Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of long chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD). Carriers are not expected to show symptoms. You and your partner or donor would both have to be carriers of LCHAD 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 or donor are carriers for LCHAD, each of your children has a 1 in 4 (25%) chance to have the condition.

Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency Explained

What is Long Chain 3-Hydroxyacyl-Coa Dehydrogenase Deficiency?

LCHAD deficiency is an inherited metabolic disorder that prevents the body from converting certain types of fats into energy. LCHAD deficiency is usually diagnosed in infancy or early childhood, when the affected individual presents with symptoms such as vomiting, low blood sugar, lack of energy, muscle weakness, liver problems, and failure to thrive. Partially metabolized fatty acids can accumulate in body tissues and cause organ damage if the disease goes untreated. This puts affected individuals at a higher risk for heart and breathing problems, comas, and seizures. The symptoms are especially noticeable when the individual goes for a long time between meals, suffers from a viral infection, or engages in intense exercise. Thus, it is important to ensure that children with LCHAD deficiency eat frequent meals, preferably high in carbohydrates and low in fats. Women whose fetuses have LCHAD deficiency are at an increased risk for pregnancy complications and should consult their doctor.

Prognosis

Prognosis is typically poor, with almost 40% of affected infants dying in the first three months of life. Morbidity in those that survive the first few months is also high, with recurrent metabolic crises and muscle problems despite therapy.

Treatment

Management is typically overseen by a group of metabolic specialists. Treatment involves a low-fat, high-carbohydrate diet. Survival can be improved by early diagnosis, but morbidity remains high despite current treatment options.

Resources

Genetic and Rare Diseases Information Center
https://rarediseases.info.nih.gov/diseases/6867/lchad-deficiency
The FOD (Fatty Oxidation Disorders) Family Support Group
http://www.fodsupport.org/index.htm
National Society of Genetic Counselors
https://www.nsgc.org/

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