



Gene MaP

Genetics for Montana Providers

Volume 1 Issue 2

July 2, 2009

Microarray Studies Increase Diagnostic Success

When a patient's chromosomes are normal, do you ever wonder what might be the cause of physical anomalies or learning problems? Microarray may supply the answer. Over the last several years, our genetics team has ordered over 130 microarray tests on individuals of all ages. This technology involves comparing the fluorescent labeling of DNA of a patient to that of a control, allowing identification of extra or missing DNA segments.

Approximately 10% of the studies we ordered showed a true abnormality. At times, microarray has allowed us to identify an "unknown marker" or unidentifiable chromosome segment attached to another chromosome that was first spotted on traditional chromosome studies. Tiny microarray abnormalities have also been found to be the cause of characteristic features and learning problems in a patient and other family members.

Additionally, this technology has the capability to look for many microduplication and microdeletion syndromes (think Williams Syndrome, Velocardiofacial Syndrome) simultaneously. Limitations are that some results can have unclear clinical implications, and a normal result will not rule out all genetic conditions (think single gene conditions), usually best diagnosed by clinical exam with possible molecular testing.

BT's story: This happy, attractive girl is the first child born to her healthy young parents, who have backgrounds in horse training and gymnastics. We first saw BT at 8 months, when she was referred by her pediatrician for short stature and unidentifiable material on chromosome 13. She had some features reminiscent of Down syndrome: a small head, small midface, lacy iris pattern, and increased space between toes 1 and 2. Microarray testing determined that BT *did not* have a variant of Down syndrome, but *did* have partial trisomy and tetrasomy of parts of chromosome 21 *outside* of the Down syndrome critical region.

This finding provided an explanation for BT's differences in development, guiding her medical care and helping

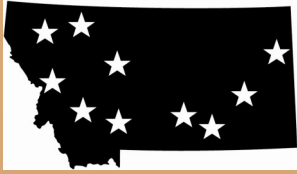
Do you sneeze when you step into sunlight after being in a dark room? You may have ACHOO Syndrome, characterized by nearly uncontrollable sneezing provoked by sudden exposure to intensely bright light (usually sunlight) for a dark-adapted subject. The number of sneezes could be two or three, or as many as 40. ACHOO is an acronym for Autosomal Dominant Compelling Hellophthalmic Outburst and



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

This project is funded (in part) under a contract with the Montana Department of Public Health and Human Services. The statements herein do not necessarily reflect the opinion of the Department.

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



Shodair Offers Genetic Outreach Clinics

Most of you know that Shodair is located in Helena. Our genetics laboratory is here, and so are our clinical genetics offices. But did you know that we see most of our clinical genetic patients not in Helena but in outreach clinics through out the state? Every 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 Sydney.

The next time you have a genetics referral, but don't think the patient will travel to Helena, don't worry! We'll work with your patients to find the outreach clinic that works best for them. Here's a list of our upcoming clinics:

<u>Billings</u>	<u>Helena</u>	<u>Kalispell</u>
July 22/23	July 14	July 8/9
August 19/20	August 6	October 6/7
September 18	August 24	
October 14/15	September 8	<u>Miles City</u>
November 18/19	September 23	September 15
December 16/17	October 1	September 17
	October 13	
<u>Browning</u>	November 24	<u>Missoula</u>
October 8	December 1	July 7
	December 15	August 5
<u>Bozeman</u>		September 1/2

Shodair Plans First-Ever Montana Metabolic Day

Do you have patients who have been diagnosed with PKU or another metabolic condition? Shodair's Metabolic Team is planning an event that will allow these patients an opportunity to discuss the importance of managing metabolic disorder, meet other people with special metabolic needs and their families, and ask questions of metabolic healthcare professionals:

Montana Metabolic Day

Saturday, August 15, 2009

9:00 am – 3:00 pm

Carroll College (Trinity Hall Lounge) in Helena

