NxGen MDx

Carnitine Palmitoyltransferase IA Deficiency

What Your Results Mean

Test results indicate that you are a carrier of carnitine palmitoyltransferase IA deficiency. Carriers are not expected to show symptoms of this deficiency. You and your partner would both have to be carriers of carnitine palmitoyltransferase IA deficiency for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment.

Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning.

Recommended Next Steps

Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner are carriers for carnitine palmitoyltransferase IA deficiency, each of your children has a 1 in 4 (25%) chance to have the condition.

Carnitine Palmitoyltransferase IA Deficiency Explained

What is Carnitine Palmitoyltransferase IA Deficiency?

Carnitine palmitoyltransferase IA deficiency (CPT1A deficiency) is an inherited metabolic disorder that prevents the body from converting certain types of fats into energy. Individuals with CPT1A deficiency 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. Without proper management, CPT1A deficiency results in organ damage. Pregnant women whose fetus has a diagnosis of CPT1A deficiency are at risk for pregnancy complications such as fatty liver.

Prognosis

Prognosis is considered favorable with treatment. However, if left untreated, irreversible neurological damage can occur and the disease can be fatal. As long as individuals are managing CPT1A deficiency with treatment, most can live fairly normal lives.

Treatment

A high-carbohydrate, low-fat diet is recommended, and fasting should be avoided. Infants are recommended to eat frequently during the day and have uncooked cornstarch continuously at night. Diet and medications are typically managed by a metabolic physician and dietitian.

Resources

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/carnitine-palmitoyltransferase-i-deficiency#
National Organization for Rare Disorders (NORD)
https://rarediseases.org/rare-diseases/carnitine-palmitoyltransferase-1a-deficiency/
National Society of Genetic Counselors
https://www.nsgc.org/

801 Broadway Ave NW, Suite 203, Grand Rapids, MI 49504 • Phone: (855) 776-9436 • Fax: (616) 710-4667 info@nxgenmdx.com
nxgenmdx.com

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