Genetics Uncoded:

Facts about

Medium Chain Acyl-CoA Dehydrogenase Deficiency

What Your Test Results Mean

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

Medium Chain Acyl-CoA Dehydrogenase Deficiency Explained

MCADD is an inherited metabolic disorder that prevents the body from converting certain types of fats into energy. MCADD 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 MCADD eat frequent meals, preferably high in carbohydrates and low in fats. 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. Management is typically overseen by a group of metabolic specialists.

How the Genetics Work

MCADD is an autosomal recessive metabolic disorder caused by pathogenic variants in the *ACADM* gene. In general, individuals have two copies of the *ACADM* gene. Carriers of MCADD have a single variant in one copy of the *ACADM* gene while individuals with MCADD 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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