Genetics Uncoded:

NxGen MDx

Facts about

Carnitine Palmitoyltransferase IA Deficiency

What Your Test Results Mean

Carriers typically show no symptoms of carnitine palmitoyltransferase IA (CPT1A) deficiency; however, carriers are at an increased risk of having a child with CPT1A deficiency. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Carnitine Palmitoyltransferase IA Deficiency Explained

CPT1A deficiency is an inherited metabolic disorder that prevents the body from converting certain types of fats into energy. Individual with CPT1A are developmentally and cognitively normal but during periods of metabolic crisis may show symptoms including vomiting, low blood sugar, lack of energy, and failure to thrive that may result in organ damage without proper management. A high-carbohydrate, low-fat diet is recommended and fasting should be avoided. Diet and medications are typically managed by a metabolic physician and dietician. Pregnant women whose fetus has CPT1A deficiency are at risk for pregnancy complications such as fatty liver.

How the Genetics Work

CPT1A deficiency is an autosomal recessive disorder caused by pathogenic variants in the *CPT1A* gene. In general, individuals have two copies of the *CPT1A* gene. Carriers of CPT1A deficiency have a single variant in one copy of the *CPT1A* gene while individuals with CPT1A 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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