Genetics Uncoded:

Clarity Genetics

Facts about

Carnitine Palmitoyltransferase II (CPT2) Deficiency

What Your Test Results Mean

Carriers typically show no symptoms of carnitine palmitoyltransferase II (CPT2) deficiency; however, carriers are at an increased risk of having a child with CPT2 deficiency. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Carnitine Palmitoyltransferase II Deficiency Explained

CPT2 deficiency is an inherited metabolic disorder that prevents the body from converting certain types of fats into energy. There are three forms of the disease: the lethal neonatal form, the severe infantile hepatocardiomuscular form, and the myopathic form. Symptoms range from liver failure and sudden death in the most severe form to episodes of muscle pain and damage triggered by exercise, stress, extreme temperature, infections, or fasting in the myopathic form. 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.

How the Genetics Work

CPT2 deficiency is an autosomal recessive disorder caused by variants in the *CPT2* gene. In general, individuals have two copies of the *CPT2* gene. Carriers of CPT2 deficiency have a single variant in one copy of the *CPT2* gene while individuals with CPT2 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 **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.

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