Volume 6, Issue 1 April 2011

State of Tennessee Department of Health

Newborn Screening Newsletter

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

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

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

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Special points of interest:

NBS UPDATES

- NBS Online Course designed for professional healthcare workers who participate in the collection or submission of newborn screening specimens. Free until 12-31-11 for TN healthcare professionals. CEUs available.
  PACE[R] Register Online @ www.nltn.org/371-10.htm
  TN Nurses CNE Register Online @ www.nltn.org/318-10.htm

INTRODUCTIONS

- Dr. Michael Warren, Director of Maternal Child Health
- Melissa Judd, RN Case Manager, NBS
- Lisa Smith, RN Case Manager, NBS

UPCOMING STATE HOLIDAYS

- April 22, 2011 Good Friday
- May 30, 2011 Memorial Day

Congenital Hypothyroidism

Congenital hypothyroidism is a condition where the thyroid gland does not make enough thyroid hormone. The thyroid gland is a small butterfly-shaped gland located in the neck. Normally, it uses iodine from food we eat to make thyroid hormone. This thyroid hormone is also known as thyroxine (T4). T4 is needed for normal growth and development. If congenital hypothyroidism is left untreated, it can lead to growth failure, mental retardation and other serious health problems.

About one baby in 3,000 is born with congenital hypothyroidism in the United States. The condition can be found in all ethnic groups.

The bad effects often do not appear until a baby is three to four months old. Some babies may have newborn jaundice, poor growth and weight gain, lengthy sleeping habits that require a baby to be awakened for feeding and severe constipation. If the baby is not treated, it can result in mental retardation, growth failure, deafness and other neurological problems.

The most common cause of congenital hypothyroidism is the incorrect growth of the thyroid gland. Early in fetal development, a baby's thyroid gland begins to form in the brain and then moves to the neck. In some cases this process is interrupted, resulting in a small thyroid gland (dysgenesis) or even the lack of a thyroid gland (agenesis). This is a permanent condition that a baby does **not** outgrow.

Congenital hypothyroidism does not appear to have a clear pattern of inheritance. There are some types of inherited hypothyroidism that are autosomal recessive. Most often, the parents of a child with an autosomal recessive condition are not affected because they are *"carriers,"* with one copy of an altered gene and one copy of the normal gene. Your endocrinologist and/or regional genetic center will let you know if your baby has this type of hypothyroidism and the chance of having another child with this condition.

Babies are tested (newborn screening) for congenital hypothyroidism before they leave the hospital. The baby's heel is pricked and a few drops of blood are taken. The blood is sent to the state laboratory to look at the thyroid-stimulating hormone (TSH, a precursor to thyroxine (T4)).

In most cases, the bad effects of congenital hypothyroidism can be prevented by replacing the missing T4 with medication given by mouth. The medication form of thyroxine is needed throughout an individual's lifetime. Follow up with an endocrinologist may be done to evaluate the amount of thyroid

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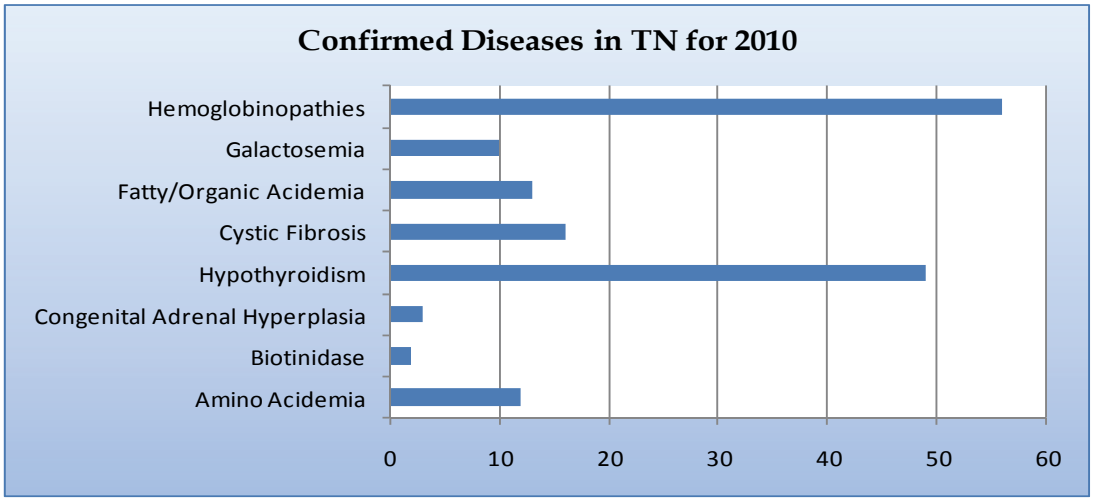

Argininosuccinic Aciduria Screening by Tandem Mass Spectrometry

The Tennessee Department of Health Laboratory Services began screening all infants born in Tennessee for Argininosuccinic Aciduria (also known as Argininosuccinate Lyase Deficiency) on May 15, 2010 by Tandem Mass Spectrometry using the primary analyte, Argininosuccinic Acid (ASA). This disorder is autosomal recessive and is caused by defects of the enzyme, Argininosuccinate Lyase. Argininosuccinic Aciduria is characterized as a urea cycle disorder.

The urea cycle only operates to eliminate excess nitrogen when protein is used by the body. On high protein diets amino acids are oxidized for energy or stored as fat and glycogen; however nitrogen from this process must be excreted. Through a series of reactions which occur in the liver cells, nitrogen is processed to urea a compound excreted by the kidneys. A deficiency of any one enzyme in the urea cycle can result in hyperammonemia.

In the urea cycle the enzyme Argininosuccinate Lyase cleaves argininosuccinate to arginine and fumarate. Deficiency in this enzyme blocks urea synthesis, leading to accumulation of amino acids, argininosuccinate and citrulline and prevents synthesis of arginine, making arginine an essential amino acid. Affected infants exhibit progressively deteriorating symptoms due to the elevated ammonia levels. Ammonia is very damaging to the central nervous system and can also eventually cause liver damage. Onset is within the first two to three days of life. Infants present with vomiting, lethargy, poorly controlled breathing, hypothermia, brain degeneration due to ammonia, enlarged liver, coma and death.

> The incidence of this disorder is 1:70,000 live births and has a high mortality rate.

Late onset may present at a few months to years of age with non-specific mental retardation, episodic seizures and enlarged liver. Hair abnormalities and or skin lesions are also physical findings in late onset due to arginine deficiency. These patients will often self select a diet low in protein. Hyperammonemia in late onset is usually not severe and accumulation occurs only during times of illness or stress. Symptoms are non-specific in the milder form.

Tandem Mass Spectrometry

Treatment usually consists of a diet low in protein, supplementation with arginine for completion of the urea cycle, drugs to scavenge ammonia and in some cases supplementation with carnitine if the patient has secondary deficiency. Liver transplant may improve the metabolic status of the patient and

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These are our All Stars!

They have <1% or <2% unsatisfactory specimens turned in for the year of 2010!. Thank you all for such wonderful work and support. We appreciate your quality work!

<1%

Centennial Medical Center	St Mary's Medical Ctr	Baptist Memorial Hosp Union City
Southern Tn Med Ctr-Columbia	Parkwest Medical Ctr	Northcrest Medical Ctr
Laughlin Memorial Hospital	Lincoln Medical Ctr	Middle TN Medical Ctr
Takoma Adventist Hospital	Maury Regional Hospital	Stone Crest Medical Ctr
Lakeway Regional Hospital	Women's Wellness & Maternity Ctr	Methodist Hospital-North
Fort Sanders Regional Med Ctr	Sweetwater Hospital Assoc.	Williamson Medical Ctr

<2%

Blount Memorial Hospital	Vanderbilt Client Service NICU	Mercy Med Ctr (Baptist) West	St Francis Hospital
Jellico Community Hosp	Vanderbilt Hosp Client Ser-Well-born	Crockett Hospital	Holston Valley Med Ctr
McKenzie Regional Hosp	Summit Med Ctr	McNairy Regional Hosp	Baptist Memorial Hosp For Women
Sycamore Shoals Hosp	Morristown-Hamblen Hosp	Blanchfield Army Comm Hosp	Sumner Regional Med Ctr
Baptist Hosp of Cocke Co	Erlanger East	Gateway Medical Ctr	Franklin Woods Comm Hosp
John Wharton Regional Med Ctr	Henry Co Med Ctr	Methodist Hosp-Germantown	Volunteer Comm Hosp
Metro-Nashville Gen Hosp	Crockett Hospital	Methodist Hospital-South	University Med Ctr
Baptist Hosp Inc			

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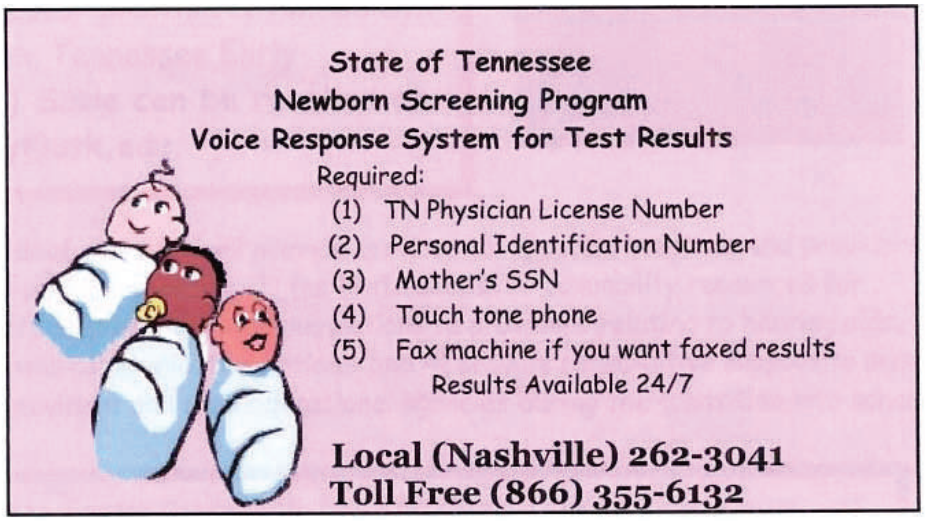
Voice Response System



VRS

Having problems or need to sign up?
Contact Nancy at
615-262-6304

Please remember that time spent giving normal results over the phone is time spent away from caring for sick babies.

2011 Tennessee General Assembly Legislative Bills Proposed

Legislation introduced by the 2011 Tennessee General Assembly includes:

- Senate Bill 65 / House Bill 373, an act to amend TCA, Title 68, relative to screening of newborns for critical cyanotic congenital heart disease.
- Senate Bill 616 / House Bill 1601, an act to amend TCA, Titles 68 and 71, relative to the care of premature infants. Its purpose is to improve health care quality and outcomes for infants born preterm through enhanced hospital discharge, follow-up care, and management processes and reduced hospitalization.

Congenital Hypothyroidism

(*Continued from page 1).*

gland present, determine its location, and watch how much thyroxine is being made.

Consult with your endocrinologist and/or regional genetic center to determine what tests and follow up are needed.

For more info go to: http://health.state.tn.us/nbs/PDFs/Congenital Hypothyroidism.pdf

Argininosuccinic Aciduria Screening…

(Continued from page 2).

offers a partial correction of the enzyme deficiency. Fasting must be avoided and especially during illness the patient must supplement with high carbohydrates and non-protein calories to avoid catabolism. Metabolic emergencies may require acute hemodialysis to lower blood ammonia levels.

The incidence of this disorder is 1:70,000 births and has a high mortality rate. Our normal cutoff for ASA is <0.18 µmol/L. We have also instituted a ratio of Argininosuccinic Acid to Arginine (ASA/ARG) that can be elevated in true cases of Argininosuccinic Aciduria. The normal cutoff for this ratio ASA/ARG is <0.13. So far three cases of Argininosuccinic Aciduria have been diagnosed.

Tennessee Department of Health

Newborn Hearing Screening

health.state.tn.us/NBS/hearing.htm

Newborn Hearing Screening – Jacque Cundall
615-262-6160 Jacque.Cundall@tn.gov

What Are The Hearing Screening Rates for Tennessee?

98% of infants received a hearing screening in 2009, 70% completed follow-up for a failed screen, 20% were lost to FU. 55 hospitals screened more than 95% of their infants.

What Type of Hearing Screening is Used in TN Hospitals?

ABR: 65.9%, OAE: 31.0%, Both ABR and OAE: 0.7%, Unknown: 2.5%

How Do I Identify Risk Factors for Hearing Loss

NEW HAND OUT: RISK INDICATORS Flip Chart to place by your hearing screening equipment.

Where Do I Send An Infant for Hearing Follow-up?

HANDOUT: Hearing Provider Sheet – An individual page of hearing providers (East, Middle, and West Tennessee Regions) is available to provide families in your hospital or practice.

How Can I Educate Parents About The Importance of Hearing Follow-up?

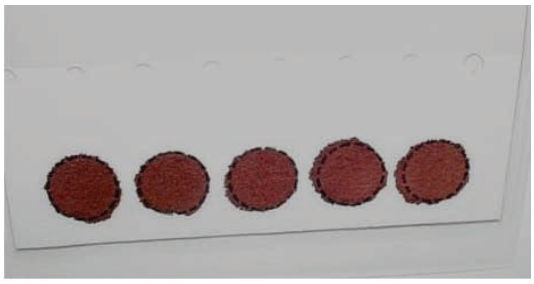
NEW DVD: A short DVD was recently distributed to the hearing screening contacts at hospitals. The DVD can be added to existing patient education loops utilized by the hospital.

NEWBORN SCREENING GALACTOSE ENZYME TESTING (GALT)

Laboratory Services, Newborn Screening has completed our validation for Galactose-1-phosphate uridyltransferase (GALT) testing.

This new assay was implemented on February 16, 2011. This assay is a multi-plex assay that allows us to test every sample for GALT and Total Galactose. This method is quantitative. The Total Galactose abnormal cutoff remains unchanged at ≥ 15 mg/dL. The GALT assay unit of measure is µmol/L, and the abnormal cutoff for the GALT assay is ≤ 40 µmol/L; as opposed to the categories of absent, low or normal GALT enzyme and numerical values for the GALT assay which were reported when the result was abnormal.

This test requires two 1/8 inch dried blood spots for analysis; therefore, it is critical that you submit good quality samples and that all five filter paper circles are filled in completely. This assures that adequate samples are available for all routine newborn screening tests and any required repeat testing. If you have any questions, please contact Christine McKeever, (615-262-6352 or Chris.McKeever@tn.gov) or Thomas Childs (615-262-6352 or Thomas.Childs@tn.gov).



Satisfactory Blood Spots for Specimen

Incomplete/Inaccurate Information

The Newborn Screening Program is receiving about 10 or more corrections a day. Corrections are sent when the original form was not filled out completely, filled out inaccurately, or illegibly before it was submitted. The most common mistakes or omissions are leaving off the date/time of birth or date/time of collection. It is also important to include the last date of transfusion when sending in specimens. Please assign someone in your organization the task of checking the information on the form with the information of the mother and infant as a last step before mailing.

Did you know missing or inaccurate information on newborn screening forms can:

Cost your hospital the same as any unsatisfactory newborn screening result when a repeat sample is collected?

Delay needed follow-up and/or diagnosis and treatment?

Be **prevented** by double checking the information on the form just before it is mailed?

Important Newborn Screening Phone Numbers at a Glance

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

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

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

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

ICD9 Codes
Ph 615-262-6300

State of Tennessee Department of Health
Newborn Screening and Genetics
630 Hart Lane
Nashville, TN 37243

Phone: 615-262-6304
Fax: 615-262-6458
E-mail: http://health.state.tn.us/NBS/index.htm

GENETICS ADVISORY COMMITTEE

The meeting is scheduled for May 5, 2011 from 10A-2P (central). It will be held at the State Laboratory Building at 630 Hart Lane in Nashville, TN. Please mark your calendars to attend!

Let's Do it Right the First Time

NOT ON OUR EMAIL LIST?

CONTACT LISA @ 615-262-6304