Carnitine Palmitoyltransferase II Deficiency

What Your Results Mean

Test results indicate that you are a carrier of carnitine palmitoyltransferase II deficiency. Carriers are not expected to show symptoms of this deficiency. You and your partner would both have to be carriers of carnitine palmitoyltransferase II 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 II deficiency, each of your children has a 1 in 4 (25%) chance to have the condition.

Carnitine Palmitoyltransferase II Deficiency Explained

What is Carnitine Palmitoyltransferase II Deficiency?

Carnitine palmitoyltransferase II deficiency (CPT II 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. In the most severe form of CPT II deficiency, the lethal neonatal form, infants first begin to show signs soon after birth. Symptoms can include respiratory failure, liver failure, seizures, cardiomyopathy (weakened heart muscle), arrhythmia, or sudden death. Infants can also have episodes of hypoketotic hypoglycemia which is characterized by low blood sugar and low levels of ketones (products of the breakdown of fat that are used for energy).

In the severe infantile hepatocardiomuscular form of CPT II deficiency, signs and symptoms typically appear during the first year of life. Symptoms can include episodes of hypoketotic hypoglycemia, seizures, an enlarged liver (hepatomegaly), cardiomyopathy, and arrhythmia.

Individuals with the mildest form of CPT II deficiency, the myopathic form, can first show signs and symptoms between childhood and late adulthood. Symptoms can include muscle pain and weakness triggered by periods of fasting, exercise, extreme temperatures, infections, or stress.

Prognosis

Prognosis depends on the type of CPT II deficiency. Infants with the lethal neonatal form usually live between a few days to a few months. Individuals with the severe infantile hepatocardiomuscular type of CPT II deficiency are at risk for liver failure, nervous system damage, coma, and sudden death. Prognosis is considered favorable for individuals with the myopathic form of CPT II deficiency, as many do not have symptoms of the disorder between episodes. Episodes are typically manageable and individuals with this mild form can usually lead a fairly normal life.

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Treatment

Treatment for the lethal neonatal form and the severe infantile hepatocardiomuscular form is symptomatic as there is no cure for CPT II deficiency. For individuals with the myopathic form, treatment typically includes a high-carbohydrate, low-fat diet where fasting is avoided. Carnitine supplements may also be recommended. Diet and medications are typically managed by a metabolic physician and dietitian.

Resources

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/carnitine-palmitoyltransferase-ii-deficiency
National Society of Genetic Counselors
https://www.nsgc.org/

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