Very Long Chain Acyl-CoA Dehydrogenase Deficiency

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of very long chain acyl-CoA dehydrogenase deficiency (VLCAD). Carriers are not expected to show symptoms. You and your partner would both have to be carriers of VLCAD for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a 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.

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 are carriers for VLCAD, each of your children has a 1 in 4 (25%) chance to have the condition.

Very Long Chain Acyl-CoA Dehydrogenase Deficiency Explained

What is Very Long Chain Acyl-Coa Dehydrogenase Deficiency?

VLCAD is an inherited disorder that prevents the body from converting certain fats to energy, particularly during fasting periods. Symptoms of VLCAD usually appear during infancy or early childhood and can include low blood sugar, lack of energy, and muscle weakness. Serious complications include liver abnormalities and life-threatening heart problems. Individuals with the later onset form may experience muscle breakdown that can cause damage to the kidneys. The later the symptoms first develop, the milder the disorder. Affected individuals exhibit the most problems during periods of fasting, illness, and exercise.

Prognosis

With early diagnosis and treatment, prognosis is very good. Affected individuals can live symptom-free with proper management. If left untreated however, the disease could be fatal.

Treatment

Treatment for affected individuals involves a special diet and the avoidance of fasting. Management is typically overseen by a group of metabolic specialists and is usually lifelong.

Resources

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

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/very-long-chain-acyl-coa-dehydrogenase-deficiency

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

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