Patient **Impact**

The statewide focus of our clinical and laboratory services provides a patient care model not possible in large hospital settings. We get to know all of our patients as individuals, and our clinical team works closely with our laboratory to ensure all of the necessary information is available to come to an accurate diagnosis whenever possible. The impact this has on patients and their families is incredible to see. Here are just two examples of the kinds of patient stories we see every day.

"You can see how much more alive she is."
MOTHER OF ZEESY • SHODAIR GENETICS PATIENT

A four-year-old girl (above) came to our clinic with a history of seizures and delays in development. As an adopted child, her family history wasn't clear, which can sometimes make diagnosing genetic conditions challenging. Through consultation between neurologists and our providers, testing for genes associated with seizure disorders was performed and a diagnosis of GLUT1 deficiency was made. This ultra-rare disorder doesn't allow glucose, the body's normal fuel, to reach the brain efficiently, starving the brain of energy. Fortunately, with this knowledge, the child was started on a ketogenic diet, changing her brain's energy source and allowing her to grow and thrive.

The **Future**

Medical genetics is a rapidly growing and evolving field. Technological advances have reduced the time and expense to find a diagnosis to weeks rather than years or even decades. Patients that we could not diagnose before now have answers and medical management that greatly improves their care and quality of life. Genetic disease touches every aspect of medicine, including oncology, cardiology, and even mental health care.

As Montana's leading resource for genetic medicine, Shodair Genetics needs to continue to be at the forefront of technology and patient access. One of our current growth initiatives is a pharmacogenetics program offering genetic testing to help choose the best medications to use to treat mental health disorders, cancer, and other diseases.

Infants born with rare genetic disease can have better outcomes if genetic care is integrated earlier. To meet this need, we are collaborating with all major hospitals in the state to make rapid whole-genome sequencing available to critically ill infants in NICUs, potentially providing lifesaving diagnoses within the first week of a baby's life. Our clinical team is growing, allowing us to see more patients with less waiting time. And all of this will take place in a new outpatient building on our campus in Helena.

The work that we do is only possible through the support of the Montana community. If you would like to support this exciting and necessary work, please contact our Foundation.

To donate to Shodair's future, contact us at 406.444.7560 or scan the QR code.

SHODAIR.ORG | 406.444.7500
2755 Colonial Drive, P.O. Box 5539, Helena, MT 59604

Shodair Children's Hospital

Medical Genetics

We help guide their future.

To heal, help, and inspire hope

The **Challenge**

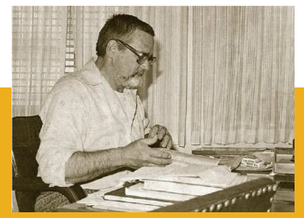
Genetic Medicine has moved to the forefront of health care throughout the world. This is in part because advances in our understanding of disease have revealed that many diseases, especially those impacting life throughout the lifespan have a genetic cause. A genetic diagnosis is increasingly becoming key to patient care, in some cases with life-altering or lifesaving effects. Genetic disorders occur in about 5% of all live births, make up 30% of pediatric hospital admissions and cause 50% of childhood deaths in the United States. Most genetic centers are based in large academic institutions and providing comprehensive genetic care close to home in a large rural state like Montana poses an extraordinary challenge.

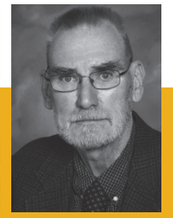
The **Response**

The Genetics Department at Shodair Children's Hospital is the only comprehensive rural clinical and laboratory genetics program of its kind in the United States, allowing patients of all ages and their families to receive state-of-the-art care close to home. Without our services, families struggling with genetic conditions would regularly have to travel long distances to academic centers in Washington, Utah, or Colorado.

OUR MISSION:

"To heal, help, and inspire hope."



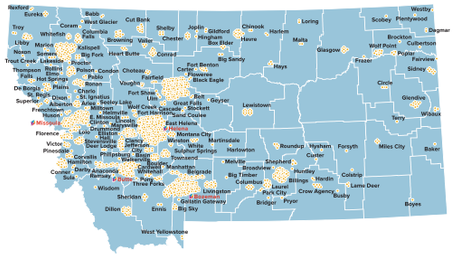


Rich **History**

The Department of Medical Genetics has been part of Shodair since 1976. Founded and formed by Phil Pallister (left) and John Opitz (right), two giants of modern human genetics, Shodair Genetics is nationally known in the genetics community as a model of rural outreach and integrated genetics care.

Regional **Impact**

Our clinical teams serve patients throughout Montana and the surrounding region, providing comprehensive support to families and providers. We also offer telemedicine direct to the patient's home when an in-person visit is not necessary or possible. Consultation services are offered to providers seeking assistance for patients with suspected genetic conditions, and our genetic counselors are contracted to support hospitals around the state.



Care teams travel to Missoula, Kalispell, Billings, and Great Falls each month, in addition to clinics held at our home hospital in Helena.

National **Recognition**

Shodair's Clinical Genetics Laboratory offers in-house, high standard-of-care genetic testing for the vast majority of genetic conditions. This includes cytogenetic testing for rapid diagnosis of Down Syndrome and other chromosomal conditions, single-gene testing for common conditions like Huntington disease, and genome-wide analysis by microarray and next-generation sequencing. Shodair's laboratory is known national for our expertise in epigenetic syndromes such as Beckwith-Wiedemann syndrome, receiving test requests from large hospital systems such as Kaiser Permanente and John Hopkins.

State-of-the-Art **Technology**

Although small, our laboratory houses the same advanced instrumentation that is found in large national reference genetics laboratories. Our sequencing and microarray platforms are capable of analyzing all 20,000 human genes at once, reducing the time and cost for patients to receive a diagnosis of a genetic syndrome. Soon we will have the capacity to perform rapid whole-genome sequencing, an emerging care model to guide treatment for critically ill infants and children with complex medical conditions.

Patient **Impact**

A 15-year old girl was evaluated for physical features concerning for Marfan syndrome, a disorder resulting in patients being tall and thin but with significantly increased risk for catastrophic cardiovascular complications. After evaluation by our care team, testing was performed not only for Marfan syndrome, but similar disorders as well. This testing revealed that the patient had a rare form of Loeys-Dietz syndrome, a disorder similar to Marfan syndrome but with less consistent physical features. This diagnosis not only put the patient on track for monitoring and medication to prevent the potentially lethal effects of this disorder but showed that her mother and grandfather also carried this genetic change. Because neither her mother nor grandfather had the same physical features that might indicate Loeys-Dietz syndrome, a diagnosis would have been easy to miss until a dangerous cardiovascular event occurred. Armed with this knowledge, all three family members are now monitored and cared for appropriately, with potentially life-saving effect.