Short Chain Acyl-CoA Dehydrogenase Deficiency

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of short-chain Acyl-CoA dehydrogenase deficiency (SCADD). 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 SCADD, each of your children has a 1 in 4 (25%) chance to have the condition.

Short Chain Acyl-CoA Dehydrogenase Deficiency Explained

What is Short-Chain Acyl-Coa Dehydrogenase?

Short-chain Acyl-CoA dehydrogenase deficiency (SCADD) is an inherited metabolic disorder that prevents the body from converting certain types of fats into energy. Its symptoms can be triggered by illness or long periods without food.

Infants affected by the disease may display episodes of vomiting, low blood sugar, and fatigue. These episodes can be fatal. Affected infants may have difficulty feeding and fail to grow at the expected rate. Some show poor muscle tone, seizures, and small head size. If the disease is untreated, the child may show developmental delays and permanent learning difficulties.

Some people with SCADD do not display any symptoms until adulthood. In these cases, the main symptom is chronic muscle weakness. Some may experience periods of pain, nausea, and shortness of breath. It is thought that many cases are so mild that they are never diagnosed.

Prognosis

In individuals with SCADD who display symptoms, response to treatment varies. Some treated individuals may live typical, healthy lives, while others may continue to experience learning delays and muscle weakness. Acute, life-threatening episodes are rare.

Treatment

For individuals who are asymptomatic, treatment entails avoiding both dehydration and long periods of fasting (fasting for longer than 12 hours). Intravenous fluids and sugars may be given to affected individuals having a metabolic crisis.

Resources

Fatty Oxidation Disorders Family Support Group
http://www.fodsupport.org/

National Society of Genetic Counselors
https://www.nsgc.org/

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