NxGen MDx

Primary Carnitine Deficiency

What Your Results Mean

Test results indicate that you are a carrier of primary carnitine deficiency. Carriers typically show no symptoms. Risk for 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 for 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 and their own personal clinical management.

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 or donor are carriers for primary carnitine deficiency, each of your children has a 1 in 4 (25%) chance to have the condition.

Primary Carnitine Deficiency Explained

What is Primary Carnitine Deficiency?

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. The disease has a variable spectrum of symptoms, from a severe infantile presentation to completely asymptomatic adults. In affected infants, periods of fasting or illness can trigger symptoms, which include low blood sugar, lethargy, and irritability. These symptoms must be treated immediately or there is a risk of coma and death. Affected individuals who present during childhood can have dilated cardiomyopathy and skeletal muscle weakness. These affected children can die from cardiac failure if not treated promptly. Some affected individuals can be mostly asymptomatic throughout their lives, though there is still risk for cardiac symptoms.

Prognosis

Prognosis is good with treatment but can be extremely poor if left untreated.

Treatment

L-carnitine supplementation is the preferred method of treatment for this disorder. Individuals respond well to treatment if started before irreversible organ damage occurs and there are relatively few side effects.

Resources

Fatty Oxidation Disorders Family Support Group
http://www.fodsupport.org/

National Society of Genetic Counselors
https://www.nsgc.org/

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