Genetics Uncoded:

Facts about Sjögren-Larsson Syndrome

What Your Test Results Mean

Carriers typically show no symptoms of Sjögren-Larsson syndrome; however, carriers are at an increased risk of having the disorder. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Sjögren-Larsson Syndrome Explained

Sjögren-Larsson syndrome in an inherited condition characterized by scaly and dry skin, neurological symptoms, and eye problems. Individuals with the disorder are unable to break down the fatty aldehydes, due to a lack of fatty aldehyde dehydrogenase. This leads to a build-up of fats causing the symptoms of the disease. Affected infants may be born prematurely. Leukoencephalopathy, a change in white matter of the brain, is frequent, causing most of the neurological symptoms that can characterize this disorder. Most affected individuals have intellectual disabilities evident at an early age. Dysarthria (speech difficulties) and delayed speech is common. Delayed motor skill functions, such as crawling or walking, is typical. There is no treatment for Sjögren-Larsson syndrome, and management is focused on symptoms and may involve physical therapy and neurological consultation. Individuals with this syndrome typically live into adulthood; however, lifespan is somewhat shortened.

How the Genetics Work

Sjögren-Larsson syndrome is an autosomal recessive disorder caused by pathogenic variants in the *ALDH3A2* gene. In general, individuals have two copies of the *ALDH3A2* gene. Carriers of Sjögren-Larsson syndrome have a variant in one copy of the *ALDH3A2* gene while individuals with the disorder have variants in both copies of *ALDH3A2*, 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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