NxGen MDx

Smith-Lemli-Opitz Syndrome

What Your Results Mean

Test results indicate that you are a carrier of Smith-Lemli-Opitz syndrome (SLOS). 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 Smith-Lemli-Opitz syndrome, each of your children has a 1 in 4 (25%) chance to have the condition.

Smith-Lemli-Opitz Syndrome Explained

What is Smith-Lemli-Opitz Syndrome?

Smith-Lemli-Opitz syndrome (SLOS) is an inherited condition characterized by small head size, developmental delays, intellectual disability or learning problems, autism-like behavioral problems, heightened sun sensitivity, and midline birth defects. Affected infants are usually severely affected with symptoms including cleft palate, weak muscle tone, feeding difficulties with persistent vomiting, and delayed growth. Male infants often have underdeveloped or deformed genitalia. Other characteristic features include distinctive facial features, small head size, intellectual disability or learning problems, and behavioral problems. Many children with this condition have autism, a developmental condition that affects communication and social interaction. Other common features include fused second and third toes, extra fingers or toes, and abnormalities of the heart, lungs, kidneys, and gastrointestinal tract. The signs and symptoms vary widely, as some affected children can produce some cholesterol, which is associated with a milder form of this condition. In general, those with the milder form have only minor physical problems with learning and behavioral problems.

Prognosis

The prognosis depends on the severity of symptoms. Those with significant heart and/or kidney issues may not survive beyond childhood. However, with adequate nutrition and medical care, many individuals with this condition can live well into adulthood. Most individuals with intellectual disability, however, are unable to live independently.

801 Broadway Ave NW, Suite 203, Grand Rapids, MI 49504 • Phone: (855) 776-9436 • Fax: (616) 710-4667 info@nxgenmdx.com
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Treatment

Although there is no cure for this condition, treatment for the various associated symptoms is available. Most individuals receive dietary supplementation with large amounts of cholesterol, which helps alleviate some symptoms. This can either be in the form of purified cholesterol or foods high in cholesterol such as egg yolks. Early intervention along with physical, occupational, and speech therapy is often necessary to assist with developmental and cognitive issues. Other issues such as gastrointestinal reflux, constipation, and vomiting can be managed with medications and consultations with specialists. Careful monitoring of weight gain and growth is also necessary. Surgical repair may be required for the hands and/or feet. Many children cannot tolerate any exposure to sunlight; others can tolerate varying periods of exposure if properly clothed and protected with sunscreen.

Resources
Smith-Lemli-Opitz/RSH Foundation
https://www.smithlemliopitz.org/
National Society of Genetic Counselors
https://www.nsgc.org/

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