Genetics Uncoded:

NxGen MDx

Facts about Primary Carnitine Deficiency

What Your Test Results Mean

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

Primary Carnitine Deficiency Explained

Primary carnitine deficiency is an inherited metabolic disorder that impairs the body's uptake of carnitine, a molecule necessary for converting lipids into metabolic energy. Signs and symptoms of primary carnitine deficiency typically appear during infancy or early childhood and can include severe brain dysfunction, a weakened and enlarged heart, confusion, vomiting, muscle weakness, and low blood sugar. The severity of this condition varies among affected individuals and can usually be effectively treated with an oral carnitine supplement. While primary carnitine deficiency can be serious and even fatal without treatment, individuals with the deficiency who take regular supplements can typically live a normal, healthy life. Management is often overseen by a group of metabolic specialists.

How the Genetics Work

Primary carnitine deficiency is an autosomal recessive disorder caused by variants in the *SLC22A5* gene. In general, individuals have two copies of the *SLC22A5* gene. Carriers of primary carnitine deficiency have a single variant in one copy of the *SLC22A5* gene while individuals with primary carnitine deficiency have variants in both copies of their genes, one inherited from each parent. Approximately 70% of variants can be detected by sequencing. 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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