

Newborn Screening



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Visit the NBS website at:

www.adph.org/newbornscreening/

Visit the following links:

www.babysfirsttest.org/

www.savebabies.org/video.html



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Congenital Hypothyroidism

Helping Alabama's Children: An Update on Congenital Hypothyroidism by the Children's of Alabama Pediatric Endocrinology Newborn Screening Partners

Thanks to its network of Newborn Screening partners, Alabama has one of the cutting edge screening programs for detecting and treating congenital hypothyroidism in the United States. Currently our state identifies 30 to 40 infants each year with congenital hypothyroidism, a condition present in infants that results from partial or complete loss of thyroid function. Through early detection and treatment, these children are given the gift of a normal, healthy life. It is estimated that by detection, 160 cases of severe intellectual disability are prevented in the United States each year, saving the health care system an estimated 195 million dollars annually.

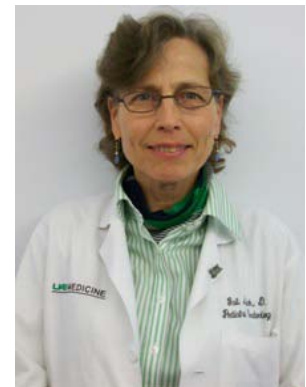
Proper screening for congenital hypothyroidism cannot be emphasized enough. It is well known that the plasma TSH level increases dramatically in normal newborns in the hours after birth, peaking at 24 hours of life. This TSH peak is followed by a slight increase in the plasma T4 during the second day of life. Thus, the timing of newborn screen collection is particularly important. When a screen is collected during the first 24 hours of life, especially during the first hour of life, there is a much higher risk of obtaining a false positive screen. Research has also shown that a second newborn screen, collected from two to four weeks of life, can be critical in detecting congenital hypothyroidism. When in doubt, a serum TSH and Free T4 can confirm the diagnosis of congenital hypothyroidism much faster than a repeated newborn screen.

The screening process for congenital hypothyroidism may involve several steps. Typically, once the diagnosis is confirmed, treatment with Levothyroxine is initiated right away. Each day that treatment is delayed can be critical for a newborn's growth and development. After diagnosis, follow-up with a pediatric endocrinologist is recommended within the first month of treatment. Thyroid function is followed every six to eight weeks for the first year of life, and a thyroid ultrasound is obtained to help differentiate the origin of the child's congenital hypothyroidism.

While some children can stop treatment at three years of age, many have no thyroid function for the remainder of their lives and will receive pediatric subspecialty care for many years. A very close relationship is built between the primary care provider, the pediatric endocrinologist, and the patient and family. Often, if any developmental delays are identified, the child is referred early for intervention and usually will go on to have normal, healthy growth and development.

The Children's of Alabama Endocrinology Clinic has made many resources available to pediatric practices and patients alike at <https://www.childrensal.org/endocrinology>. Just click on the tab for "Newborn Screening" located on the left of the screen. You can reach the clinic staff regarding patient concerns by calling (205) 638-9107 and asking to speak with the clinic's newborn screening coordinator or the endocrinologist on call. It is our desire to be an easily accessible resource for Alabama's birthing facilities and medical providers.

Thank you for all that you do to keep Alabama's children healthy! Together we can make a difference in children's lives.



Gail Mick, MD
Children's of Alabama
Pediatric Endocrinology



Leslie N. Pitts, CRNP
Children's of Alabama
Pediatric Endocrinology



NBS letters soon to be faxed ...

The Alabama Newborn Screening Program is currently working on a system that will automatically fax letters to providers as opposed to mailing out letters. Be on the lookout for provider letters to be sent via fax.

Newborn Screening Nurse Survives MDA Lock-Up



SERATIA JOHNSON, NBS NURSE COORDINATOR, SUPPORTED THE MUSCULAR DYSTROPHY ASSOCIATION (MDA) BY AGREEING TO BE "ARRESTED" FOR HAVING A "BIG HEART" IN THE MDA LOCK-UP FUNDRAISER. SERATIA RAISED OVER FOUR-HUNDRED DOLLARS FOR THIS YEAR'S EVENT.

AAP Meeting & Fall Update 2012

The Alabama Newborn Screening Program recently exhibited at the 2012 Annual Alabama Chapter of the American Academy of Pediatrics (AAP) Meeting & Fall Update, held at the Wynfrey Hotel in Birmingham, September 28-30.

There were approximately 130 physicians and other pediatric clinicians in attendance. Some of the educational topics included Pulse Oximetry Screening in Healthy Newborns and Hearing Screening in the Medical Home.



Dr. Heather Taylor (left), University Medical Center, and Jill Smith, Au.D, CCC-A (right), Audiologist, Children's of Alabama, were in attendance at the 2012 AAP Meeting.



Cindy Ashley (left), NBS Program Director, and Rachael Montgomery (right), NBS Nurse Supervisor, at the 2012 AAP Meeting and Fall Update.

VAUGHN REGIONAL MEDICAL CENTER PARTICIPATES IN NBS TRAINING

The Alabama Newborn Screening Program recently conducted training at Vaughn Regional Medical Center (VRMC) on September 20, 2012. There was approximately twenty nursing staff that participated in either the morning or afternoon training sessions. The purpose of the training was to provide a review of proper collection techniques and address specific issues identified with specimen collection.

Seratia Johnson, Newborn Screening Nurse Coordinator, conducted the training held at VRMC. Approved standards for newborn screening specimen collection as outlined by the Clinical and Laboratory Standards Institute (CLSI) were presented. The training included a Powerpoint presentation and brief video that was followed by the observation of some of the nursing staff collecting specimens.

Participants had the opportunity to ask questions and provide feedback. One nurse commented on how great a specimen may look after it is collected, but then it will come back unsatisfactory. According to Seratia, "It is important to allow specimens to dry first and review a second time prior to sending to the Bureau of Clinical Laboratories. A specimen may appear uniform when wet, but then dry and reveal uneven saturation."

A common question from participants is "Do you have to start over if the drops do not absorb in the indicated circle?"



Nurses at VRMC listen as Seratia Johnson, NBS Nurse Coordinator, conducts her training presentation.

Because the circles are meant as a guide for appropriate size and number of blood spots, you do not need to start over as long as you have collected the indicated amount of blood without overlapping the drops.

One thing Seratia would like training participants to take away with them is to understand that their collection technique will not improve overnight, and it takes practice to become proficient with newborn screening specimen collection.

The Alabama Newborn Screening Program has been committed to the improvement of specimen collection and continues to work with providers across the state to assist with training if needed. For further information regarding newborn screening specimen collection training, please contact Seratia at 334-206-5729.

Family Highlight



Dr. Jeb Hornsby with wife Jennifer, Caden, John Luke, and Ella Kate

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

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

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. Ella Kate is progressing. She's gone from expressing around 20 words at the beginning of August 2012 to about 175 words in October 2012. 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" reports Mrs. Hornsby.

ADPH complies with the guidelines set by the Joint Committee on Infant Hearing (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 an 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. For further information, please visit the following link: www.jcih.org.

ROUTINE SECOND SCREENING IN NEWBORNS

The Alabama Newborn Screening Program is often asked if a second newborn screen or "B" screening is mandatory. It has always been the policy of the Alabama Department of Public Health to recommend second test screening between two and six weeks of age on all babies.

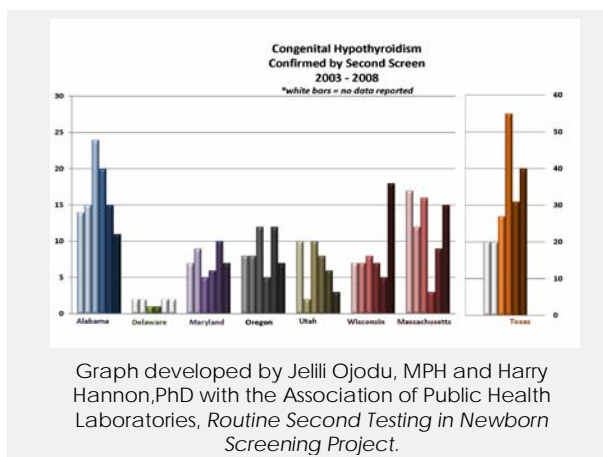
In Alabama, only the initial newborn screen is mandated; however, the vast majority of medical providers in the state have made it practice to collect a routine second newborn screen at two-six weeks of age.

According to Danita Rollin, Laboratory Division Manager with the Bureau of Clinical Laboratories, "Our policy is to provide the best standard of care possible for babies in the state. There is a potential for a newborn to present with problems after the first

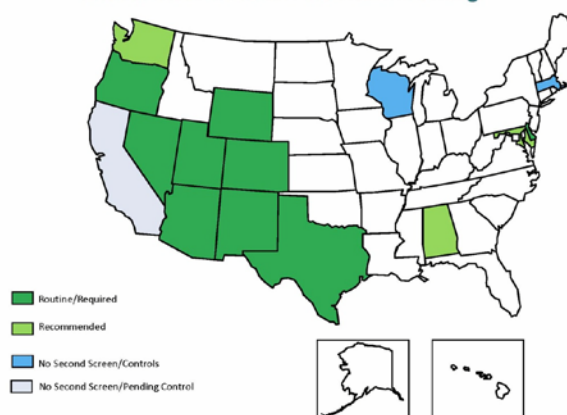
specimen has been collected, tested, and reported as normal. This has become a more urgent issue since many states have expanded their screening panels."

In Alabama, six newborns were identified on a repeat screen with Congenital Hypothyroidism (CH) and one with Congenital Adrenal Hyperplasia (CAH) in 2011. In addition, there was one infant born in Alabama this year that had an initial normal screen and identified with Cystic Fibrosis from a second newborn screen obtained at two weeks of age.

According to a study completed by the Association of Public Health Laboratories, "Scientific literature indicates that cases of CH and CAH are missed on the initial screens that are detected by a routine second screen."



States with Routine Second Screening



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Meet ADPH's Newborn Hearing Coordinator



The Alabama Newborn Screening Program is excited to welcome Ada Wall, BSN, RN, as the Newborn Hearing Screening Coordinator for the Alabama Department of Public Health. Ada comes to the Newborn Screening Program from the Bureau of Home and Community Services with over 15 years of nursing experience.

According to Ada, "I have some big shoes to fill but am looking forward to the challenge." Ada is glad to be a part of the Newborn Screening Program and plans to continue efforts to improve the hearing services provided to Alabama's babies.



Ada Wall, BSN, RN
Newborn Hearing Screening Coordinator

About Our Organization



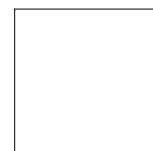
Newborn screening is mandated by Statutory Authority Code of Alabama 1975, Section 22-20-3. The Alabama Department of Public Health is responsible for administrative oversight of the Alabama Newborn Screening Program, which establishes protocol to ensure early identification

and follow-up of infants affected with certain genetic or metabolic conditions. Early diagnosis may reduce morbidity, premature death, mental retardation, and other developmental disabilities.

The program works in partnership with pediatric

sub-specialists throughout the state to ensure all babies identified with abnormal results receive appropriate follow-up. The Alabama Bureau of Clinical Laboratories is the sole provider in the state for the blood analysis of newborn screening.

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