Genetics Uncoded:

NxGen MDx

Facts about Salla Disease

What Your Test Results Mean

Carriers typically show no symptoms of Salla disease; however, carriers are at an increased risk of having a child with Salla disease. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Salla Disease Explained

Salla disease is an inherited condition that affects the cell's ability to export free sialic acid from the lysosomes, causing severe impairment of the nervous system. Infants with Salla disease usually have poor muscle tone and progressive neurological problems during the first year of life. Signs and symptoms of Salla disease include intellectual disability and developmental delay, seizures, problems with movement and balance, and muscle spasticity. Motor and mental skills gradually decline in these individuals, and though most adults with the disease live normal lifespans, they are profoundly disabled. In more rare and severe cases, the disease progresses more quickly and may lead to decreased lifespan.

How the Genetics Work

Salla disease is an autosomal recessive disorder caused by variants in the *SLC17A5* gene. In general, individuals have two copies of the *SLC17A5* gene. Carriers of Salla disease have a single variant in one copy of the *SLC17A5* gene while individuals with Salla disease have variants in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with the disorder 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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