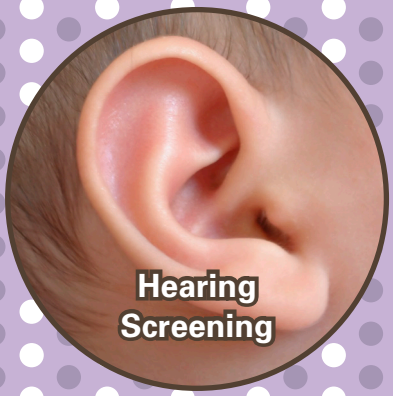
Alabama Newborn Screening Program



Delivering You the Facts

ADPH
Alabama Department of 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

Alabama Newborn Screening Timeline

1964	PKU
1978	Congenital Hypothyroidism
1987	Hemoglobinopathies
1992	Galactosemia
1994	Congenital Adrenal Hyperplasia
1997	Voice Response System (VRS)
04/2004	Biotinidase Deficiency
10/2004	**Amino Acid Disorders** Citrullinemia (CIT) Homocystinuria (HYC) Maple Syrup Urine Disease (MSUD) Tyrosinemia (TYR) Argininosuccinate Aciduria (ASA) **Organic Acid Disorders** Propionic Acidemia (PROP) Methylmalonic Acidemia (Vitamin B12 disorders) (CBL, A, B) Methylmalonic Acidemia (methylmalonyl-CoA mutase) (MUT) Multiple Carboxylase (MCD) **Fatty Acid Disorders** Medium Chain acyl-CoA Dehydrogenase deficiency (MCAD) Carnitine Uptake Defect (CUD)
10/2006	**Organic Acid Disorders** Glutaric Acidemia (OA) Isovaleric Acidemia (OA)
04/2007	**Fatty Acid Disorders** Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD) Long Chain 3-hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD) Trifunctional Protein Deficiency (TFP)
08/2007	**Organic Acid Disorders** 3-Methylcrotonyl-CoA carboxylase (3-MCC) Beta ketothiolase (BKT) 3-Hydroxy 3-methylyglutaric aciduria (HMG)
01/2008	Universal Newborn Hearing Screening*
04/2008	Cystic Fibrosis (CF) (IRT/DNA)
2009	Cord Blood Collection and Testing discontinued
06/2013	Critical Congenital Heart Disease (CCHD)

***started voluntarily in 2001 and mandated 2008**

Delivering You the Facts

What is Newborn Screening?
In Alabama, all newborns are screened for some rare but serious disorders before they leave the hospital. These inherited disorders can lead to death, mental retardation, or physical disability if not identified early. However, early identification through newborn screening can help prevent death and result in normal growth and development. Babies with these conditions may look healthy at birth and have no family history of problems.

Some babies may be found to have a single disease causing gene and be a carrier of certain conditions; however, not all carriers will be identified through newborn screening. You should consult with your health care provider if you have concerns about genetic testing.

Newborn Screening Includes:
👣 Blood Spot Screen (heel-stick) – a sample of blood is taken at 24 – 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 very safe and take only minutes to perform.

♥ Pulse Ox Screen - a test to detect congenital heart defects. A probe is placed on both the baby's hand and foot to measure oxygen levels. The test is very safe 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 disorder, but that further testing is necessary. Your doctor will be called right away if a screen is referred for further evaluation.

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

How Many Newborn Screens will my Baby Need?
In Alabama, it is recommended that an infant receive a routine second newborn screen at two to six weeks of age on all full term infants with a normal first test screen. Some infants may have an unreliable blood spot screen submitted and may be asked to repeat the newborn screen. Please follow up immediately to have a repeat newborn screen if requested.

Blood Spot Screen

- **Biotindase Deficiency** - a treatable, inherited disorder of biotin. It is caused by the lack of an enzyme called biotinidase. Treatment includes oral biotin for lifetime.
- **Congenital Adrenal Hyperplasia (CAH)** - a family of genetic diseases involving the adrenal glands. There are two forms of CAH: salt-wasting and simple-virilizing. Salt-wasting CAH may 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 disorders. Female infants may present with ambiguous genitalia. Males have normal genitalia but later experience advanced growth and puberty. Treatment includes salt replacement and hormone replacement.
- **Congenital Hypothyroidism (CH)** - occurs when infants are unable to produce enough of the hormone thyroxin or T4, which is necessary for normal metabolism, growth, and brain development. Occassionally thryoid problems are inherited, but typically it is a case of the thryoid not forming properly. Infants who are not identified and treated promptly suffer mental retardation and variable degrees of growth failure, deafness, and neurological abnormalities. The condition can be treated with simple oral doses of thyroid hormone.
- **Cystic Fibrosis (CF)** - an inherited disease that cause 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. Studies have shown babies diagnosed with CF through newborn screening benefit from better nutritional status than babies whose diagnosis is delayed. 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** - inherited disorder caused by a lack of an enzyme that converts galactose or milk sugar to glucose. A lack of the enzyme results in toxic buidlup of galactose in the body and can lead to death if the disorder is untreated. Treatment includes removal of galatose from the diet.
- **Hemoglobin SS Disease** - a blood disease that 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 treatment with penicillin can dramatically reduce the risk of these adverse effects. Affected babies should receive all regular childhood vaccinations including hemophilus influenza B and pneumococcal in order to prevent serious bacterial infections. Additional treatments may include intermittent pain medications and regular blood transfusions.
- **Hemoglobin SC Disease** - another form of sickle cell disease that is often milder then Hemoglobin SS Disease, and routine penicillin treatment may not be recommended. Children with Hemoglobin SC disease inherit one sickle cell gene and one gene for another abnormal type of hemoglobin called HbC.
- **Hemoglobin S/beta-thalassemia**- In this form of sickle cell anemia, the child inherits one sickle cell gene and one gene for beta thalassemia. Symptoms are often milder than HbSS, though severity varies among affected children. Routine treatment with penicillin may not be recommended for all affected children.

Blood Spot Screen

Metabolic Disorders:

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 disorders but simply picks out which infants may be more likely to have one of these disorders.

Amino Acid Disorders

- Citrullinemia
- Homocystinuria
- Maple Syrup Urine Disease (MSUD)
- Tyrosinemia
- Argininosuccinate Aciduria (ASA)
- Phenylketonuria (PKU) - amino acid disorder in which affected individuals have an inability to properly process the esstential amino acid phenylalanine, which accumulates and damages the brain. PKU can result in severe mental retardation unless detected soon after birth and treated with a special formula. Treatment includes a low-phenylalanine diet at least throughout childhood, adolescence, and for females during pregnancy.

Fatty Acid Disorders

- Carnitine Uptake Defect (CUD)
- Carnitine palmitoyl transferase 2 (CPT2)
- Medium chain acyl-CoA dehydrogenase deficiency (MCADD) - most commonly identified fatty acid disorder 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.
- Long chain L-3 hydrosyacyl-CoA dehydrogenase deficiency (LCHADD)
- Very long chain acyl-CoA dehydrogenase deificiency (VLCADD)
- Trifunctional protein deficiency (TFP)

Organic Acid Disorders

- Propionic Acidemia
- Methylmalonic Acidemia (CBL, A, B)
- Methylmalonic Acidemia (methylmalonyl-CoA mutase)
- Multiple carboxylase (MCD)
- Glutaric Acidemia (GA-1)
- Isovaleric Acidemia (IVA)
- 3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC)
- 3-Hydroxy-30methyglutaric aciduria (HMG)
- Beta ketothiolase deficiency (BKT)

Please visit the Alabama Newborn Screening website at www.adph.org/newbornscreening to view descriptions of these rare conditions.

Hearing Screen

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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 hearng loss** - occurs when there is damage to the inner ear. This may be caused by diseases, birth injury, toxic drugs, viruses, or genetic syndromes.

In addition, there are various 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. Thus, 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. Thus, it is important to diagnose hearing problems early because a hearing loss could affect a baby's speech and language development. In addition, early detection makes talking, learning, and adjusting to hearing devices easier.

How is the Hearing Screen Performed?
There are two types of screening methods that may be used. Both tests are very safe, take only minutes to perform, and are non-invasive. Most babies sleep through the hearing screening.
- **Auditory Brainstem Response (ABR)** - determines the infant's ability to hear soft sounds normally by inserting miniature earphones and attaching electrodes to measure brain wave responses to the sound. This diagnostic testing is recommended by the Joint Committee on Infant Hearing (JCIH) for high risk newborns admitted to the NICU greater than five days and should be completed as a second test method if an infant is initially tested with ABR.
- **Otoacoutstic Emissions (OAE)** - measures inner ear function by inserting a miniature microphone in the ear canal via a soft probe tip and measuring tones from the ear by sending responses to a special computer.

What if a Baby does not Pass the Hearing Screen?
If a baby does not pass the first hearing screen then an attempt should be made to repeat the screen 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.

Family Highlight: Hearing Screening

Meet the Hornsby Family! Their newborn screening journey began when their baby girl, Ella Kate, failed her newborn hearing screen after five attempts using an automated auditory brainstem response (AABR) hearing screen. At two days of age, Ella Kate failed an otoacoustic emissions (OAE) hearing screen at her pediatrician's office. 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 by the JCIH for newborn hearing screening and follow-up. These guidelines specify that infants who fail an AABR must have a repeat AABR for follow-up screening, and both ears should be tested even if only one ear failed. Infants who fail an initial OAE may be re-screened with either OAE or AABR, since the AABR is a more sensitive and comprehensive test. No more than two valid initial attempts should be performed. If the infant fails both, then a referral for a diagnostic hearing evaluation should be made as soon as possible.

Pulse Oximetry Screen ♥

What is the Pulse Ox Screen?
Pulse oximetry is a simple and painless 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 used to measure how much oxygen is in the blood. Pulse ox is routinely used and can be used to monitor an infant's oxygen level during a procedure or treatment. It can also be helpful in determining if an infant's heart and lungs are healthy. Pulse ox can also help to identify babies with low levels of oxygen in their blood that may have serious heart problems. A doctor or nurse practitioner may ask for more testing such as an ultrasound of the heart, or 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 a serious CHD before he or she leaves the newborn nursery.

What is Congenital Heart Disease (CHD)?
CHD is a problem in the structure of the heart or the blood flow through the heart. CHD is the most common birth defect and the cause is not really known.

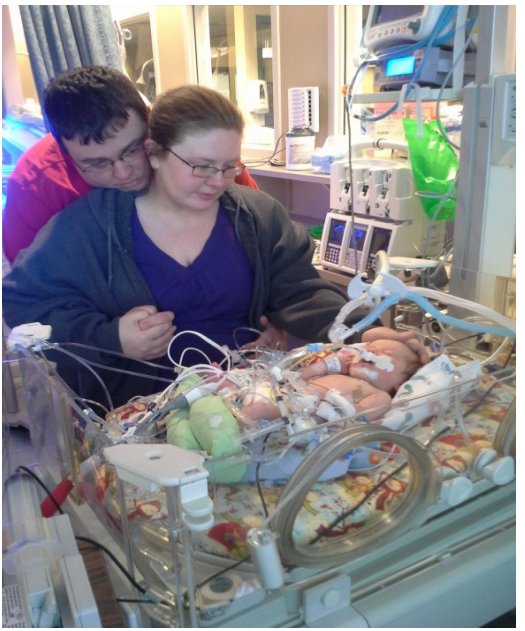
What is a Normal Reading?
Pulse ox readings in the right hand and foot that are 95 or higher and have a difference of three or less between the right hand and foot are normal in healthy children. Children with heart or lung problems may have lower readings. A low pulse oximetry reading can be normal 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 tell you what a normal pulse ox range is for your child. It is possible that your baby's doctor will order additional tests.

Can a Baby with Serious CHD 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, your primary care doctor will advise you.

For more information please visit
www.cdc.gov/ncbddd/pediatricgenetics/CCHDscreening.html

Family Highlight: 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."

Alabama NBS Confirmed Diagnoses

Disorders	2007	2008	2009	2010	2011	2012	2013
Biotinidase Deficiency	1	0	1	0	0	0	0
Classical Galactosemia	2	0	0	4	0	3	1
Cystic Fibrosis	*	7	23	13	19	14	12
Hearing Loss	55	43	29	31	68	60	54
Critical Congenital Heart Disease	*	*	*	*	*	3	2
Endocrine Disorders							
Congenital Hypothyroidism	18	16	30	33	29	32	36
Congenital Adrenal Hyperplasia	1	7	5	3	5	4	2
Hemoglobinapathy							
Sickle Cell Disease	51	57	62	56	66	52	53
Sickle Cell Trait	2038	1818	1940	1860	1835	1866	1817
Amino Acid Disorders							
Phenylketonuria	6	3	4	3	5	2	4
Homocystinuria	1	1	0	0	0	0	0
Maple Syrup Urine Disease (MSUD)	0	0	0	0	0	0	1
Citrullinemia	0	0	0	0	0	0	0
Tyrosinemia	-	-	-	1	0	0	0
Fatty Acid Disorders							
Carnitine Uptake Defect	1	2	3	0	2	1	1
Medium chain Acyl-CoA Dehydrogenase Deficiency (MCAD)	3	3	6	5	5	4	3
Long chain Acyl-CoA Dehydrogenase Deficiency (LCAD)	0	0	0	0	0	0	1
Very long chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)	1	0	0	0	1	1	1
Trifunctional Protein Deficiency	0	0	1	0	0	0	0
Organic Acid Disorders							
Glutaric Acidemia	1	1	0	0	1	0	0
Isovaleric Acidemia	0	0	0	0	0	0	0
Propionic Acidemia	0	0	0	1	0	0	0
Methylmalonic Acidemia (MMA)	0	2	2	0	1	1	1
3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC)	0	0	1	0	0	2	0
2-Methylbutyryl-CoA Dehydrogenase Deficiency	0	1	0	0	0	1	0

* Disorder not included in screening panel

References

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2. General Accounting Office. Newborn Screening: Characteristics of State Programs. Washington, DC: General Accounting Office, 2003. Publication GAO-03-449. Data from the National Newborn Screening and Genetics Resource Center.
3. American Academy of Pediatrics Section on Hematology/Oncology Committee on Genetics. Health Supervision for Children with Sickle Cell Disease, Pediatrics, volume 109, number 3, March 2002, pages 526-535.
4. National Center for Hearing Assessment and Management, Utah State University.
5. Children's National Medical Center. Congenital Heart Disease Screening Program Toolkit: A Toolkit for Implementing Screening. Washington, D.C.: Children's National Medical Center; 2009.

Notes:

Alabama Newborn
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