Volume 1, Issue 1 Winter 2012

Newborn Screening Newsletter

NEWBORN SCREENING: MUCH MORE THAN A PKU

SPECIAL POINTS OF INTEREST:

- Newborn Screening saves lives
- It's the law
- The kind folks at the Newborn Screening Program are here to facilitate a smooth screening process.
- Utah Screens for 37 genetic and metabolic disorders

YOU CAN'T STOP PROGRESS!

Welcome to the first quarterly Newborn Screening Newsletter! We are so excited to keep our community providers informed about the importance of and advancements in Newborn Screening! You, as the medical home, are crucial to the process of screening our newest residents!

Just to kick things off, I'd like to give a brief history of newborn screening in Utah. In 1965, PKU was the found to be the most common *preventable* cause of mental retardation in adults. An inability to metabolize phenylketonuria causes it to build up in the brain thereby causing brain damage. A simple screen at birth was able to detect the disorder before irreversible symptoms began. In 1979, the Utah Department of Health began to oversee the follow-up of the screening process. At that time, Utah screened for PKU, Galactosemia, and Hypothyroidism. In September of 2001, hemoglobinopathies were added. In January 2006, amino acid, fatty acid, and organic acid disorders were added as well as biotinidase deficiency and congential adrenal hyperplasia. In January 2009, Cystic Fibrosis was added to the panel.

Utah now screens for 37 disorders!

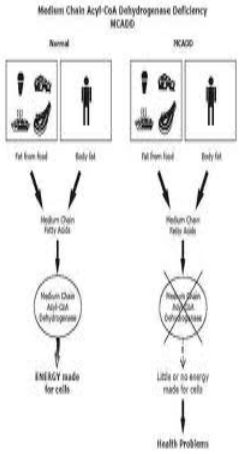
I am looking forward to hearing from each of you regarding what you would like to read about and how I can bring the Newborn Screening Program to the streets!

Terri Buchanan-Burdick RN BSN

IF A BABY IS DISCHARGED FROM THE NICU...

Babies who have had the pleasure of a NICU stay have most likely had both screens collected. NICU protocol is to collect the second newborn screen at 8 days of life. Baby gets discharged and our diligent community providers collect another specimen at the 2-week appointment. Now baby has three newborn screens. It is preferable to over screen rather than under screen however we will most likely call your office and request permission to destroy the unnecessary third sample. If you have a patient who has had an extended hospital stay, please feel free to call our office and ask about the status of the second screen.

MCADD: A fatty acid disorder



A mutated *ADADM* gene is responsible for providing instructions for making an enzyme called medium-chain acyl-Co A dehydrogenase, this enzyme breaks down medium chain fatty acids providing the heart and muscles with energy, and *in times of fasting*, provide energy to the liver and other tissues.

MCADD is an autosomal recessive fatty acid disorder. The early signs and symptoms are lethargy and hypoglycemia caused by the body's inability to break down fatty acids for fuel. The build up of unusable fatty acids in the liver, tissues and brain over time cause the more severe symptoms of brain and liver damage as well as possible cardiac arrest.

The good news is with early detection MCADD is relatively easy to manage. Baby may not fast any longer than the weight allows. No longer than 3 hours during the newborn period. As the child grows, time between meals can be extended but anytime the kid gets an ear infection or other common childhood malady , they must go directly to the ED for IV glucose.

MCADD has only been included in the newborn screening panel since 2006. Once identified via newborn screening we will call the pediatrician to have confirmatory labs collected. Familial compliance is critical as this disorder is absolutely life threatening.

With early detection , management is relatively easy

Meet your Newborn Screening Members

Kim Hart, MS LCGC

Kim is the Newborn Screening Program Manager and a Genetic Counselor. She has worked for the Department of Health for just over a year, replacing Fay Keune. Prior to joining the team she worked with the fine folks at ARUP. Kim is married and has two children, who turned three in November. She enjoys running, swimming, camping, and herding her twins.

Tanayia Pomikala, BS

Tanayia has been the brain of our operation for three years. She is the Health Program Representative and if she doesn't have an answer to your question, she knows who will. Prior to Newborn Screening, Tanayia worked in Medicaid.

Tanayia is from Gary, IN, just like Michael Jackson. She is married, has two children, and three step-children. She is a mean bunko player and is always smiling.

Tanayia's direct line is 801.584.8256, call her and let her make your day brighter.

Meet your newborn screening program members, continued

Karen Roylance, RN BSN

Karen has worked for the Newborn Screening Program for 5 years. She has worked for the great state of Utah for 10 years and has spent 16 years in public health. Karen is the Cystic Fibrosis expert in our office. She follows up on abnormal screens and schedules sweat tests as necessary. She is a great resource for all your CF needs.

Karen is available for newborn screening in-services in your office. Call and set something up. Her direct line is 801.584.8260

Karen has 3 children and 4 grandchildren. She loves to travel and ride her motorcycle through the beautiful, local, canyon roads.

Terri Buchanan-Burdick, RN BSN

Terri started working with the Newborn Screening Program in June, when Jan Kilpack retired. Previously, Terri worked in the NICU at the University of Utah Hospital for almost five years. She is a graduate of Wesminster College.

If you have any questions about hemoglobinopathies or congenital hypothyroidism she is happy to answer them or direct you to a helpful resource. If you wonder about the correct protocol for a baby who has received a transfusion, give her a call.

Terri is also available for newborn screening in-services in your office. Call and set something up. Her direct line is 801.584.8530

Terri likes to camp, raft, run, climb rocks, and spend time with her kids.

Nicole Tattersal, RN BSN

Nicole is the newest member of our team. She started with Newborn Screening in November. Nicole has nursing experience in research, and mental health. She got her nursing degree from the University of Utah.

Nicole has two dogs, Ozzy and Kaija. She has season Jazz tickets and spends a lot of time with her niece and nephew. Nicole is also an ordained minister, she is able to marry you and bury you as well as answer questions about metabolic disorders.

Nicole's direct line is 801.584-8279