Genetics Uncoded:

Facts about

Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency

What Your Test Results Mean

Carriers typically show no symptoms of long chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD); however, carriers are at an increased risk of having a child with LCHAD. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency Explained

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. Management is typically overseen by a group of metabolic specialists. Women whose fetuses have LCHAD deficiency are at an increased risk for pregnancy complications and should consult their doctor.

How the Genetics Work

LCHAD is an autosomal recessive metabolic disorder caused by pathogenic variants in the *HADHA* gene. In general, individuals have two copies of the *HADHA* gene. Carriers of LCHAD deficiency have a single variant in one copy of the *HADHA* gene while individuals with LCHAD deficiency 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 **1-855-776-9436** to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.

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