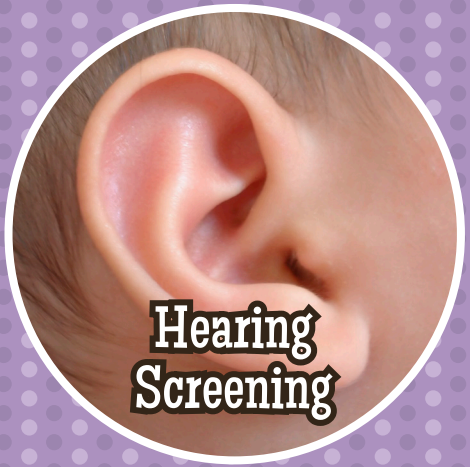
Alabama Newborn Screening Program



DELIVERING YOU THE FACTS

ALABAMA PUBLIC HEALTH

Alabama's Listening! UNIVERSAL NEWBORN HEARING SCREENING

ALABAMA Newborn Screening PROGRAM

Alabama Department of Public Health Newborn Screening Program
www.adph.org/newbornscreening 1-866-928-6755

Delivering You the Facts

What is Newborn Screening?
In Alabama, all newborns are screened for some rare but serious conditions before leaving the hospital. These inherited conditions can lead to intellectual disability, physical disability, and even death if not identified and treated early; however, early identification through newborn screening can help prevent these complications and result in normal growth and development. Babies with these conditions often look healthy at birth and have no known family history.

Although Newborn Screening is intended to detect the presence of genetic conditions, an infant may instead be identified as a "carrier" or have a change in one copy of a gene but not have the disease. Not all carriers will be identified through newborn screening, especially for conditions such as cystic fibrosis. For more information on the impacts of being a carrier visit: http://www.chw.org/medical-care/genetics-and-genomics-program/medical-genetics/single-gene-defects/autosomal-recessive.

Newborn Screening Includes:

- **Blood Spot Screen (heel-stick)** – a lab test to screen for certain inheritable conditions. A sample of blood is taken before 48 hours of age by pricking the baby's heel and placing several drops of blood on special paper.
- **Hearing Screen** – a test to detect hearing loss in newborns. There are two types of screening methods that may be used. Both tests are noninvasive and take only minutes to perform.
- **Pulse Ox Screen** – a test to detect critical congenital heart disease. A probe is placed on both the baby's hand and foot to measure oxygen levels. The test is noninvasive and takes only a few minutes to perform.

Getting Your Baby's Newborn Screening Results
You should let the hospital know who your baby's doctor will be and ask about the newborn screening results at your baby's first doctor visit.

What Does it Mean if My Baby has an Abnormal Newborn Screening Result?
A positive or abnormal result does not mean that your baby has a condition, but that further testing is necessary. Your doctor will be called right away if your baby is identified as needing further testing.

What if My Baby is Identified with a Condition?
Most babies with a newborn screening condition can grow and develop normally if a condition is identified and treated early. Sometimes lifelong treatment and monitoring is needed. It is important to act quickly if your baby needs more tests or requires treatment.

How Many Newborn Screens will my Baby Need?
In Alabama, it is recommended that all full-term infants with a normal first blood spot screen receive a routine repeat blood spot screen between two and six weeks of age. Some infants may have an inadequate blood spot specimen submitted and need to have an additional newborn screen collected. Please follow up immediately to have a repeat newborn screen collected, if requested.

Blood Spot Screen

- **Biotinidase deficiency** - a treatable, inherited condition of biotin. It is caused by the lack of an enzyme called biotinidase. Treatment includes oral biotin.
- **Congenital adrenal hyperplasia** - a family of genetic diseases involving the adrenal glands. There are two forms of CAH: salt-wasting and simple-virilizing. Salt-wasting CAH can cause life-threatening salt loss from the body, if untreated. There are also milder subtypes called non-classical CAH that are not life-threatening but can result in acne, excess growth, and pubertal conditions. Treatment includes salt replacement and hormone replacement.
- **Congenital hypothyroidism** – a condition which occurs when infants are unable to produce enough of the hormone thyroxin or T4, which is necessary for normal metabolism, growth, and brain development. Thyroid problems can be inherited, but typically they occur when the thyroid does not form properly. Prompt identification and treatment can prevent intellectual disability and variable degrees of growth failure, deafness, and neurological abnormalities. The condition is treated with oral doses of thyroid hormone.
- **Cystic fibrosis** - an inherited disease that causes thick, sticky mucus to build up in the lungs, digestive system, and other organs of the body. The mucus can lead to chronic lung infections and difficulty digesting food and nutrients, causing poor growth and development. Treatment may include a

high-calorie diet, respiratory therapy to help clear mucus from the lungs, and medications to improve breathing and prevent lung infections.

- **Galactosemia** – an inherited condition caused by a lack of the enzyme that converts galactose or milk sugar to glucose. This lack of enzyme results in toxic build-up of galactose in the body, and can lead to death if the condition is left untreated. Treatment includes removal of galactose from the diet.
- **Hemoglobin SS disease** – an inherited blood disease, also known as sickle cell anemia, which can cause severe pain, damage to the vital organs, stroke, and sometimes death in childhood. Young children with sickle cell anemia are especially prone to dangerous bacterial infections such as pneumonia and meningitis. Vigilant medical care and preventive treatment with penicillin can dramatically reduce the risk of these adverse effects. Additional treatments may include intermittent pain medications and blood transfusions.
- **Hemoglobin SC disease** - another inherited blood disease that is often milder then Hemoglobin SS Disease for which routine preventive treatment with penicillin is often recommended. Children with Hemoglobin SC disease inherit one sickle cell gene and one gene for another abnormal type of hemoglobin called Hemoglobin C. Although it is not as severe as Hemoglobin SS Disease, it is important to ensure vigilant medical care, especially during illness.
- **Hemoglobin S/beta-thalassemia** - another inherited blood disease in which a child inherits one sickle cell gene and one gene for beta thalassemia. Symptoms are often milder than Hemoglobin SS Disease, although severity varies among affected children. Routine preventive treatment with penicillin may not be recommended for all affected children.
- **Severe combined immunodeficiencies** - a genetic condition in which the immune system does not develop normally. Affected infants are extremely vulnerable to infections. Newborns may appear normal the first weeks of life because they carry their mother's antibodies, but they usually do not survive past their first birthday if not diagnosed and treated early. The most commonly recommended treatment is bone marrow transplantation in the first three months of life.

Metabolic Conditions

Alabama began screening for certain metabolic conditions using tandem mass spectrometry (MS/MS) in October 2004 and has since added metabolic conditions detectable by expanded newborn screening. MS/MS screening does not diagnose infants with conditions; however, it does help to identify infants who may be affected by one of these conditions.

Amino Acid Conditions

- **Argininosuccinic aciduria**
- **Citrullinemia**
- **Maple syrup urine disease**
- **Homocystinuria**
- **Tyrosinemia**
- **Phenylketonuria (PKU)** – an amino acid condition in which affected individuals have an inability to properly process the essential amino acid phenylalanine, which accumulates and damages the brain. PKU can result in severe intellectual disability unless detected soon after birth and treated with a special formula. Treatment includes a low-phenylalanine diet.

Fatty Acid Conditions

- **Carnitine uptake defect**
- **Very Long-chain acyl-CoA dehydrogenase deficiency**
- **Long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency**
- **Trifunctional protein deficiency**
- **Medium-chain acyl-CoA dehydrogenase deficiency** – the most commonly identified fatty acid condition in Alabama. May result in seizures (caused by low blood sugar), liver failure, coma, and death. Identifying affected infants before they become ill is vital to preventing a crisis. Treatment includes avoidance of fasting and nutritional supplements.

Organic Acid Conditions

- Propionic acidemia
- Methylmalonic acidemia (methylmalonyl-CoA mutase)
- Methylmalonic acidemia (cobalamin disorders)
- Isovaleric acidemia
- 3-Methylcrotonyl-CoA carboxylase deficiency
- 3-Hydroxy-3-methyglutaric aciduria
- Holocarboxylase synthase deficiency
- Beta Ketothiolase deficiency
- Glutaric acidemia

For additional information, view the Disorder Descriptions at http://adph.org/newbornscreening.

Hearing Screen

What is Hearing Loss?

There are two main types of hearing loss:

- **Conductive hearing loss** - occurs when sound cannot enter into the inner ear. This may be caused by wax buildup, fluid in the ear, or structural abnormalities. It can usually be corrected with medical or surgical intervention.
- **Sensorineural hearing loss** - occurs when there is damage to the inner ear. This may be caused by disease, birth injury, toxic drugs, or genetic syndromes.

In addition, there are varying degrees of hearing loss. They include mild, moderate, severe, and profound hearing loss. It is important to note that milder hearing losses or hearing losses that affect only one ear may not be apparent; therefore, it is important to follow-up even though a baby may appear to hear normally.

Why Should a Baby's Hearing be Screened?
The first two years of a baby's life are critical for learning speech and language; therefore, it is important to diagnose hearing problems early. In addition, early detection makes talking, learning, and adjusting to hearing devices easier.

How is the Hearing Screen Performed?
There are two types of screening methods that may be used. Both tests are very safe, take only minutes to perform, and are noninvasive. Most babies sleep through the hearing screening.

- **Auditory Brainstem Response (ABR)** - determines the infant's ability to hear soft sounds normally by placing miniature earphones in or over the ears, and attaching electrodes to measure brain wave responses to the sound. This test method is recommended by the Joint Committee on Infant Hearing (JCIH) for high risk newborns admitted to the NICU greater than five days and should be completed as the repeat test method if an infant is initially tested with ABR.
- **Otoacoustic Emissions (OAE)** - measures inner ear function by inserting a small probe into the ear canal and measuring vibrations, or emissions from the ear which are a result of stimulating the cochlea with soft sounds. The absence of emissions indicates the need for additional testing.

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

For additional information, view Newborn Hearing Screening at http://adph.org/newbornscreening.

Pulse Oximetry Screen

What is the Pulse Ox Screen?
Pulse oximetry is a simple, noninvasive test that measures how much oxygen is in the blood. Another term for pulse oximetry is "pulse ox." It is done at 24-48 hours of age.

How is Pulse Ox Performed?
The pulse ox is placed by a sticky strip, like a band-aid™, with a small red light, or "probe," on the baby's hand or foot. The probe is attached to a wire, which is attached to a special monitor that shows the pulse ox reading. The pulse ox test takes just a few minutes to perform when a baby is still, quiet, and warm. If a baby is crying, squirming, or cold it may take longer, or not be possible. You can help comfort your baby and keep him or her warm, calm, and quiet while the test is being performed.

Why is Pulse Oximetry Used?
Pulse ox is routinely used to measure how much oxygen is in the blood. It can be used to monitor an infant's oxygen level during a procedure or treatment, and is helpful in determining if an infant's heart and lungs are healthy. Pulse ox is also used to identify babies with low levels of oxygen in their blood that may have serious heart problems. A doctor or nurse practitioner may order additional testing, such as an ultrasound of the heart, also known as an echocardiogram or "echo", when a low pulse ox reading is identified. The echo will screen for a serious problem in the structure of the heart or the blood flow through the heart. Pulse ox can help identify a baby with critical congenital heart disease before he or she leaves the newborn nursery.

What is Critical Congenital Heart Disease (CCHD)?
CCHD is a problem in the structure of the heart or the blood flow through the heart which requires treatment, usually surgical, in the first year of life. Congenital cardiac defects are the most common birth defect and the cause is not usually known.

What is a Normal Reading?
Pulse ox readings in the right hand and foot that are 95 or higher and have a difference of three or less between the right hand and foot are normal

in healthy children. Children with heart or lung problems may have lower readings. A low pulse oximetry reading can be normal in newborns whose lungs and heart are adjusting after birth. If your child has a problem with his or her heart or lungs, your doctor or nurse will discuss this with you. In addition, your baby's doctor may order additional tests.

Can a Baby with CCHD have a Normal Pulse Ox Reading?
It is possible that the pulse ox test will not detect all forms of problems in the baby's heart. Your baby should continue to have normal visits with his or her primary care doctor. If a problem with the heart is suspected, discuss this with your primary care doctor who will advise you.

For more information, visit http://www.cdc.gov/ncbddd/pediatricgenetics/documents/CCHD-factsheet.pdf.

Family Highlights – Hearing Screening

Meet Ella Kate! Her newborn screening journey began when she failed her newborn hearing screen after five attempts using the automated auditory brainstem response (AABR) hearing screen. She was then referred to an otolaryngologist. At two weeks of age, Ella Kate had another OAE completed by the otolaryngologist and did not pass. She was scheduled to go back in another month, and at that time, failed a third OAE.* She was finally referred to Children's of Alabama after failing multiple hearing screens.

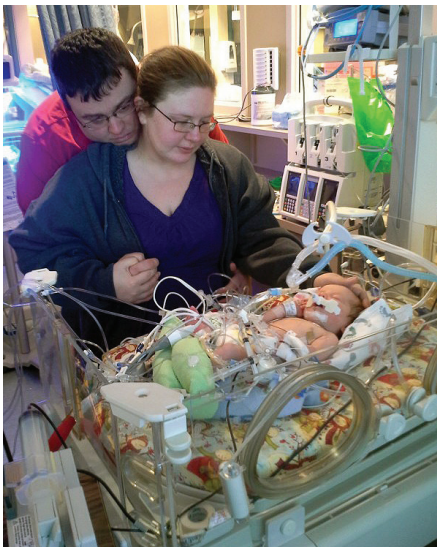
At less than six weeks of age, Ella Kate was diagnosed with moderate to severe hearing loss in her right ear and severe hearing loss in her left ear. According to Ella Kate's mother, Jennifer Hornsby, "It was heart-wrenching. You never imagine life will pan out this way, but God had a plan." Ella Kate

was finally fitted with hearing aids at more than 3 months of age. She started speech therapy, and at 16 months of age, Ella Kate received her first cochlear implant. About a year later, Ella Kate received her second implant. "The cochlear implants have been such a blessing. It has been amazing to see her progress in such a short time. She is not caught up with her peers yet, but we look forward to the day she will be! Every journey is different, but we are so thankful to be able to share ours."

*ADPH complies with the guidelines set forth by the JCIH for newborn hearing screening and follow-up. For additional information on hearing screening guidelines, visit http://www.jcih.org/posstatemts.htm.

Family Highlights – Pulse Oximetry Screening

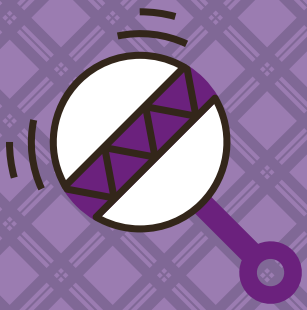
First time parents Kyle and Leslie Ethridge had an uneventful pregnancy and birth of their first child Grayson in December 2012. Grayson was evaluated by his pediatrician after delivery, and the new parents were told everything was fine. The night before they were scheduled to leave the hospital the nurse took Grayson for routine vital signs and also performed the pulse oximetry screen, which revealed a 97% oxygen saturation in his right hand and a 87% oxygen saturation in his foot.



Grayson was quickly transferred to Baptist Medical Center South and then flown to Birmingham for open heart surgery. He was diagnosed with ventricular/atrial septal defect and an interrupted aortic arch. Grayson sees a pediatric cardiologist on a regular basis and has had superb news every visit. According to Mrs. Ethridge, "Family, friends, and faith kept us sane during this trial. We have come out stronger and are very proactive in raising awareness for pulse ox screening to detect congenital heart defects. Newborn screening saved my baby's life."

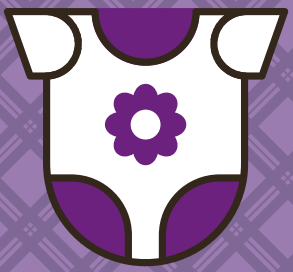
About **1 in 125 newborns** have a **Congenital Heart Defect**

More than **1 in 300 newborns** have a condition detectable through newborn screening

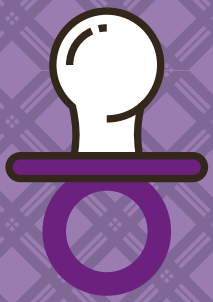


Newborn Screening is one of the **greatest public health achievements** of the 20th century

Each year, **12,000 babies** with serious, but treatable conditions grow up **healthy,** thanks to newborn screening



Every baby born in the United States can undergo newborn screening.



As many as **6 in 1000 newborns** may have **hearing loss** at birth

ALABAMA

Newborn Screening

PROGRAM

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