Genetics Uncoded:

Facts about

Megalencephalic Leukoencephalopathy with Subcortical Cysts

What Your Test Results Mean

Carriers typically show no symptoms of megalencephalic leukoencephalopathy with subcortical cysts (MLC); however, carriers are at an increased risk of having a child with MLC. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Megalencephalic Leukoencephalopathy with Subcortical Cysts Explained

MLC is an inherited brain disorder that causes progressive neurological complications. Affected individuals are born with enlarged heads (megalencephaly) and abnormalities in the brain's white matter (myelin, which insulates and protects nerve fibers). 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. Treatment is supportive and most individuals with MLC live well into adulthood, though they may lose their ability to walk independently.

How the Genetics Work

The majority of MLC is caused by pathogenic variants in the *MLC1* gene. In general, individuals have two copies of the *MLC1* gene. Carriers of MLC have a single variant in one copy of the *MLC1* gene while individuals with MLC have variants in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with MLC is 25%.

Questions? Contact us at **1-855-776-9436** to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.

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