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ChildrensNational.org

LeukoSEQ: Novel Leukodystrophy Genetics at Children's National

Announcing Next Generation CLIA certified Whole Genome Sequencing Study in Leukodystrophy

Dear Clinicians and Investigators,

We are very excited to announce the initiation of a whole genome sequencing study sponsored by the Myelin Disorders Bioregistry Project at Children's National Health System, for patients with recently identified and unsolved pediatric leukodystrophies.

The purpose of this study is to **investigate the diagnostic efficacy, clinical utility, and cost effectiveness of next-generation sequencing (NGS) for leukodystrophies by determining if NGS improves diagnostic effectiveness in the leukodystrophies relative to current diagnostic approaches and whether NGS results in changes to healthcare-related costs.**

Our hope is that the results of this LeukoSeq study will help to refine our understanding of leukodystrophies, develop new benchmarks for early diagnosis and clinical management of these conditions, and ultimately alleviate the burden of care shared by families and clinicians.

To help make this study a success, we are seeking your participation through the referral of qualified patients. Patients who participate in the study will be involved for approximately 16 months. Requirements include a blood sample donation, clinical records, and MRI studies followed by participation in health outcomes monitoring. All patients will receive results of CLIA-certified Whole Genome Sequencing. If patients do not meet the qualifying criteria for the LeukoSeq study, they can still be referred into the Bioregistry.

Who can participate in this research study?

- Patients with abnormalities of the white matter signal on neuroimaging (MRI) with T2 hyperintensity, which must be diffuse or involve specific anatomical tracts consistent with a genetic diagnosis
- White matter abnormalities identified less than two months prior to enrollment
- No evidence of an acquired cause for the white matter abnormalities (infection, trauma, birth related injury)
- No pre-existing diagnosis
- Less than 18 years of age at time of enrollment
- Availability of both biologic parents for blood sampling
- Willingness to adhere to study protocols and follow-up for 16 months

What does participation include?

- Providing MRI and clinical data
- Providing a blood sample for the patient and both parents
- Willingness to participate in biweekly follow-up on changes in medical care in the 12 months after test results are made available
- Availability of a Genetic Counselor on staff or available to assist in the review of genetic testing and results

How will you benefit?

- Your patients will receive results from CLIA-certified Whole Genome Testing

About the Myelin Disorders Program

The Myelin Disorders Program at Children's National Health System is a multidisciplinary clinical and bench research program and international leader in the study of inherited disorders of cerebral white matter, or leukodystrophies. Children's National hosts two of the largest clinical programs in pediatric neurology and genetics that researches, diagnoses, and treats leukodystrophies. We provide integrated care by a team of neurologists, pediatricians, rehabilitation physicians, and genetic counselors, ensuring that children receive the best care and the most appropriate treatment.

The program lead, Adeline Vanderver, MD, PhD, Neurologist and Director, Myelin Disorders Program has been instrumental in the creation of the Global Leukodystrophy Initiative (GLIA), a consortium of researchers and clinicians devoted to the discovery of novel variations responsible for pediatric white matter disorders and to translate these findings into clinical treatments. Under the leadership of Dr. Vanderver, Children's National serves as a global resource for information, research, advocacy, and treatment for families searching for information and care for their child.

For more information about the Myelin Disorders Program and our Bioregistry project, please contact Adeline Vanderver, MD, (PI) at avanderv@childrensnational.org or myelindisorders@childrensnational.org.

Regards,

Roger Packer, MD
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