Megalencephalic Leukoencephalopathy with Subcortical Cysts Type 1

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of megalencephalic leukoencephalopathy with subcortical cysts type 1. Carriers are not expected to show symptoms. You and your partner or donor would both have to be carriers of megalencephalic leukoencephalopathy for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning.

Recommended Next Steps

Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner or donor are carriers for megalencephalic leukoencephalopathy with subcortical cysts type 1, each of your children has a 1 in 4 (25%) chance to have the condition.

Megalencephalic Leukoencephalopathy with Subcortical Cysts Type 1 Explained

What is Megalencephalic Leukoencephalopathy with Subcortical Cysts Type 1?

Megalencephalic leukoencephalopathy with subcortical cysts type 1 is an inherited brain disorder that causes progressive neurological complications. Affected individuals are born with an enlarged brain (megalencephaly) and abnormalities in the brain's white matter (myelin, which insulates and protects nerve fibers). Additionally, they may develop cysts in the brain that can grow in size and number. They may have mild developmental delays early in life, but over time, the white matter wastes away, causing increased muscle spasticity, difficulty coordinating movements, and mild-to-moderate intellectual disability. Some affected individuals also experience seizures, poor muscle tone, and difficulties talking and swallowing.

Prognosis

Prognosis is unfavorable. Affected individuals typically lose the ability to walk and are wheelchair bound by adolescence. Lifespan varies but is typically reduced.

Treatment

There is no cure for megalencephalic leukoencephalopathy and treatment is symptomatic. Minor head trauma can lead to seizures, temporary motor deterioration, or coma. Due to this risk, affected individuals should wear a helmet when there is risk for head trauma.

Resources

Genetic and Rare Diseases Information Center
https://rarediseases.info.nih.gov/diseases/3445/megalencephalic-leukoencephalopathy-with-subcortical-cysts
Genetics Home Reference
https://ghr.nlm.nih.gov/condition/megalencephalic-leukoencephalopathy-with-subcortical-cysts
National Society of Genetic Counselors
https://www.nsgc.org/

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