



Having a baby is exciting, and you may have questions. Don't be afraid to ask your doctor for more information about anything you don't fully understand.

Florida Newborn Screening does not store specimens beyond the time necessary to conduct quality assurance activities. Residual dried bloodspot specimens are destroyed six months after testing is complete. Some abnormal samples are kept unidentified for quality assurance and method validation purposes. Florida Newborn Screening does not use residual dried bloodspot specimens for biomedical research.

See *Florida Statutes* 383.14 and 383.145 at [www.leg.state.fl.us](http://www.leg.state.fl.us) for laws governing Florida Newborn Screening.

For more information about newborn screening contact:

Children's Medical Services  
Newborn Screening Program  
1-866-804-9166  
1-866-289-2037  
[www.floridanewbornscreening.com](http://www.floridanewbornscreening.com)



## Newborn Screening *an important beginning*



"This one small screening has changed the entire outcome of our daughter's life." —a new parent

Your baby is precious and, as a new parent, you want to do everything you can to keep your baby safe and healthy. An important step is make sure your baby gets newborn screening tests before you both leave the hospital.

- Your baby's blood spots are kept by the lab for only six months and then destroyed
- Newborn Blood Screening tests your baby's blood for different diseases and conditions. Newborn Hearing Screening checks to see if your baby has any hearing problems. Early detection and treatment of health issues can help your baby grow up healthier.
- These tests are an important beginning of a long, healthy life for your baby.

### Newborn Blood Screening

#### Why does my baby need Newborn Screening tests?

- All babies are tested because even babies who look healthy may have a rare health problem.
- If problems are found early, treatment can begin and may be able to prevent serious problems like mental retardation or death.
- It is important to remember that the newborn screening test is a screening test and not a diagnostic test. There will be false positive test results and false negative test results.
- Some disorders will detect an infants' carrier status in the process to detect a true positive.

#### How will my baby be tested for the Newborn Blood Screening?

- Before you leave the hospital, several drops of blood will be taken from your baby's heel.
- The hospital will send the blood sample to the Florida Newborn Screening Lab in Jacksonville.

#### How will I get the results of the test?

- Your baby's doctor will be given the results by the hospital. Be sure to tell the hospital the correct name and phone number of your baby's doctor so they can send the results.
- Parents are directly notified of test results if there is a problem.
- Ask about the results when you see your baby's doctor.

#### Why do some babies need to have the blood screening repeated?

- Some babies need to have the screening repeated because there is a problem with the blood sample.
- All babies who leave the hospital earlier than 24 hours must have the screening repeated within 5 days of age.
- A few babies need to have the screening repeated because the first test showed a possible health problem.
- If your baby needs to have the screening repeated, get it done right away.
- Our screening tests are very sensitive. Sometimes the results look positive even if there are no problems. Get your baby retested to be sure.

#### What babies need to have additional testing?

- All babies with abnormal screening results need more testing.
- You will be contacted if your baby needs to have additional testing.
- If your baby needs more testing, get it done right away.
- Make sure your hospital and doctor have your correct address and phone number.

#### What if my baby is uninsured?

Find out if Florida's KidCare health insurance is for you. Visit [www.floridakidcare.org](http://www.floridakidcare.org) or call 1-888-540-5437.

## Disorders found by Newborn Blood Screening

All disorders, except for hypothyroidism, are inherited from parents. Some children who are diagnosed with one of these disorders will need close supervision by a medical specialist and their team.

**Biotinidase Deficiency** is when the body is unable to use the vitamin biotin. It can result in seizures, hearing loss, and death in severe cases. Treatment is simple and involves daily doses of biotin.

**Endocrine Disorders** are caused by decrease or absence of certain hormones. Treatment involves lifelong hormone replacement therapy.

**CONGENITAL ADRENAL HYPERPLASIA** can result in lethargy, vomiting, poor feeding, death, and a female being identified as a male at birth.

**CONGENITAL HYPOTHYROIDISM** can result in mental and growth retardation.

**Cystic Fibrosis** is a progressive lung disease with other organ failures and lung and food digestion problems. Treatment leads to decreased hospitalizations and better growth, development and lung functioning.

**Galactosemia** is the failure to break down the milk sugar galactose. It can lead to cataracts, liver cirrhosis, mental retardation and/or death. Treatment is elimination of galactose from the diet usually by substituting soy.

**Amino Acid Disorders** are caused by the body's failure to breakdown certain proteins. If untreated the baby could have poor feeding, vomiting, neurological symptoms, mental retardation, coma, and death. Treatment may involve strict dietary management of the amino acid and medication. Florida currently screens for seven amino acid disorders.

**Fatty Acid Oxidation Disorders** are caused by the body's failure to break down fatty acids to convert to energy. If not diagnosed, fatty acid disorders may result in excessive fat buildup in the liver, heart and kidneys. If left untreated, low blood sugar, vomiting, seizures, lethargy, liver disease, developmental delay, coma,

and death could occur. Treatment may include regular food intake, a special low fat diet and medicine. The child will need close supervision by a metabolic specialist and dietician. Florida currently screens for ten fatty acid oxidation disorders.

**Organic Acidemias** are a group of metabolic disorders that lead to a build up of organic acids in blood and urine. Most of these disorders can be severe and can be present within the first week of life. The baby can develop poor feeding, liver and kidney problems, mental retardation, and possibly death. Treatment involves a strict diet that must be followed for life and medication. Florida currently screens for nine organic disorders.

**Hemoglobinopathies** are disorders of the red blood cells. The baby can develop life-threatening infections, pneumonia, pain episodes and stroke. Sickle cell anemia causes clogged blood vessels resulting in severe pain and other severe health problems. Family education, immunizations, penicillin and prompt treatment of acute illness improves the outcome for the child. Florida currently screens for three hemoglobinopathies.

**Severe Combined Immunodeficiency (SCID)** is a group of inherited disorders that affect the immune system. At birth, these infant's 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 infections that will develop into a life threatening illness. Early diagnosis and treatment will provide a better outcome.

**Critical Congenital Heart Disease (CCHD)** Babies born with congenital heart disease have an abnormality in the structure of the heart which is present at birth. Some babies can look and act healthy at first, and are sent home before the heart defect is detected. These babies are at risk for developing serious complications. Pulse oximetry is a non-invasive, painless test used to detect oxygen levels in the baby's blood. Pulse oximetry screening can identify some, but not all babies with CCHD before they show signs or symptoms. Once early diagnosis is made, babies receive specialized care and treatment, leading to longer, healthier lives.

For a complete listing of disorders screened visit: <http://www.doh.state.fl.us/cms/nbscreen-disorder.html>.



### What if I have questions?

Ask your baby's doctor if you have any questions or concerns. Visit [www.doh.state.fl.us/cms/nbscreen](http://www.doh.state.fl.us/cms/nbscreen) for additional information.

## Newborn Hearing Screening

Hearing loss is a defect in the way a newborn hears. Treatment may involve the use of hearing aids or other listening devices and early intervention services.

**AUDITORY BRAINSTEM RESPONSE (ABR):** Small headphones are placed over your baby's ears. The headphones send soft clicking sounds into your baby's ears. Sensors or electrodes placed on different parts of the head measure your baby's responses to sounds.

**OTOACOUSTIC EMISSIONS (OAE):** A tiny earplug is inserted into your baby's ear. The earplug sends soft sounds into the ear and a computer reads the "echoes" that come back from your baby's ear.



Automated Auditory Brainstem Response



Otoacoustic Emissions



### Why should my baby's hearing be tested?

- We test all babies to find the ones who may not be able to hear.
- If we find hearing loss early, there are many ways we can help your baby.
- Make sure your baby's hearing is tested before you leave the hospital.

### What should I know about the Newborn Hearing Test?

- The test is safe and painless and can be done in about 10 minutes.
- The test results are available before you leave the hospital.
- Be sure to tell the hospital the name of your baby's doctor so they can send a copy of the results for your baby's record.

### Why do some babies need another test?

- Some babies may need another test because of: **fluid in the ear; noise in the testing room; baby was moving a lot; or baby has hearing loss.**
- Most babies who need another test have normal hearing. Some will have hearing loss.
- If your baby does not pass the hearing test, make sure he or she is tested again as soon as possible.

### Can a newborn baby pass the hearing test and still have hearing loss?

- Yes, some babies will hear well enough to pass the first test, but lose their hearing later because of: **some illnesses; some medicines; some injuries; and family history of hearing loss.**
- If you are ever worried about how your baby hears or talks, call your baby's doctor right away and ask about getting a hearing test.

Each year in the U.S., as many as 2 to 3 babies out of each 1,000 born, are deaf or hard of hearing.

Watch for signs of hearing loss as your baby grows. Use this list as a guide to things babies with normal hearing should be able to do for their age.

#### BIRTH-3 MONTHS

- **Jumps or blinks to loud sounds**
- **Wakes up to loud sounds**
- **Quiets or smiles when spoken to**
- **Makes vowel sounds like "ohhh," "ahhh"**

#### 3-6 MONTHS

- **Looks for sounds with eyes**
- **Starts babbling "baba," "mama," "gaga"**

#### Responds to your voice

- **Enjoys toys that make noise**
- **Uses a variety of sounds, squeals, chuckles**

#### 6-9 MONTHS

- **Turns head toward loud sounds**
- **Responds to his or her name**
- **Begins to imitate speech sounds**
- **Understands "no-no" or "bye-bye"**

### What about my newborns eyes?

The American Academy of Pediatrics recommends that a physician perform an eye evaluation that includes the red reflex test at all well-infant and well-child visits.