



# Gene MaP

## Genetics for Montana Providers

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*Please direct your ideas or comments regarding this newsletter to: [mtgene@shodair.org](mailto:mtgene@shodair.org)*

### Pharmacogenetic Testing: How It Can Help Your Patients

By Amy Crunk, MS, CGC

We all know that patients react differently to medications. We generally use trial and error to fit the right medication and dosage to a patient. Pharmacogenetic testing allows us to fit the right medication and dosage to the patient based on the patient's genetic make-up.

Many drugs, including SSRI antidepressants, are metabolized by CYP450 enzymes. The Genetics Laboratory at Shodair Children's Hospital looks at the genotypes of three CYP450 enzymes — CYP2D6, CYP2C19, and CYP2C9 — to determine how a patient will metabolize drugs. Extensive metabolizers are considered "the norm" or the average in respect to the speed they metabolize a drug. Intermediate metabolizers have slower enzyme function and may need a lower dose of a medication to have the appropriate response. If conversion to an active metabolite is required, then a higher dose may be necessary. Poor metabolizers have little or no enzyme function, causing a drug to build up in the body to toxic levels, which has led to death for some patients. Ultra extensive metabolizers have fast enzyme function and may need a larger dose of a drug in order for the drug to have an effect, or a smaller dose if the metabolite is active.

Pharmacogenetic testing is available for patients who have had an adverse drug reaction, drug sensitivity, or may not be experiencing therapeutic benefit from medications. Individuals who have a family history of drug intolerance are also good candidates. For some patients, pharmacogenetic testing will allow quicker therapeutic benefit without first going through trial and error.

For more information on pharmacogenetic testing, please contact our Genetic Laboratory at (406) 444-7532 or visit our website at [shodair.org](http://shodair.org). Click on Genetic Services, then click on Laboratory Services in the menu bar on the left.

### Shodair Launches New Website

Shodair Children's Hospital recently completed a website makeover, making it easier for physicians and patients to access more information about our programs and services. Referring patients is even easier now — just visit [shodair.org](http://shodair.org) and click on **Genetic Services** for instructions. For lab services, you can now download our Genetic Laboratory requisition forms. If you'd like to bookmark our new website, simply:

1. Start Internet Explorer
2. Visit our page at [shodair.org](http://shodair.org)
3. Click on the "Favorites" menu
4. Select "Add to Favorites..."
5. Click "Add"



## 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.

To make a genetics referral, visit our website at [shodair.org](http://shodair.org), or fax patient information to our Clinic Coordinator, Barb Doggett, at (406) 444-1064. You may also call Barb at (406) 444-7530. Please note that clinic dates and locations are subject to change.

### MCAD Deficiency: When Newborn Screening Relies on Diagnostic Testing By Tessa Pitman, MS

Medium Chain Acyl-CoA Dehydrogenase deficiency, or MCADD, is a metabolic disorder that is screened for on the expanded newborn screening panel. It is the single most common fatty acid oxidation disorder, more common than PKU, with an incidence of about 1 in 10,000 individuals (about 1 in 50 individuals are carriers of this recessive condition). Typically, affected individuals are asymptomatic at birth; however, they can present clinically anytime in the first few years of life with hypoglycemia, vomiting, and lethargy secondary to illness. Without intervention, this can quickly progress to coma and death. The prognosis is excellent once the diagnosis is made and treatment involving avoidance of fasting, vigilance of illness (that induces fasting), and possibly carnitine supplementation can be started.

As part of the Newborn Screening Follow-Up Program, the genetics team at Shodair assisted in diagnosis of two cases of MCADD in 2009. One case involved an asymptomatic, term, female with a normal birth weight. The initial newborn screen was abnormal. The acylcarnitine profile on the newborn screen distinctively fit the pattern for MCADD. Biochemical diagnostic tests were recommended. Interim recommendations were provided that included the avoidance of fasting and vigilance of illness with or without fever.

Instead of diagnostic tests, a repeat newborn screen was obtained. The repeat screen was normal. The question soon became, "Is this really MCADD, given that the second screen was normal?"

The answer is yes. For normal term infants, the median concentration of individual acylcarnitines is highest in the first two to three days of life and declines until concentrations stabilize at day seven or greater. Therefore, for fatty acid oxidation disorders the first screen is more telling than the second screen. Often times, the second screen is normal, providing a false sense of security that the child is really unaffected. This is why for MCADD we recommend diagnostic tests rather than a repeat screen. Diagnostic studies were eventually obtained and the diagnosis was confirmed. Molecular testing was also performed, which also confirmed the diagnosis.

### Genetic Fun Fact

We all learned in high school that the ability to roll one's tongue is an autosomal dominant trait. If you have one tongue rolling gene, you can roll your tongue. Twin studies on this trait provide a different answer. Identical twins, who share all their genes, should always share the tongue rolling trait, but they don't. It seems that other unknown factors play a "roll" in tongue rolling!

**We hope you enjoy this quarterly newsletter; please feel free to forward it your colleagues. To request additions/removals from our email list, contact us at [mtgene@shodair.org](mailto:mtgene@shodair.org).**