Volume 6, Issue 3 October 2011

http://health.state.tn.us/NBS/index.htm

State of Tennessee Department of Health

Newborn Screening Newsletter

NBS Metabolic Follow Up
Phone: 615-262-6304
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NBS Hearing Follow Up
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Acylcarnitines and Amino Acids

When follow up nurses from the State Lab call positives, they frequently call abnormal acylcarnitines and amino acids. An acylcarnitine is simply the analyte that is found in the blood indicating the level of fatty and/organic acids. An amino acid is critical to life, and has many functions in metabolism. The concentrations of various acylcarnitines and amino acids are measured using advanced techniques of tandem mass spectrometry or MS/MS.

Fatty Acid Oxidative Disorders (FAOD) are inherited conditions that affect the way a person's body breaks down certain fats (fatty acids). A person with a FAOD cannot break down their stored fat for energy. Consequently, the body begins to fail once the food the person has eaten runs out. In addition, fatty acids build up in the blood. In the case of FAODs, the inability to break down fats for energy and the buildup of fatty acids can cause serious health problems.

The symptoms of these conditions vary and depend on the type of fatty acid. Without treatment, children with an fatty acid oxidation disorder may experience periods of poor feeding, lack of energy, difficulty breathing, low blood glucose (sugar) and vomiting. These episodes can become serious enough to lead to developmental delay, seizures, coma and even sudden death.

Organic acidemias are inherited conditions that affect the way a person's body uses protein. A person with an organic acidemia cannot properly break down certain components of protein for energy, growth, and development. Typically, these compounds are amino acids that are not completely broken down. Since the body cannot properly break down these amino acids, certain organic acids build up in the blood and urine. High levels of certain organic acids can cause serious health problems. The symptoms of these conditions vary and depend on the type of organic acidemia.

Amino acidemias are inherited conditions that affect the way a person's body uses a part of food called amino

(Continued on page 2)

Special Points of Interest

Tennessee Child Health Week
OCTOBER 3-9, 2011

Genetics Advisory Committee Meeting
OCTOBER 20, 2011

Upcoming State Holidays

- Nov. 11: Veterans Day
- Nov. 24-25: Thanksgiving
- Dec. 25: Christmas Day
- Jan. 1: New Years Day

GAC

The second Genetics Advisory Committee (GAC) Meeting will be held at the Department of Health State Laboratory Building in Nashville, TN on October 20, 2011. We have lots of interesting and new things to discuss.

Acylcarnitines and Amino Acids (continued)

(Continued from page 1)

acids. A person with an amino acidemia cannot breakdown a specific amino acid in food. Amino acids are needed for proper growth and development, but too much can cause serious health problems. In the case of an amino acidemia, a specific amino acid(s) builds up in the blood, and may penetrate and damage the brain and other organs of the body. The high levels of amino acid(s) ultimately cause serious health problems.

In many cases, therapy with special or restricted diet and/or prescription medication is used. Each treatment depends on the specific disorder. Children and adults with these disorders require follow-up care at a medical center or clinic that specializes in these types of metabolic conditions. Regular blood tests are used to monitor an individual's health.

The chart below highlights some of the more common analytes detected on Tandem Mass Spectrometry (MS/MS).

For more information go to: http://health.state.tn.us/nbs/information.htm

TANDEM MASS SPECTROMETRY (MS/MS)			
PRIMARY ANALYTE	**ANALTYE TYPE**	**DISORDER GROUP**	**DISORDER**
C5-OH	Acylcarnitine	Organic Acidemia	3 Methyl Crotonyl CoA Carboxylase Deficiency (3MCC)
C5	Acylcarnitine	Organic Acidemia	Isovaleric Acidemia (IVA)
C16-OH, C18:1-OH	Acylcarnitine	Fatty Oxidative	Long Chain Hydroxyl AcylCoA Dehydrogenase Def. (LCHAD)
C8	Acylcarnitine	Fatty Oxidative	Medium Chain AcylCoA Dehydrogenase Deficiency (MCAD)
C4	Acylcarnitine	Fatty Oxidative	Short Chain AcylCoA Dehydrogenase Deficiency (SCAD)
Citrulline	Amino Acid	Amino Acidemias	Citrullinemia (CIT)
Methionine	Amino Acid	Amino Acidemias	Homocystinuria (HCY)
Leucine	Amino Acid	Amino Acidemias	Maple Syrup Urine Disease (MSUD)
Phenylalanine	Amino Acid	Amino Acidemias	Phenylketonuria (PKU)

Child Health Week: October 3 - 9, 2011
"A Healthy Tennessee Begins With A Healthy Me"

The health and well-being of our children is fundamental to the future progress of our state. As Child Health Day, a national event first proclaimed by President Calvin Coolidge in 1928, celebrates its 84th observance this year, the State of Tennessee has set apart a special week to focus on the health and well-being of its children.

Many efforts are in place across Tennessee to promote the health of children. For example, programs like GetFitTN, Project Diabetes and Coordinated School Health offer community-driven approaches to partnering with families, teachers and other caregivers. Initiatives like TENNderCARE, CoverKids, and Children's Special Services ensure that children have access to recommended preventive care and resources needed to manage childhood illnesses. Other programs like WIC, the State Immunization Program and the Newborn Screening Program make sure young children get a healthy start on life. Collectively, these efforts, along with many others, make it easier for parents to help children live healthier lives and prepare for a healthy future.

Every Tennessean can have a positive impact on the health and well-being of children. Parents can help their kids stay active and encourage healthy eating habits. Schools and communities can create environments that foster healthy lifestyles. Health providers can promote regular checkups and immunizations and help children and families manage childhood diseases. Together, we can secure a healthier future for all of Tennessee's children.

For more information: http://health.tn.gov/MCH/CHW.shtml

Department of Health State Laboratory

Our Newborn Screening laboratory standard has always been to report all suspected positive results to the Maternal and Child Health (MCH) Follow-up group, generally within 1 to 2 working days after the specimen is received in the laboratory. The follow up program then notifies the provider by telephone to initiate treatment of the patient, confirmation testing and follow-up of the patient. The reports of normal and presumptive positive specimens are generally mailed within 5 to 7 working days from time of receipt. We felt positively that we could improve our turnaround time for reporting results.

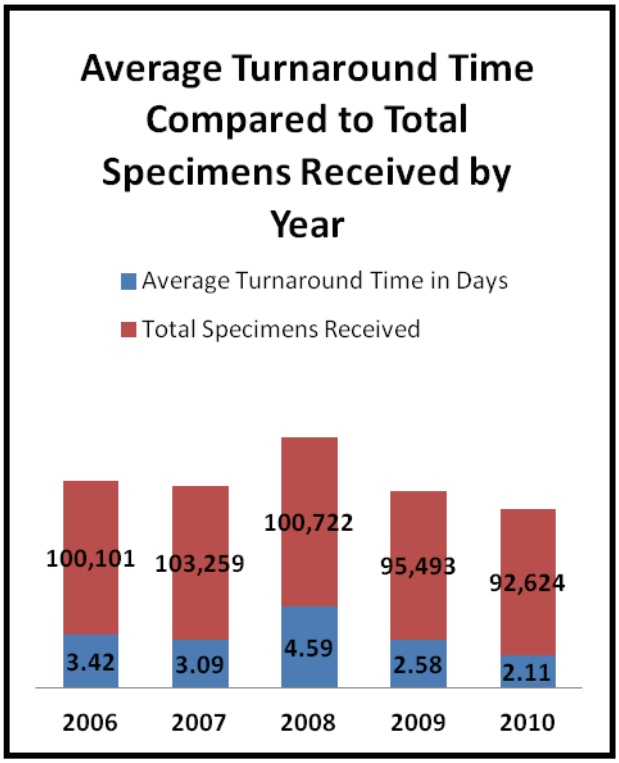
Division of Laboratory Services Newborn Screening is glad to report a significant decrease in turnaround time (TAT) for reporting out screening results.

To achieve this goal several factors had to come together. We looked at our process and refined it to meet the goal of reporting and mailing specimens to include normal and presumptive positives in less than 2.5 days after the receipt of specimen. Once the specimens are received, they are removed from the envelopes. Each specimen gets a date and time of receipt stamp. Each specimen is checked twice by two different techs for quality and quantity. Next, a unique identifier (Tennessee Department of Health lab number) is assigned to each sample. Subsequently, the blood cards are separated from the form then punching and processing of each specimen occurs. Next, data entry staff enters into Natus our laboratory information management system (LIMS) demographic information from the specimen form. This information once keyed is verified for accuracy and completeness by double entry by a different data entry staff member.

Once all specimens received on a particular day are keyed and verified, the lab is notified that the batch is complete and ready for reporting. Data entry consistently meets a goal keying and verifying all specimens received the day before by 10 a.m. the following day. A final demographic check is done by the lab into the LIMS then results are merged and released to follow-up. Lab reports have a set run time to print at the end of the work day. Lab reports are reviewed for accuracy by NBS management and are retrieved by data entry staff where they are readied for mail pickup by 9:30 a.m. The major refinement came from the instituting of merging results into our LIMS and the cooperation of our data entry staff members ensuring that demographics are keyed in a timely fashion. Based on this, the chart below illustrates how successful we have been in decreasing our turnaround time.

Our future goal for the lab is to continue to decrease our turnaround time to reporting out results the very next day as well as web based demographic entry and reporting.

By Thomas Childs, B.S. MT (ASCP)
Newborn Screening Manager ❖



Welcome

Claudia Weber, M.S.Ed.
Tennessee Early Intervention System (TEIS)
Newborn Hearing Liaison

Claudia has been with TEIS for a number of years and began as the newborn hearing liaison in July 2011. She contacts families in need of further hearing screening. She has the ability to report follow-up by a remote data link to the Newborn Hearing Screening Program Neometrics data system.

Early Childhood Hearing Outreach (ECHO) in Early Head Start Agencies

Three Early Head Start agencies participated in the ECHO training held 8-19-11 at the Signal Center in Chattanooga. Centers include Northwest Early Head Start, Chattanooga Early Head Start, and Knoxville Early Head Start. These centers serve over 250 infants and toddlers to age 3 years.

Participants learned how to conduct hand held automated otoacoustic emissions (OAE) screening on children enrolled in several Head Start Centers in the Chattanooga area. The plan is to incorporate the hearing screening in all early head start centers. Not all hearing loss can be indentified at birth. Periodic, objective screening throughout early childhood is also important because gradual or sudden hearing loss can occur at any age. Some infants and children have risk indicators that place them at a greater risk for developing a hearing loss. Failing to recognize hearing loss may result in delayed development of language, social and cognitive skills.
http://www.kidshearing.org

Medical Providers

Newborn Hearing Guidelines
for Pediatric Medical Home Providers

- **Newborn:** Assure infant receives a newborn hearing screen.
- **1 month:** Obtain a repeat hearing screen if initial screen did not pass. This may be completed by the medical provider, as an out-patient at some hospitals, or by a pediatric audiologist. Report retests to the Hearing Program.
- **3 months:** If infant does not pass rescreen, refer for a diagnostic evaluation by a pediatric audiologist. If diagnosed with hearing loss, make referrals for evaluation by otology, genetics, ophthalmology, and early intervention (TEIS). Report results to the Hearing Program.
- **6 months:** Assess if child with hearing loss is enrolled in early intervention services and if referrals for other evaluations complete.
- **Infants with RISK INDICATORS:** JCIH standards recommend infants and toddlers identified with risk indicators for progressive and delayed onset hearing loss be evaluated at periodic intervals every 6 -12 months until the age of three years old.
- **Refer to AAP algorithm for Early Hearing Detection and Intervention (EHDI)**
  http://www.medicalhomeinfo.org

Pediatric Audiologist

Reminders for audiologists/otolaryngologists:

- Report infant hearing evaluations on children birth – 3 years to the Newborn Hearing Screening (NHS) Program
- Report hearing risk follow-up to NHS
- Report all hearing loss on children birth to 3 years to NHS

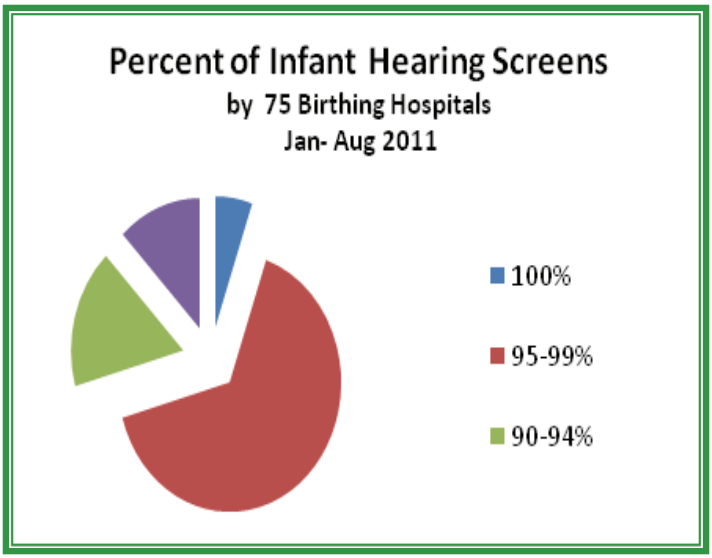
Pediatric Hearing Providers

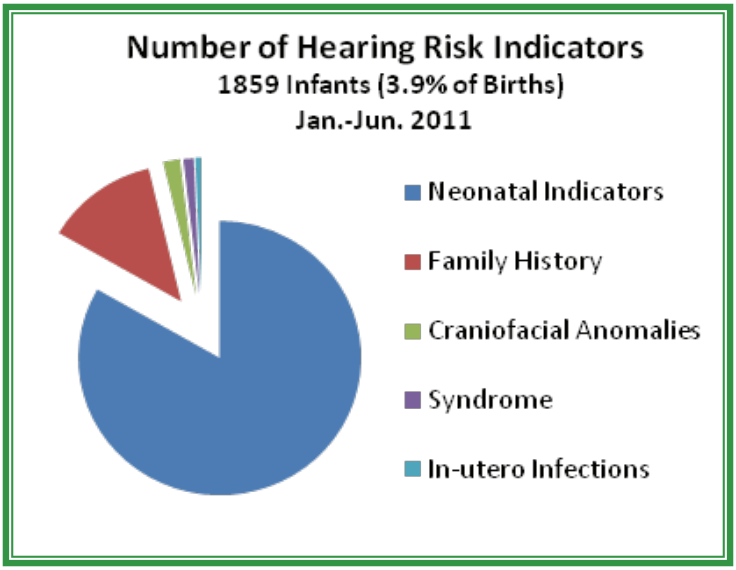
- List of providers available for EAST, MIDDLE and WEST TN
- This resource is sent to parents in their follow-up letters.

Hospitals

Reminders for hospital hearing screeners:

- Complete all information on the report form.
- Document a screen for both ears. If unable to screen one ear list as REFER.
- Do not screen more than 3 times at one test period. Remember this is only a "screen".
- Assess and document hearing risks.
- Report reason for no screen - infants that have transferred, refused, still in the hospital, or expired.





Hearing Loss
Jan. – June 2011

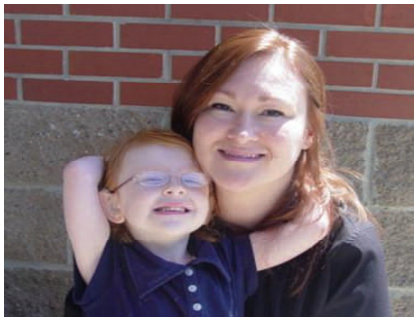
- 1,437 (3.7%) infants did not pass the initial hearing screen
- 25 infants identified with hearing loss.
- 70 pending results after at least one audiology evaluation.

Family Support

TENNESSEE HANDS & VOICES™

www.handsandvoices.org

Tennessee now has an official chapter of Hands &Voices dedicated to supporting families of children who are deaf and hard of hearing without bias in communication methods or methodology.



Melanie Bacon – Lead Parent TN H&V
And daughter Morgan

Start-up Family Activities were held:

- Nashville 9-18-10
- Rutherford Co 6-18-11
- Humboldt 7-9-11
- Knoxville 8-14-11
- Memphis -9-10-11

http://www.tnhandsandvoices.org
901-235-2021
tnhandsvoices@yahoo.com

FAMILY VOICES of Tennessee

part of a national grassroots network of families and friends speaking on behalf of children with special health care needs

1-888-643-7811

Parent Support and Advocacy by parents of children with hearing loss.

- East TN-Camille Keck - camille_k@tndisability.org
- Middle TN–Tonya Bowman - tonya_b@tndisability.org
- West TN-Jenny Williams - jenny_w@tndisability.org
- West TN-Treva Sease - treva_s@tndisability.org

EHDI

The Early Hearing Detection and Intervention Program

The CARE Project Training

Facilitates acceptance and advocacy for families after the diagnosis of hearing loss.

By Dr. Johnnie Sexton, AuD
www.thecareproject.me

This program is for families and professionals and shares experiences by families and professionals.

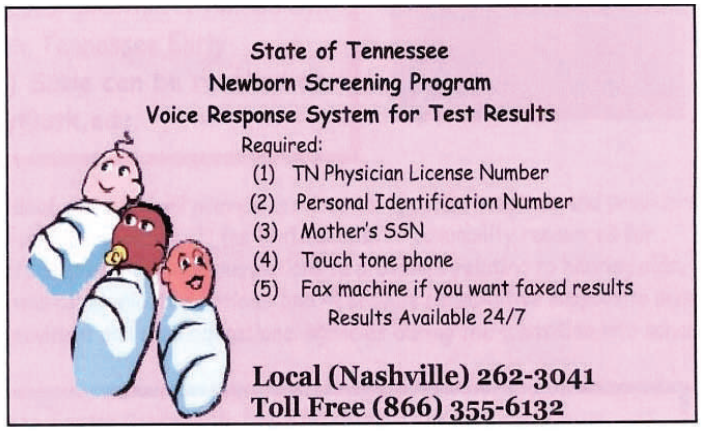
Nashville: August 19, 2011
Bill Wilkerson

Memphis: October 18, 2011
LeBonheur
8-4:30 Cindy Gore
mlh.cont.ed@gmail.com

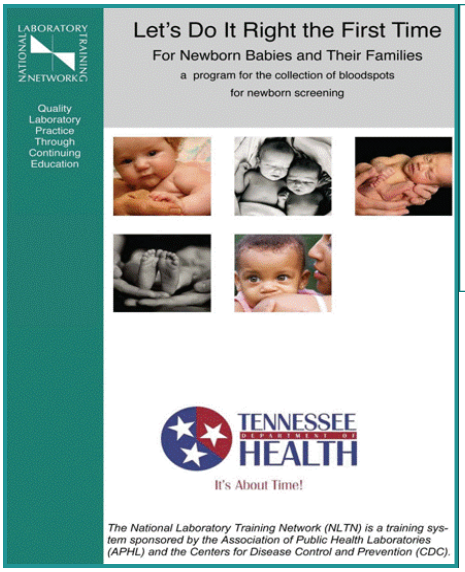
Knoxville: October 20, 2011
School for the Deaf
8-4:30 Susie McCamy
865-974-4137
smccamy@utk.edu

Visit the State of Tennessee Department of Health Newborn Screening Program Website

Information available for Provider, Parents and Hospitals.
http://health.state.tn.us/nbs/index.htm



Having problems or need to sign up for the Voice Response System (VRS)? Contact Nancy at 615-262-6304



Metabolic DVD available free upon request.

Please call 615-262-6304

www.nltn.org/318-10.htm
www.nltn.org/317-10.htm

CEU's available

Hearing DVD available free upon request.

Please Call 615-262-6160

Importance of Newborn Screening
http://www.cdc.gov/Features/NewbornScreening

Important Newborn Screening Phone Numbers at a Glance

NBS Metabolic Follow-Up
Ph 615-262-6304
Fax 615-262-6458

Pamphlets
Ph 615-262-6304
Fax 615-262-6458

ICD9 Codes
Ph 615-262-6300

NBS Hearing Follow-up
Ph 615-262-6160
Fax 615-262-6159

NBS State Laboratory
Ph 615-262-6473
Fax 615-262-6447

Newsletter Email List
Ph 615-262-6304

Filter Papers
Ph 615-262-6391
Fax 615-262-6455

Billing
Ph 615-262-6300
Fax 615-262-6396