Sjögren-Larsson Syndrome

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of Sjögren-Larsson syndrome. Carriers typically show no symptoms. Risk for current or future pregnancies is dependent on your partner's carrier status. Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for 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 and their own personal clinical management.

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 Sjögren-Larsson syndrome, each of your children has a 1 in 4 (25%) chance to have the condition.

Sjögren-Larsson Syndrome Explained

What is Sjögren-Larsson Syndrome?

Sjögren-Larsson syndrome (SLS) in an inherited condition characterized by scaly and dry skin, neurological symptoms, and eye problems. Babies with SLS have red skin at birth, which becomes dry, rough, and scaly with a yellowish or brownish tone later in infancy. These changes in the skin are usually found throughout the body, but individuals' faces are typically not affected. These children often experience mild to severe itchiness due to these skin changes. SLS also affects individuals' nervous systems, with most affected individuals having some degree of intellectual disability ranging from mild to severe. People with SLS can also have speech difficulties (such as delayed speech or difficulty pronouncing words), seizures, and developmental delay (such as delayed crawling or walking). In general, about half of individuals with SLS will need wheelchair assistance. People with SLS can also be nearsighted and be particularly sensitive to light.

Prognosis

Prognosis is variable. With appropriate medical care, individuals with SLS can live well into adulthood. The quality of life for these individuals can often depend on the severity of their symptoms and the access to symptomatic treatment.

Treatment

Although a cure for SLS does not exist, treatments do exist to help manage the symptoms of the condition. Individuals with SLS should consider following special diets early in the disease to limit daily fat intake. Affected individuals should also try to keep their skin as hydrated as possible with daily baths and special moisturizing agents that help treat the scaly nature of the skin. It has been shown that application of calcipotriol (a vitamin D-like substance) to the skin can also help. Some research shows that treatment with a drug called zileuton can help control the itchiness of the skin, but the drug has not currently been FDA approved for this purpose. Any individuals with SLS experiencing seizures should be treated with anti-seizure therapies by a neurologist. People with SLS who have difficulty walking may benefit from physical therapy and may eventually need mobility aids, such as a wheelchair. Individuals should also seek care from an ophthalmologist if they experience any eye-related symptoms.

Resources

Foundation for Ichthyosis & Related Skin Types
https://ghr.nlm.nih.gov/condition/multiplesulfatasedeficiency
The Arc
https://www.thearc.org/
United Leukodystrophy Foundation
https://ulf.org
National Society of Genetic Counselors
https://www.nsgc.org/

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