from direct heat or sunlight.
- Do verify that all demographic sections have been accurately completed before mailing.
- Do fold the biohazard flap to keep blood spots from touching each other in the mail.
- Do check that a return address is present on the mailing envelope.
- Do mail specimen cards by overnight delivery, to be received at the BPHL-Jacksonville no later than three days after collection. Mark for early/first delivery.
- Do mark specimens mailed on Friday for “Saturday delivery” on the shipping label.
- Do keep a list of specimens that were sent and the tracking information so documentation is available should a delivery issue arise.

DON’T

- Do not allow blood spots to come into contact with any surface while drying.
- Do not refrigerate specimens.
- Do not place specimens in the envelope until completely dry.
- Do not place specimens in outdoor mailboxes located on the street.
- Do not transport specimens in plastic bags.
- Do not batch specimen cards beyond the same day.
- Do not dry specimen cards vertically.
- Do not submit copies of the card with the original specimen.
- Do not tape or staple the biohazard flap.

Specimen Transit Protocol

It is critical for newborn screening specimens to be received for testing in a timely manner. Some conditions included on Florida's newborn screening panel can cause death or permanent, irreversible damage in the first few days of life. Early identification is key in preventing or reducing negative outcomes.

Specimens should be mailed or transported as soon as they are properly dried (3–4 hours), and no later than 24 hours after collection. In all cases, specimens should be received at the BPHL-Jacksonville within three days of collection.

The recommended shipping method for specimens is overnight courier. Mark packages for early delivery. Specimens received before 10:00 a.m. will be processed the same day.

Specimens mailed on Friday should be marked for Saturday early delivery as the BPHL-Jacksonville is open to receive deliveries and process specimens on Saturdays.

A specimen will not be tested if it is received more than 14 days after the collection date.

Specimen cards should never be left in the heat. Please see storage and shipping recommendations on the back of the specimen card.

In case of emergencies (i.e. hurricane, etc.), DOH will notify collection facilities of alternative mailing addresses or procedures.

Specimen Retention and Storage Policy

After testing, the specimen card is stored for a period of six months. This allows 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.

At the end of the six-month period, the specimen card will be destroyed according to appropriate biomedical waste procedures.

Condition Definitions and Screening Results

Florida Newborn Screening Condition Panel

Amino Acidemias

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

Secondary Conditions
7. Citrullinemia Type II (CIT II)
8. Hypermethioninemia (MET)
9. Benign Hyperphenylalaninemia (H-PHE)
10. Biopterin Defect in Cofactor Biosynthesis (BIOPT BS)
11. Biopterin Defect in Cofactor Regeneration (BIOPT REG)
12. Ornitine Transcarbamylase Deficiency (OTC)
13. Carbamoyl Phosphate Synthase Deficiency (CPS)
14. Tyrosinemia Type II (TYR II)
15. Tyrosinemia Type III (TYR III)

Fatty Oxidation

Core Conditions
16. Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
17. Very Long-chain acyl-CoA Dehydrogenase Deficiency (VLCAD)
18. Long-chain L-3- Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)
19. Trifunctional Protein Deficiency (TFP)
20. Carnitine Uptake Deficiency (CUD)

Secondary Conditions
21. Carnitine Palmitoyltransferase Type I Deficiency (CPT-I)
22. Carnitine Palmitoyltransferase Type II Deficiency (CPT-II)
23. Short Chain Acyl-CoA Dehydrogenase Deficiency (SCAD)
24. Glutaric Acidemia Type II (GA II)

Organic Acidemias

Core Conditions
25. Isovaleric Acidemia (IVA) Glutaric Acidemia Type I (GA I)
26. 3-Hydroxy-3-Methylglutaric Aciduria (HMG)
27. Holocarboxylase Synthase Deficiency (MCD)
28. Methylmalonic Acidemia (MUT)
29. Methylmalonic Acidemia -Cobalamine Conditions (Cbl A, B)
30. 3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC)
31. Propionic Acidemia (PA) (PROP)
32. β-ketothiolase Deficiency (βKT)

(Organic Acidemias cont'd)

Secondary Conditions
33. Methylmalonic Aciduria with Homocystinuria (Cbl C,D)
34. Isobutyrylglycinuria (IBG)
35. 2-Methylbutyrylglycinuria (2MBG)
36. 3-Methylglutaconic Aciduria (3MGA)
37. 2-Methyl-3-Hydroxybutyric Acidemia (2M3HBA)
38. Ethylmalonic Encephalopathy (EE)

Endocrine Conditions

39. Congenital Adrenal Hyperplasia (CAH)
40. Primary Congenital Hypothyroidism (CH)

Enzyme Conditions

41. Galactosemia (GALT)
42. Biotinidase Deficiency (BIOT)

Hemoglobinopathies

Core Conditions
43. Hemoglobin SC Disease (FSC)
44. Sickle-β Thalassemia (FSA)
45. Sickle Cell Anemia (FS)

Secondary Conditions
46. Various Other Hemoglobinopathies

Lysosomal Storage Conditions

47. Mucopolysaccharidosis Type I (MPS I)
50. Pompe Disease

51. Severe Combined Immunodeficiency (SCID)

Secondary Condition
52. T-cell Related Lymphocyte Deficiencies

53. Cystic Fibrosis (CF)

54. X-linked Adrenoleukodystrophy (X-ALD)

55. Spinal Muscular Atrophy (SMA)

56. Critical Congenital Heart Disease (CCHD)

57. Hearing Loss

Metabolic Conditions

These conditions, which are rare, fall into three different biochemical categories: amino acidemias, organic acidemias, and fatty acid oxidation conditions. If left untreated, these conditions may cause developmental delays or even death. Not all these conditions are difficult to treat. Some require family education and oral medication. Some are treated with frequent feedings of normal food, while others require supplementation with medical foods or avoidance of certain foods.

Once a diagnosis is made, most conditions require lifelong management and follow-up.

Amino Acidemias

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

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

Amino Acidemia Conditions on Florida's Newborn Screening Panel:

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

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 remove toxic metabolites.

Fatty Acid Oxidation

Fatty acid oxidation conditions are inherited conditions 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. A fasting state can occur in infants in as little as four hours. Failure to diagnose fatty acid conditions 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 conditions have been misdiagnosed as Sudden Infant Death Syndrome (SIDS).

Fatty Acid Oxidation Conditions on Florida's Newborn Screening Panel:

- Medium chain Acyl-CoA Dehydrogenase deficiency (MCAD)
- Very long-chain acryl- 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

Infants may need to be fed often, even throughout the night, to avoid fasting. Depending on the condition, supplemental carnitine, a low-fat diet, and home glucose monitoring may be prescribed.

Organic Acidemias

Organic Acidemia conditions are inherited conditions 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 conditions have severe forms that present in the first week of life and constitute an emergency. Infants generally appear healthy at birth, but can develop poor feeding, liver and kidney problems, irritability, lethargy, vomiting, severe metabolic ketoacidosis, developmental delay, and possibly death if left untreated.

Organic Acidemias on Florida's Newborn Screening Panel:

- 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)
- Proprionic Acidemia (PROP)
- Mitochondrial acetoacetyl-CoA thiolase deficiency (BKT)

Treatment

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

Testing Methodology

Tandem mass spectrometry (MSMS)

Special Considerations

Low birth weight, Total Parenteral Nutrition (TPN), and carnitine supplementation may cause false positive results for certain metabolic conditions.

Repeat specimens for term infants (more than 37 weeks gestational age) with abnormal borderline levels for metabolic conditions or infants on TPN will not be referred to a contracted Genetics Referral Center or need a repeat specimen if the initial specimen is valid and results are normal. This policy does not apply to presumptive positive results. All infants with presumptive positive results will be referred to a contracted Genetics Referral Center for confirmatory testing.

Follow-up Actions for Abnormal Screening Results - Metabolic Conditions

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	Refer baby to a contracted Genetics Referral Center for evaluation and confirmatory testing.
Presumptive Positive	

X-linked Adrenoleukodystrophy (X-ALD)

X-ALD is a genetic condition that occurs primarily in males and mainly affects the nervous system and adrenal glands. There are three distinct types of X-ALD: childhood cerebral form, adrenomyeloneuropathy type, and Addison's disease only.

Children with the cerebral form of X-ALD experience learning and behavioral problems that usually begin between the ages of 4 and 10. Additional signs and symptoms of the cerebral form include aggressive behavior, vision problems, difficulty swallowing, poor coordination, and impaired adrenal gland function. The rate at which this condition progresses is variable but can be extremely rapid, often leading to total disability within a few years. The life expectancy of individuals with this type depends on the severity of the signs and symptoms and how quickly the condition progresses.

Testing Methodology

Newborn screening for X-ALD involves **three steps**:

1) All dried blood specimens obtained from routine NBS are tested for very long chain fatty acids (VLCFA) using MSMS.

2) If step one is abnormal, specimens will be tested for VLCFA using liquid chromatography MSMS.

3) If step two is abnormal, DNA sequencing is performed.

Treatment

Children who have adrenal insufficiency will be treated with corticosteroids and will need close monitoring by an endocrinologist. Those with the childhood cerebral form will need close monitoring by a neurologist and may receive allogenic hematopoietic stem cell transplantation (HCST) to stop the progression of X-ALD.

Special Considerations

Symptoms can vary depending on the type and age of onset. Although X-ALD primarily affects males, affected females can become symptomatic in adulthood.

Approximately one in every 17,000 newborns is born with X-ALD.

Follow-up Actions for Abnormal Screening Results- X-ALD

Determination	Action
Within Normal Limits	No further action needed.
Presumptive Positive	Refer baby to a contracted Genetics Referral Center for evaluation and confirmatory testing once DNA sequencing is complete.

Lysosomal Storage Disorders

Lysosomal storage disorders (LSD) are a group of rare conditions which occur when lysosomes (the recycling center of the cell) are unable to breakdown substances, such as protein or carbohydrates, due to missing enzymes. These substances can build up in cells and become toxic, damaging the body's cells and organs.

Mucopolysaccharidosis Type I (MPS I)

An inherited condition that affects many different parts of the body. MPS I has a wide spectrum of severity and affected newborns are categorized as having either the severe form or attenuated (less severe) form.

Signs and symptoms of severe MPS I generally appear within the first year of life and progress rapidly. Signs and symptoms of attenuated MPS I are usually milder and appear later in childhood. Early signs of MPS I include umbilical or inguinal hernias, macrocephaly, coarse facial features, swollen abdomen caused by hepatomegaly and splenomegaly, corneal clouding, hearing loss, frequent nasal drainage, and varying degrees of developmental delay and learning disabilities.

Testing Methodology

Newborn screening for MPS I involves **three steps**:

1) All dried blood specimens obtained from routine NBS are subjected to analysis of the alpha-L-iduronidase (IDUA) enzyme. If analysis is normal, the sample is deemed within normal limits.

2) If the IDUA analysis is <8% of the daily median, samples will be sent for biochemical analysis. If the analysis is normal, the sample is deemed within normal limits.

3) If step two is abnormal, sequencing of the IDUA gene is performed.

Treatment

Treatment for MPS I requires a multi-disciplinary approach from a team who specializes in neuromuscular conditions. This team may include primary care providers, cardiologists, pulmonologists, gastroenterologists, neurologists, audiologists, ophthalmologists, orthopedists, physical therapists, dentists; ear, nose, and throat specialists; and developmental specialists. Genetic counseling is imperative.

The two main treatment options for MPS I are HSCT and enzyme replacement therapy (ERT). Surgeries, physical therapy, medications, and dietary treatments may be beneficial depending on disease progression and/or symptoms.

Special Considerations

Approximately one in every 100,000 newborns is born with the severe form of MPS I. Approximately one in every 500,000 newborns is born with the attenuated form of MPS I.

Follow-up Actions for Abnormal Screening Results- MPS-1

Determination	Action
Within Normal Limits	No further action needed.
Presumptive Positive	Refer baby to a contracted Genetics Referral Center for evaluation and confirmatory testing after DNA sequencing is complete.

Pompe Disease

An inherited condition that affects many different parts of the body. There are two forms of Pompe Disease: Infantile Onset Pompe Disease (IOPD) and Late Onset Pompe Disease (LOPD).

Signs and symptoms of IOPD begin before birth or shortly after birth and include myopathy, hypotonia, failure to thrive, difficulty breathing, trouble feeding, respiratory infections, and hearing problems.

Signs and symptoms of LOPD are milder and slower to progress than the IOPD type and may include myopathy and difficulty breathing. Signs and symptoms may develop anytime from childhood to adulthood.

Testing Methodology

Newborn screening for Pompe Disease involves **two steps**:

1) All dried blood specimens obtained from routine NBS are subjected to analysis of the acid alpha-L-glucosidase (GAA) enzyme. If analysis is normal, the sample is deemed within normal limits.

2) If the GAA analysis is <15% of the daily median, sequencing of the GAA gene is performed.

Treatment

The treatment of Pompe Disease requires a multi-disciplinary approach from a team who specializes in neuromuscular disorders. This team may include primary care providers, neurologists, orthopedists, cardiologists, dieticians, and other healthcare professionals may need to be involved in the treatment plan. Genetic counseling is imperative.

Treatments may include ERT, physical therapy, respiratory therapy, and dietary treatments.

Special Considerations

Approximately one in every 40,000 newborns is born with Pompe disease.

Follow-up Actions for Abnormal Screening Results- Pompe Disease

Determination	Action
Within Normal Limits	No further action needed.
Presumptive Positive	Refer baby to a Genetics Referral Center for evaluation and confirmatory testing after DNA sequencing is complete.

Enzyme Conditions

Galactosemia

Galactosemia is an inherited condition which occurs when there is a deficiency of any of the three enzymes required to break down galactose (milk sugar). The condition results in elevated levels of galactose in the blood. Many infants with Galactosemia appear fine at birth but symptoms can become severe within a few days of ingesting breast milk or lactose-based formula.

Some of the early symptoms of Galactosemia include poor feeding, poor sucking reflex, vomiting, jaundice, irritability, and seizures. If left untreated, Galactosemia can cause developmental delays, cataracts, failure to thrive, or death.

Testing Methodology

Newborn screening for Galactosemia involves **two steps**:

1) All dried blood specimens obtained from routine NBS are tested for galactose 1-phosphate uridyl transferase (GALT) enzyme levels.

2) If the GALT level is low, total galactose (TGAL) is performed.

Treatment

Treatment for Galactosemia includes eliminating dietary galactose.

Special Considerations

GALT enzyme is sensitive to heat and can degrade over time. Specimens exposed to heat or delayed in processing may have false positive results.

Infants receiving lactose-free formula may have false negative results.

If the initial NBS specimen's results are normal, repeat specimens with abnormal results for Galactosemia will not be referred to a contracted Genetics Referral Center or need an additional repeat specimen.

A dried blood specimen must be collected before an infant receives a blood transfusion. Galactosemia results will not be reported within 120 days of transfusion. If transfusion occurs prior to screening, a repeat must be obtained 120 days after the last transfusion.

Approximately one in every 30,000–60,000 newborns is born with classic galactosemia.

Follow-up Actions for Abnormal Screening Results - Galactosemia

Determination	Action
Within Normal Limits	No further action required
Borderline	An immediate repeat newborn screening specimen must be collected.
Presumptive Positive	The baby is referred to a contracted Genetics Referral Center for

	evaluation and confirmatory testing.

Biotinidase Deficiency

Biotinidase deficiency is an autosomal recessive condition of biotin recycling. Biotin is an essential vitamin widely present in foods such as egg yolks, soybeans, and cereals. Biotin-dependent 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 delays, and metabolic acidosis.

Testing Methodology

All dried blood specimens obtained from routine NBS are tested for Biotinidase enzyme activity.

Treatment

Treatment for Biotinidase deficiency involves daily oral biotin.

Special Considerations

If the initial NBS specimen's results are normal, repeat specimens with abnormal results for Biotinidase will not be referred to a contracted Genetics Referral Center or need an additional repeat specimen.

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.

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 decreased Biotinidase activity (false positive).

A dried blood specimen must be collected before an infant receives a blood transfusion. Biotinidase results will not be reported within 120 days of transfusion. If a transfusion occurs prior to screening, a repeat specimen should be obtained no sooner than 120 days after the last transfusion.

Approximately one in every 60,000 newborns is born with Biotinidase deficiency.

Follow-up Actions for Abnormal Screening Results - Biotinidase Deficiency

Determination	Action
Within Normal Limits	No further action needed.
Borderline	An immediate repeat newborn screening specimen must be collected.
Second Borderline Result	Refer baby to a contracted Genetics Referral Center for

Presumptive Positive	evaluation and confirmatory testing.

Congenital Adrenal Hyperplasia (CAH)

Congenital Adrenal Hyperplasia (CAH) is a group of autosomal recessive conditions 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 other conditions on the newborn screening panel, 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.

Testing Methodology:

All dried blood specimens obtained from routine NBS are tested for 17 Hydroxyprogesterone (17-OHP) levels.

Treatment

Treatment consists of hormone replacement therapy.

Special Considerations

Results are dependent upon the baby’s age and weight at time of collection.

Approximately one in every 10,000 newborns is born with CAH.

Follow-up Actions for Abnormal Screening Results for CAH

Determination	Recommended Action
Within Normal Limits	No further action needed.
Borderline Results <24 hrs. of age or <2,500 grams	A repeat newborn screening specimen must be collected. Infants in the NICU should follow the NICU guidelines for specimen collection.
Borderline Results > 24 hrs. of age & > 2,500 grams	A repeat newborn screening specimen should be collected when the infant is two weeks of age.
Repeat Borderline Results CAH > 24 hrs. & > 2,500 grams	If repeat results are **increasing** in value, refer baby to an Endocrine Referral Center for evaluation and confirmatory testing. If repeat results are **decreasing** in value, obtain a repeat NBS specimen. Baby should be examined for ambiguous genitalia, hyponatremia, and hyperkalemia. **If symptoms are present or CAH is suspected, contact NBS Follow-up Program for referral to an Endocrine Referral Center for evaluation and confirmatory testing.**
Presumptive Positive	Refer baby to a contracted Endocrine Referral 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 as the newborn's condition dictates whether serum thyroid studies are warranted.

Newborn screening is not designed to detect late onset 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 a hoarse cry.

Without treatment, CH may result in mental and/or growth delays, and developmental delay. CH occurs in one out of every 3,500 births and is twice as common in females as it is in males.

Testing Methodology

All dried blood specimens obtained from routine NBS are tested for T4 (thyroxine) and TSH (thyroid stimulating hormone) levels.

Treatment

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

Special Considerations

Results are dependent upon the infant's age at time of collection. Date and time of collection are important to note, results are interpreted differently once the infant is greater than 24 hours of age. TSH levels are normally elevated in the first 24 hours of life.

Approximately one in every 3,000–4,000 newborns is born with CH.

Follow-up Actions for Abnormal Screening Results - CH

Determination	Follow-Up Action
Within Normal Limits	No further action needed.
Date of Birth Hypothyroidism or Neonatal Hyperthyroxinemia	An immediate repeat newborn screening specimen must be collected.
Low T4	The baby should have a repeat NBS specimen collected at 2–3 weeks of age or have a free T4 and TSH collected by a local clinical laboratory to confirm results.
Borderline	An immediate repeat newborn screening specimen must be collected or have a free T4 and TSH collected by a local clinical laboratory. **If free T4 and TSH is completed by a local clinical laboratory, fax results to 850-922-5385.**
Second Borderline	Refer baby to a contracted Endocrine Referral Center for evaluation and confirmatory testing.
Presumptive Positive	

Hemoglobinopathies

Hemoglobinopathies, including Sickle Cell Disease, are a group of inherited conditions caused by abnormal hemoglobin (Hgb) molecules in the red blood cells. These abnormal hemoglobin molecules are detectable 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 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 having "trait." Most hemoglobinopathy 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. Special efforts to ward off dehydration are important for trait patients.

Testing Methodology:

All dried blood specimens obtained from routine NBS are tested for the presence of F, S, A, C, E, D, and BARTS hemoglobins.

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.

Special Considerations

Regardless of age, a NBS specimen should be collected prior to transfusion. Samples collected within 120 days of a transfusion are invalid for hemoglobinopathy testing. If a valid specimen is not collected prior to transfusion, a repeat specimen should be obtained at least 120 days after the last transfusion.

Approximately one in every:

- 375 African American** newborns is born with sickle cell disease.
- 835 African American** newborns is born with hemoglobin S/C disease.
- 20,000 newborns is born with another hemoglobinopathy.

** 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.

Follow-up Actions for Abnormal Screening Results – Hemoglobinopathies

Determination	Action
Normal	No further action needed.
Trait	A letter is mailed to the parent/guardian and primary care provider on record advising of detected trait.
Presumptive Positive	Refer infant to a contracted Hemoglobinopathy Referral Center for evaluation and confirmatory testing.

Cystic Fibrosis (CF)

Cystic Fibrosis is one of the most common inherited diseases in the United States and primarily affects the lungs and digestive system. The CF gene 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. Excess secretions lead 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 cause high salt content in the sweat of individuals with CF. A quantitative sweat test is the gold standard diagnostic test and should be performed at a contracted CF Referral Center.

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), and frequent greasy, bulky stools, or difficulty in bowel movements.

If left untreated, CF will lead to impaired growth, usually below the 10th percentile, severe chronic lung infections, or early death.

Testing Methodology

Newborn screening for CF involves **two steps**:

1) All dried blood specimens obtained from routine NBS are tested for levels of trypsinogen using the Immunoreactive Trypsinogen Test (IRT).

2) Each testing day, the top 4% of specimens with the highest IRT results are analyzed for DNA variants.

Treatment

There is no cure for CF, but symptoms may be managed and reduced by a care team with experience treating CF. Treatments may include physical therapy, respiratory therapy, nutritional counseling, and medications. Surgeries, including lung or liver transplant, may be necessary in severe cases. Genetic counseling is recommended.

Special Considerations

It is possible for an infant to have a variant(s) not detected through NBS as not every specimen is tested for CF variants nor does screening include all known CF variants. Providers should not use NBS results to rule out the presence of disease, particularly with symptomatic presentation or family history. Providers may refer to a local CF Referral Center for evaluation.

IRT levels may decline into the normal range by 10 days of age and may not be picked up through repeat screenings.

IRT levels may be falsely elevated in premature or sick infants.

Approximately one in every 3,500 Caucasian newborns; 7,000 Hispanic newborns; and 17,000 African American newborns is born with CF.

Follow-up Actions for Abnormal Results – Cystic Fibrosis

Determination	Follow-Up Action
Within Normal Limits	No further action needed.
CF Borderline* (IRT >96th percentile, no variants detected)	NICU: Reliability of IRT levels cannot be determined. Collect a repeat newborn screening specimen before discharge. Home: Physician discretion strongly advised. If signs and symptoms or a family history of CF is suspected, contact the NBS Follow-up Program.
Presumptive Positive (IRT >96th percentile, one or two CF variants detected)	Refer infant to a contracted CF Referral Center for evaluation and confirmatory testing.

*Referral to a CF Center for infants with multiple specimens with an elevated IRT without CF variants is at the discretion of the NBS Follow-up staff or primary care provider.

Florida's NBS panel includes analysis for 74 variants utilizing the Agena CFTR Panel:

American College of Medical Genetics (ACMG) Recommended Variants	621+1G>T 711+1G>T 1507 del 1717-1G>A 1898+1G>A 2184delA	2789+5G>A 3120+1G>A 3659delC 3849+10kbC>T A455E F508del	G85E G542X G551D N1303K R117H R334W	R347P R553X R560T R1162X W1282X
Additional Variants	394delTT 406-1G>A 935delA 1078delT 1506V 1507V 1677delTA 1898+5G->T 2055del9->A 2143delT 2183AA->G 2307insA 3199del6	3791delC 3876delA 3905insT A559T CFTR del e2, 3 D1152H E60X F508C G178R G330X G551S G1244E G1349D	K710X L206W M1101K Q493X Q890X R75X R117C R347H R560K R1066C R1158X R1162L R1162Q	S549N S549R_1645A>C S549R_1647T->G S1196X S1251N S1255P S1255X T5/T7/T9 V520F W1089X Y122X Y1092X

Severe Combined Immune Deficiency (SCID)

Severe Combined Immune Deficiency (SCID) is a group of inherited conditions which affect the immune system. At birth, these infants appear healthy while the mother's immune system continues to protect them from infection for the first few weeks of life. If left untreated, these infants become extremely vulnerable to infection that can quickly become a life-threatening illness.

Testing Methodology

All dried blood specimens obtained from routine NBS are tested for T-cell receptor excision circles (TREC).

Treatment

Affected infants should be isolated from large crowds and anyone with a contagious illness to prevent infection. It may be necessary to keep the newborn on preventative antibiotics.

SCID may be treated by bone marrow transplant.

Special Considerations

Surgical procedures, particularly cardiac, and extreme prematurity may result in false positive results.

If SCID or other T-cell deficiency is suspected, live vaccines, particularly Rotavirus, should be withheld until diagnosis is made.

Approximately one in every 40,000–75,000 newborns is born with SCID.

Follow-up Actions for Abnormal Screening Results – SCID

Determination	Follow-Up Action
Within Normal Limits	No further action needed.
Borderline	Immediate repeat NBS specimen is required.
Repeat Borderline	Refer infant to a contracted SCID Referral Center for evaluation and confirmatory testing.
Presumptive Positive	

Spinal Muscular Atrophy (SMA)

Spinal muscular atrophy (SMA) is a group of conditions which affect the motor neurons of the spinal cord. These neurons control muscles used for activities such as breathing, crawling, and walking. The loss of motor neurons causes progressive muscle weakness and atrophy. Symptoms typically begin to appear around one month of age.

Testing Methodology

All dried blood specimens obtained from routine NBS are tested for homozygous deletion of exon 7 in the SMN1gene.

Treatment

Management begins with diagnosis, with symptoms being addressed by different specialists on the care team.

SMA may be treated by SMA enhancing therapies approved by the Food and Drug Administration (FDA).

Special Considerations

Approximately one in every 10,000 newborns is born with SMA.

Follow-Up Actions for Abnormal Screening Results – SMA

Determination	Follow-Up Action
Within Normal Limits	No further action needed.
Presumptive Positive	Refer infant to a contracted Genetics Referral Center for evaluation and confirmatory testing.

Critical Congenital Heart Disease (CCHD)

Infants born with CCHD have an abnormality in the structure of the heart. Some infants may look and act healthy at first and could be sent home before the CCHD is detected. Please refer to the CCHD Screening Algorithm.

A passing CCHD screening result does not exclude the possibility of cardiac disease. A full cardiac evaluation should be completed if clinically indicated, regardless of a passing CCHD screening result.

CCHD screening is designed to identify seven primary conditions:

- Hypoplastic Left Heart Syndrome
- Pulmonary Atresia
- Tetralogy of Fallot
- Total Anomalous Pulmonary Venous Connection
- Transposition of the Great Arteries
- Tricuspid Atresia
- Truncus Arteriosus

CCHD screening can identify seven secondary conditions:

- Aortic Arch Atresia or Hypoplasia
- Atrioventricular Septal Defect (ASD)
- Coarctation of the Aorta
- Double-Outlet Right Ventricle
- Ebstein's Anomaly
- Interrupted Aortic Arch
- Ventricular Septal Defect (VSD)

Testing Methodology

Pulse oximetry screening is completed per recommended protocol at the bedside and reported on the specimen card.

Treatment

CCHDs typically require a procedure or surgical intervention.

Special Considerations

Respiratory distress/disease may cause a failed CCHD result.

Infants receiving oxygen supplementation are not candidates for CCHD screening. Complete the screening when the infant is no longer receiving oxygen supplementation.

CCHD screening is not required if an echocardiogram has been performed.

Approximately two in every 1,000 newborns are born with CCHD.

Follow-up Actions for Abnormal Screening Results – CCHD

CCHD screening is a point-of-care test completed by the hospital or birth provider. Evaluation and diagnosis are completed immediately after a failed screening. Completion of a diagnostic echocardiogram (echo) is the Gold Standard of care after a failed screening.

A pulse oximetry measurement on both the right hand and either foot should be completed after 24 hours of age. A difference of more than 3% between the two readings indicate the possible presence of a CCHD.

CCHD screening results should be reported on the newborn screening specimen card. **All applicable fields of the pulse oximetry section of the specimen card are required,** not just "Pass" or "Fail." Oxygen saturations, in addition to the echo outcome or reasons not screened, if applicable, must be reported.

The NBS Follow-up Program manages reported failed CCHD results. Facilities are contacted regarding the outcome of all failed screenings, including information about echocardiogram results, diagnosis, and surgeries/procedures.

CCHD SCREENING ALGORITHM

RIGHT HAND	EITHER FOOT											<90
100	100	99	98	97	96	95	94	93	92	91	90	*
99	100	99	98	97	96	95	94	93	92	91	90	*
98	100	99	98	97	96	95	94	93	92	91	90	*
97	100	99	98	97	96	95	94	93	92	91	90	*
96	100	99	98	97	96	95	94	93	92	91	90	*
95	100	99	98	97	96	95	94	93	92	91	90	*
94	100	99	98	97	96	95	94	93	92	91	90	*
93	100	99	98	97	96	95	94	93	92	91	90	*
92	100	99	98	97	96	95	94	93	92	91	90	*
91	100	99	98	97	96	95	94	93	92	91	90	*
90	100	99	98	97	96	95	94	93	92	91	90	*
<90	*	*	*	*	*	*	*	*	*	*	*	*

Green = PASS (Negative screen)

Red = Rescreen in 1 hour

Red for 3 consecutive screens = FAIL (Positive Screen)

***Red = Any result <90 is an automatic FAIL. (Positive screen)**

Immediately notify the provider of a failed screening.

Hearing Screening

Newborn hearing screening services are provided to identify newborns with hearing loss, ensure follow up testing and referral for intervention services, in accordance with Chapter 383, sections 383.145, 383.146, F.S. Additionally, Chapter 64C-7.006 F.A.C. relates to the reporting of hearing testing.

The information contained in this document is intended to be a general overview of newborn hearing screening only. The Florida Newborn Hearing Screening and Early Hearing Detection and Intervention (EHDI) Program has released *The Florida Guidelines for Newborn Hearing Screening* available online at www.floridanewbornscreening.com/hearing. Health care facilities providing newborn hearing screenings are encouraged to review these Guidelines and contact the Newborn Hearing Screening and EHDI Program at 866-289-2037 for more information.

Legislative Intent

The intent of this section is to provide a statewide comprehensive and coordinated interdisciplinary program of early hearing impairment screening, identification, and follow up care for newborns. The goal is to screen all newborns for hearing impairment to alleviate the adverse effects of hearing loss on speech and language development, academic performance, and cognitive development.

Requirements for screening of newborns; insurance coverage; referral for ongoing services:

(a) Each licensed hospital or other state-licensed birthing facility that provides maternity and newborn care services shall provide that all newborns are, prior to discharge, screened for the detection of hearing loss, to prevent the consequences of unidentified disorders.

(b) Each licensed birth center that provides maternity and newborn care services shall provide that all newborns are, prior to discharge, referred to a licensed audiologist, a physician licensed under Chapter 458 or Chapter 459 F.S., or a hospital or other newborn hearing screening provider, for screening for the detection of hearing loss, to prevent the consequences of unidentified disorders. The referral for appointment shall be made within 30 days after discharge. Written documentation of the referral must be placed in the newborn's medical chart.

EHDI and Hearing Health Care Partners

All hearing care providers involved in the care of newborns are essential partners with the Florida EHDI team. Together, the goal is to encourage early diagnosis of hearing loss and provide every child the opportunity for early intervention based on the 1-3-6 benchmarks. These benchmarks include:

- Hearing screening by 1 month of age
- Diagnosis by 3 months of age
- Early intervention enrollment by 6 months of age.

Program Oversight

One individual (preferably an audiologist) from each Newborn Hearing Screening Program is to serve as the contact person for the EHDI program. The designated contact person is responsible for:

- Ensuring that persons who perform hearing screening are sufficiently trained, coordinating follow-up services, and managing all correspondence.
- Providing oversight and participating in the writing of individual program screening policies and procedures.
- Providing re-education when needed and taking corrective action as necessary to improve and maintain program performance.
- Monitoring program statistics and quality assurance.
- Overseeing hearing screeners and monitoring schedules to ensure 365 days of coverage.
- Performing a case reconciliation every month to assure all results are reported to the EHDI program.

Performing a Newborn Hearing Screening

Best practices for performing a high-quality hearing screening are available in the Florida Guidelines for Newborn Hearing Screening.

Reporting Results to the Newborn Hearing Screening and EHDI Program

Florida Statutes mandate that all babies are screened for hearing loss at birth, unless a parent refuses the screen. All hearing screens and diagnostic test data are reported annually to the Centers for Disease Control and Prevention (CDC).

For information on mandatory reporting to the EHDI program see the Florida Guidelines for Newborn Hearing Screening.

Missed Hearing Screen

Every effort shall be made to complete the mandated hearing screening prior to hospital discharge. When a hearing screening is not completed and a newborn is discharged before an initial hearing screening, a process will be in place for the hospital or birthing facility to contact the family and arrange for an outpatient hearing screening within **30 days**.

Follow-up Hearing Testing Needed

At the time of discharge for newborns who did not pass (fail/refer) the final hearing screening in one or both ears, it is recommended the hospital ensure:

- A written, preferably signed, copy of the hearing screening results, and follow up appointment information is provided to the parent or legal guardian.
- The outpatient appointment is scheduled prior to discharge, including the location, date, and time for follow up rescreening or diagnostic hearing testing.

Contact Information

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 Newborn Screening Nurse Line: (866) 804-9166
Secure Newborn Screening Fax Line: (850) 922-5385

Toll-free Newborn Hearing Screening Line: (866) 289-2037
Secure Newborn Hearing Screening Fax Line: (850) 245-4049

Website:
www.floridanewbornscreening.com

Bureau of Public Health Laboratories-Jacksonville

Physical address (For mailing specimens)**:**
1217 N. Pearl Street
Jacksonville, FL 32202

Mailing address (For correspondence only, **do not use for mailing specimens.**)**:**
PO Box 210
Jacksonville, FL 32231

Phone numbers:
Main Phone Line: (904) 791-1500
Customer Service Lines: (904) 791-1644
(904) 791-1645
(904) 791-1646
(904) 791-1647
Secure Fax Line: (904) 791-1671

DOH Contracted Referral Centers

See the following pages for coverage area maps.

Condition	Referral Center	Phone
Endocrine (CAH, CH)	University of Florida **Gainesville**	(352) 265-7337
	University of Miami **Miami**	(305) 243-5707
	University of South Florida **Tampa**	(813) 396-2980
Cystic Fibrosis	Nemours Children's Specialty Care **Pensacola**	(850) 505-4700
	Nemours Children's Specialty Care **Jacksonville**	(904) 697-3600
	University of Florida **Gainesville**	(352) 273-8380
	Nemours Children's Specialty Care **Orlando**	(407) 650-7715
	Orlando Health - Arnold Palmer Hospital for Children (Only Winnie Palmer & South Lake Hospital births) **Orlando**	(321) 841-6350
	University of South Florida **Tampa**	(813) 259-8700
	Johns Hopkins All Children's Hospital **St. Petersburg**	(727) 767-3995
	Lee Memorial Health- Golisano Children's Hospital **Ft. Myers**	(239) 437-5500
	Memorial Healthcare System - Joe DiMaggio Children's Hospital **Hollywood**	(954) 265-6333
	Nicklaus Children's Hospital **Miami** (South Miami & Monroe)	(305) 662-8380
	University of Miami **Miami** (North Miami)	(305) 243-6642
Genetics (Biotinidase, Galactosemia, LSDs, MSMS, SMA, X-ALD)	University of Florida **Gainesville**	(352) 294-5050
	University of Miami **Miami**	(305) 243-9633
	University of South Florida **Tampa**	(813) 259-8772

Hemoglobinopathies	UF Health Pediatric Specialty Services - Studer Family Children's Hospital **Pensacola**	(850) 416-1890
	University of Florida **Gainesville**	(352) 273-9120
	Nemours Children's Specialty Care **Jacksonville**	(904) 697-3793
	Johns Hopkins All Children's Hospital **St. Petersburg**	(727) 767-4176
	University of South Florida **Tampa**	(813) 396-9725
	Orlando Health - Arnold Palmer Hospital for Children (Only Winnie Palmer & South Lake Hospital births) **Orlando**	(321) 843-8588
	AdventHealth **Orlando**	(407) 303-2080
	Lee Memorial Health System - Golisano Children's Hospital of SW Florida **Ft. Myers**	(239) 343-5333
	Memorial Healthcare System - Joe DiMaggio Children's Hospital **Hollywood**	(954) 265-2234
	Broward Health - Salah Foundation Children's Hospital **Ft. Lauderdale**	(954) 355-4527
	University of Miami **Miami**	(305) 243-0850
SCID	University of Florida **Gainesville**	(352) 294-5252
	University of Miami **Miami**	(305) 243-2222
	University of South Florida **St. Petersburg** (@ Johns Hopkins All Children's Hospital)	(727) 767-4150
SMA Treatment Centers	University of Florida **Gainesville**	(352) 294-5757
	University of Miami **Miami**	(305) 243-0171
	University of South Florida **Tampa**	(813) 844-8126
	Nemours Jacksonville **Jacksonville**	(904) 697-3151
	Nemours Orlando **Orlando**	(407) 221-8272

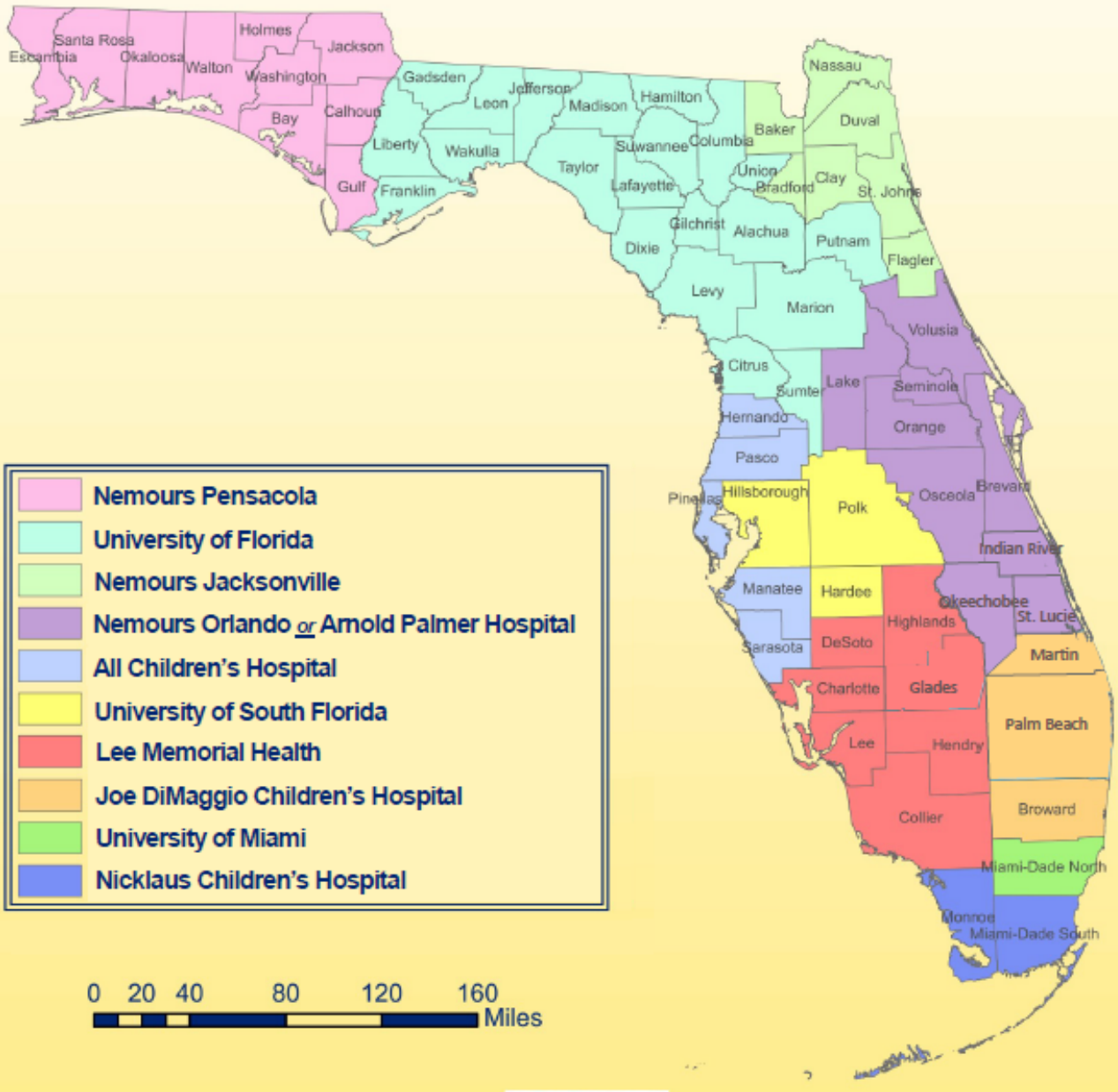
Newborn Screening Endocrine, Genetics, & SCID

Referral Centers

Legend

- University of Florida
- University of Miami
- University of South Florida

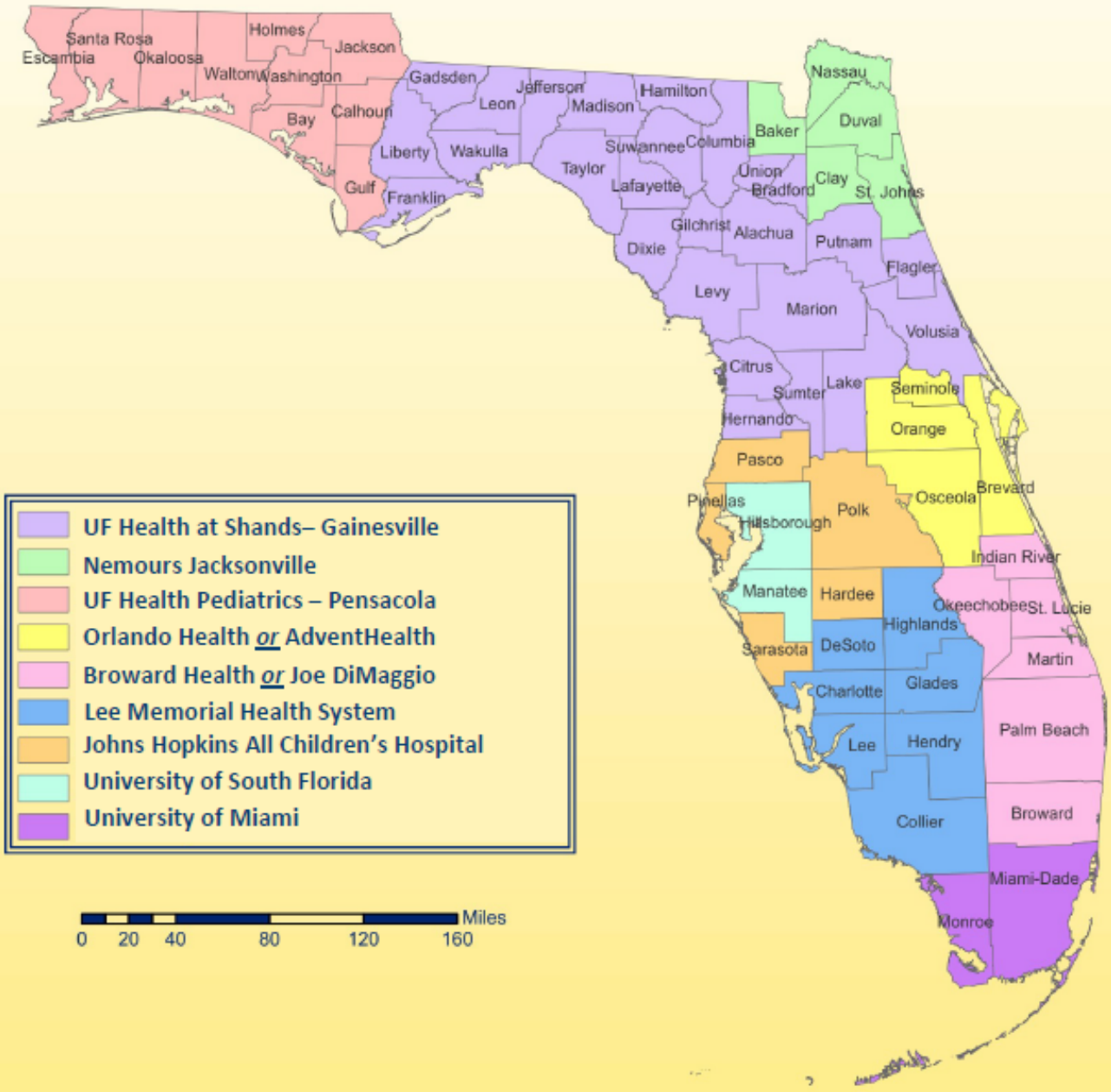
Newborn Screening Cystic Fibrosis Referral Centers



Florida HEALTH

5/24/2021

Newborn Screening Hemoglobinopathy Referral Centers



Rev. 2/11/2020

Newborn Screening SMA Treatment Centers

Resources

Florida Newborn Screening Program
Resourceful website for hospitals, physicians' offices, parents, and other members of the community for information regarding all areas of the Florida NBS Program.
www.floridanewbornscreening.com

Florida Newborn Screening Results
Online access to newborn screening results for registered users.
https://www.fnsr.net

CMS Kids and Early Steps
Website for families and providers to assist with accessing Children's Medical Services programs and Managed Care Plan.
http://www.cms-kids.com/

Florida Kidcare
Health insurance for Florida children from birth through age 18.
www.floridakidcare.org

Newborn Screening Resources

Baby's First Test
Parent-oriented website with generous amounts of information regarding newborn screening disorders and practices across the country.
www.babysfirsttest.org

Centers for Disease Control and Prevention (CDC)
Portal for information regarding the latest recommendations and information regarding newborn screening on a Federal level. Resources and multimedia tools available.
https://www.cdc.gov/newbornscreening/

March of Dimes
Website with information and resources for families and providers regarding birth defects, infant loss, prematurity, and infant mortality.
http://www.marchofdimes.com/home.asp

National Newborn Screening and Genetics Resource Center
An independent national newborn screening resource site with information for healthcare professionals and families.
http://genes-r-us.uthscsa.edu/

NewSTEPS
A national newborn screening resource site in partnership with the Association of Public Health Laboratories (APHL) featuring national data, links to articles and other resources.
www.newsteps.org

Southeastern Regional Genetics Group (SERGG)
Website to keep up with meetings for the Southeastern Regional Genetics Group. Links to presentations from previous conferences are posted for viewing.
http://www.sergginc.org/

Hearing Loss Resources

AG Bell Florida
Website for the Alexander Graham Bell Association's Florida Chapter. Resources for helping families and providers with support and education for children with hearing loss.
https://www.agbell.org/Connect/Local-Chapters/Florida

Florida Association of Speech-Language Pathologists and Audiologists
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.
https://www.flasha.org/

Florida Department of Education Bureau of Exceptional Education and Student Services (BEESS)
Resource page for the FL Department of Education's Bureau of Education and Exceptional Education Student Services.
http://www.fldoe.org/academics/exceptional-student-edu/beess-resources/

Florida Hands & Voices
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.
http://www.flhv.org/

Florida School for the Deaf and Blind
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. Includes resources for teachers of the deaf and hard of hearing.
www.fsdb.k12.fl.us

Florida's Alliance for Assistive Services and Technology
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.
www.faast.org

UF Health Florida and Virgin Islands (FAVI) Deaf-Blind Collaborative
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.
www.deafblind.ufl.edu