GENETICS UNCoded:
FACTS ABOUT CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY

**AKA** | Carnitine palmitoyltransferase 2 deficiency, CPT2 deficiency

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### What Your Test Results Mean

Testing results indicate that you are a carrier of carnitine palmitoyltransferase II (CPT2) deficiency. Carriers typically show no symptoms of CPT2 deficiency; however, carriers are at an increased risk of having a child with CPT2 deficiency. Risk for the current or future pregnancies is dependent on your partner’s carrier status. Carrier testing of your partner is recommended in addition to consultation with a genetic counselor.

### 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 hepatocardiomyosascular 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 mutations in the CPT2 gene. In general, individuals have two copies of the CPT2 gene. Carriers of CPT2 deficiency have a single mutation in one copy of the CPT2 gene while individuals with CPT2 deficiency have mutations in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

### Recommended Next Steps

Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

### Questions?

Contact us at (855) 776-9436 to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.