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Genetics for Montana Providers

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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

Shodair Welcomes New Geneticist: Samuel P. Yang, M.D., FAAP

Medical geneticist Samuel Yang, M.D. joined Shodair's Genetics Department last month.

A graduate of Johns Hopkins School of Medicine, Dr. Yang completed an internship in pediatrics at Stanford Medical Center, followed by a residency in pediatrics at UC Davis Medical Center. Dr. Yang also completed a Fellowship in Genetics at Children's Hospital of Central California.

Dr. Yang is board certified by the American Board of Pediatrics, and the American Board of Medical Genetics with both clinical and clinical biochemical specialties.



Most recently, Dr. Yang worked at Lanterman Development Center in Pomona, California. Prior to that, he was associated with the Permanente Medical Group in San Francisco, Children's Hospital of Los Angeles, and the California Department of Developmental Services.

We're excited to welcome Dr. Yang to our team! While Dr. Yang will see patients with a variety of genetic concerns along with existing Shodair geneticists Dr. John Johnson and Dr. Jim Reynolds, he will specialize in treating patients with metabolic conditions through the Newborn Screening Follow Up program operated by the Montana Department of Public Health and Human Services and Shodair Children's Hospital.

To refer a patient to Dr. Yang, call (406) 444-7530 or 1-800-447-6614.

Genetic Testing and Hereditary Colorectal Cancers By Katherine Berry, MS, CGC

Imagine living for years knowing that numerous family members had colon cancer, and some survived with the help of surgeries, while others, less fortunate, did not. Imagine at age 40 that you went for a colonoscopy, but felt so anxious that you left the medical center before it was done. After that, you had no coverage to pay for the procedure. Now, at 47, you know that your sister carries an APC gene mutation causing FAP (familial adenomatous polyposis), and your risk of carrying the gene approaches 50%. Such is the story of a woman we recently saw for genetic counseling and gene testing.

While most colorectal cancers are sporadic, about 10% are hereditary. HNPCC, hereditary nonpolyposis colon cancer (Lynch syndrome) accounts for 3-5%, while FAP accounts for up to 1%. Identifying a patient who may be carrying one of these mutations may be valuable in determining who is at increased risk for future cancers and may benefit from

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increased surveillance, preventative surgery or medications---and who isn't at risk and doesn't need them. Implications for children and other relatives are another compelling reason to consider families for counseling and testing.

HNPCC can cause early onset colorectal cancer (often but not always before 50), as well as endometrial, stomach, ovarian, kidney/urinary, brain, small bowel, pancreatic, and other cancers. Any patient with one or more of these, or a combined personal and family history that show a combination of these cancers, may benefit from testing of the several genes which can cause this condition. Testing of tumor cells to determine whether they show microsatellite instability (MSI) or immunohistochemistry (IHC) findings typical of tumors caused by this condition may indicate whether HNPCC is more or less likely, depending on the result. Recent studies have shown MSI and IHC to be useful in evaluating tumors of older individuals, since some of them carry these mutations as well.

FAP causes many polyps in the colon and increases the risk for cancers of the duodenum, pancreas, thyroid, stomach, brain (and liver, in children). This condition carries a high cancer risk for young people if unrecognized and untreated by preventative surgery. However, with gene testing, screening, and colon surgery when appropriate, the most serious manifestations of this condition may be avoided.

We had the privilege of telling our patient above that she was free of the family mutation and its conferred cancer risks. She was thrilled with the implications for her future health and that of her children.

Genetics clinic cancer consultations are available locally on a timely basis to assess individuals with a personal and/or family history of cancer in order to estimate genetic cancer risk and discuss genetic testing options. Services are provided by a team of a medical geneticist and a genetic counselor. The process includes a discussion of insurance coverage. Insurance preauthorization for visits and testing is available. Individuals without insurance may qualify for waived or reduced charges, both for the visit and for testing.

To request a schedule of our outreach clinics, or to make referrals, please contact the Department of Medical Genetics at Shodair Children's Hospital at (406) 444-7530 or toll-free at 1-800-447-6614. You may also visit our website at www.shodair.org or email us at mtgene@shodair.org for more information.

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, 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.

Genetic Fun Fact

When you clasp your hands with the fingers entwining, is the right thumb closest to your body or the left? It is thought that which thumb is closest to your body has a genetic component. One study found that 55% of the population are left-thumb closest, 44% are right-thumb closest, and the remaining 1% have no preference. What's yours?

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.

