Shodair CHILDREN'S HOSPITAL

GeneMaP
Genetics for Montana Providers

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Children's Miracle Network Hospitals Helping Local Kids

Shodair Children's Hospital is Montana's only Children's Miracle Network affiliated hospital. Children's Miracle Network is a non-profit organization dedicated to saving and improving the lives of children by raising funds for premier children's hospitals across North America. For more information, visit: www.cmn.org

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Please direct your ideas or comments regarding this newsletter to: **mtgene@shodair.org**

Maternal Serum Screening: Which test to offer your patients?

By Amy Crunk, MS, CGC

Maternal serum screening is the standard of care for pregnant women. Since the introduction of alpha-fetoprotein (AFP) to screen for Down syndrome (DS) in 1984, maternal serum screening has grown to include first trimester screening, second trimester screening, and sequential and integrated screening. How is one to know which screen is best for their patients?

To help physicians navigate through all these choices, the American College of Obstetricians and Gynecologists released a practice bulletin in 2007. The table below lays out the different screening options and their detection rates for DS.

Screen	DS detection rate
Triple Screen AFP, hCG, estriol	**70%**
Quad Screen AFP, hCG, estriol, inhibin A	**80%**
Serum Integrated Screen PAPP-A + Quad Screen	**85%**
First Trimester Screen NT, hCG, PAPP-A	**87%**
Sequential Screen First Trimester screen + Quad Screen Results at both steps	**90%**
Integrated Screen First Trimester screen + Quad Screen Results after Quad Screen	**95%**

AFP= alpha-fetoprotein, hCG= human chorionic gonadotropin, NT= nuchal translucency, PAPP-A= pregnancy-associated plasma protein A

A limitation of the first trimester screen is the need for specific training and ongoing audits to perform nuchal translucency (NT) measurements. This makes the availability of first trimester screening, and hence the sequential and integrated screen, limited in Montana.

ACOG recommends first trimester screening with NT measurement and biochemical markers. When NT measurements are not available, ACOG recommends serum integrated screening. For patients that present for the first time in the second trimester, then the quad screen is recommended by ACOG. The serum integrated screen, as well as the quad and triple screen, are available through Shodair Children's Hospital. We are also available to speak with you and your patients if there is an abnormal result. If you or your patients have additional questions about maternal screening options, call us at 1-800-447-6614. Or, you can refer to the ACOG Practice Bulletin on Screening for Fetal Chromosomal Abnormalities in Obstetrics & Gynecology, Vol, 109, No. 1, January 2007.

Shodair Offers Genetic Outreach Clinics

Did you know that we see most of our clinical genetic patients not in Helena, but in outreach clinics through out the state? Each month, we have clinics in Billings, Bozeman, Great Falls, and Missoula, as well as in Helena. We also see patients in Butte, Kalispell, Browning, Miles City and Sidney. Here's a list of our upcoming clinics for early 2010:

Billings
Jan. 13/14
Feb. 17/18
Feb. 19 ***Cleft***
Mar. 17/18

Bozeman
Jan. 21
Feb. 25
Mar. 25

Browning
Mar. 26 ***Cleft***

Butte
Jan. 19
Mar. 23

Helena
Jan. 12
Jan. 26
Feb. 23
Mar. 5 ***Cleft***
Mar. 16

Great Falls
Jan.6
Feb. 10
Mar. 10

Missoula
Jan. 5
Feb. 3
Feb. 4 ***Cleft***
March 2/3

To make a genetics referral, simply fax patient information to our Clinic Coordinator, Barb Doggett, at (406) 444-1064 or call her at (406) 444-7530. Please note that clinic dates and locations are subject to change.

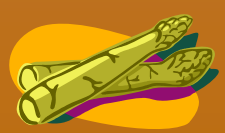
Loeys-Dietz Syndrome

By Amy Crunk, MS, CGC

Four-year-old Noah was originally referred to us for a diagnosis of autism. But in between the referral and our visit, Noah had an echocardiogram because of a murmur; the focus of our visit changed substantially. Noah was found to have a dilated aortic root. Noah's father had passed away in the last year from an aneurysm. We now were spending most of our visit discussing familial aortic aneurysms and possible genetic testing.

Since Noah and his family did not fit the clinical criteria for Marfan's syndrome, we decided to do gene testing for a new familial aneurysm syndrome: Loeys-Dietz Syndrome (LDS). LDS was first described by Drs. Loeys and Dietz in 2005. LDS is a connective tissue disorder similar to Marfan's syndrome. Both have aortic dilation and aneurysm, pectus excavatum or pectus carinatum, and flexible joints. LDS is different in that there is arterial tortuosity and aneurysms can occur in arteries other than the aorta. Individuals can also have characteristic facial features such as hypertelorism.

Noah did have a change in the TGFBR2 gene known to cause LDS. He was also found to have arterial tortuosity. Noah's mom has been a great advocate for Noah. She contacted Dr. Dietz's office about the diagnosis to get as much information as she could. Noah is being followed for his aortic dilation and being checked for other complications that may arise. We will be following up with Noah and his mom to further discuss his LDS diagnosis as well as discuss his autism diagnosis, which is not related to LDS.



Genetic Fun Fact

Have you ever had dinner, gone to the bathroom and wondered why your urine smells funny? Then you remembered that you ate asparagus. Being able to produce the odor and being able to smell the odor after eating asparagus are genetic traits. It seems that some people are able to produce the odor, some not, and some are able to smell the odor, and some not. Something to think about the next time you have asparagus!