Genetics Uncoded:

Clarity Genetics

Facts about Smith-Lemli-Opitz Syndrome

What Your Test Results Mean

Carriers typically show no symptoms of Smith-Lemli-Opitz syndrome (SLOS); however, carriers are at an increased risk of having a child with SLOS. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Smith-Lemli-Opitz Syndrome Explained

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. The disorder affects the body's ability to produce cholesterol. Cholesterol is important for pre- and postnatal development because it is a structural component of cell membranes and the protective substance covering nerve cells (myelin) and also plays a role in the production of certain hormones and digestive acids. Treatment for SLOS is symptomatic. Life expectancy is less than two years in the most severe forms of the disorder but is dependent on the presence of severe symptoms.

How the Genetics Work

The clinical features of SLOS can be explained by variants in the *DHCR7* gene. In general, individuals have two copies of the DHCR7 gene. Carriers of SLOS have a single variant in one copy of the *DHCR7* gene while individuals with SLOS 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 **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.

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