Medium Chain Acyl-CoA Dehydrogenase Deficiency

NxGen MDx

What Your Results Mean

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

Medium Chain Acyl-CoA Dehydrogenase Deficiency Explained

What is Medium Chain Acyl-CoA Dehydrogenase Deficiency?

MCAD is an inherited metabolic disorder that prevents the body from converting certain types of fats (medium chain fatty acids) into energy. MCAD is usually diagnosed in infancy or early childhood, when the affected individual presents with symptoms such as vomiting, low blood sugar, lack of energy, and failure to thrive. Partially metabolized fatty acids can accumulate in body tissues and cause organ damage if the disease goes untreated. The symptoms are especially noticeable when the individual goes for a long time between meals or suffers from a viral infection. Thus, it is important to ensure that children with MCAD eat frequent meals, preferably high in carbohydrates and low in fats. When left untreated, individuals may be at risk for seizures, breathing difficulties, liver problems, brain damage, coma, and sudden death.

Fatty acid oxidation disorders, such as MCAD, have been reported to increase the risk for maternal complications in the third trimester of pregnancy when the baby is affected. Additional pregnancy monitoring may be recommended in these cases.

Prognosis

The prognosis of an affected individual depends on the severity of the symptoms but is usually good if the disease is diagnosed early and careful medical management is provided.

Treatment

The primary focus of treatment is to avoid fasting. Infants will require frequent feedings, and toddlers will need a low-fat diet and could receive uncooked cornstarch at bedtime to sustain glucose levels overnight. Affected individuals should take simple carbohydrates by mouth (e.g., glucose tablets or sweetened, non-diet beverages) or IV if needed, to avoid fasting.

Resources

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

National Organization for Rare Disorders
https://rarediseases.org/rare-diseases/medium-chain-acyl-coa-dehydrogenase-deficiency/

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

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