Methylmalonic Acidemia, *MMAA*-Related

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of methylmalonic acidemia, *MMAA*-related. Carriers are not expected to show symptoms. You and your partner or donor would both have to be carriers of methylmalonic acidemia, *MMAA*-related 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 or donor are carriers for methylmalonic acidemia, *MMAA*-related, each of your children has a 1 in 4 (25%) chance to have the condition.

Methylmalonic Acidemia, *MMAA*-Related Explained

What is Methylmalonic Acidemia, *MMAA*-Related?

Methylmalonic acidemia is an inherited disorder in which the body is unable to process certain proteins and fats properly. Symptoms typically present in infancy and vary from mild to life-threatening. Affected infants can experience vomiting, dehydration, weak muscle tone (hypotonia), developmental delay, excessive tiredness (lethargy), an enlarged liver (hepatomegaly), and failure to gain weight and grow at the expected rate. Long-term complications can include feeding problems, intellectual disability, chronic kidney disease, and inflammation of the pancreas. Without treatment, this disorder can lead to coma and death in some cases.

Prognosis

Prognosis varies and is often dependent on the age of onset of symptoms, the severity of symptoms, and the responsiveness to treatment. In general, early diagnosis and treatment is associated with a better outcome. Some affected individuals have lifelong learning problems, intellectual disability, seizures, or growth delay, even with treatment.

Treatment

Treatment for methylmalonic acidemia involves vitamin B12 supplementation for the vitamin B12 responsive subtypes, L-carnitine supplements, a special protein managed diet, avoidance of fasting, and management of symptoms as they appear. Methylmalonic acidemia, *MMAA*-related is typically non-vitamin B12 responsive.

Resources

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/methylmalonic-acidemia#resources

Genetic and Rare Diseases Information Center
https://rarediseases.info.nih.gov/diseases/7033/methylmalonic-acidemia

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

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