NxGen MDx

Glutaric Acidemia Type I

What Your Results Mean

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

Glutaric Acidemia Type I Explained

What is Glutaric Acidemia Type I?

Glutaric acidemia type I is an inherited metabolic disorder caused by a missing enzyme, known as glutaryl-CoA dehydrogenase. Without this enzyme, the body cannot break down the amino acids lysine and tryptophan. Symptoms vary widely; some individuals are only mildly affected, while others have severe problems. Affected individuals may present with metabolic crisis or low blood sugar, leading to severe vomiting and lethargy, seizures, and other complications in times of illness, fever, or fasting. If untreated, glutaric acidemia type I can lead to brain damage, intellectual disabilities, loss of motor skills, and death.

Prognosis

With early diagnosis and appropriate treatment and management, prognosis is typically favorable. If left untreated or improperly managed, neurologic damage can be irreversible.

Treatment

To avoid a metabolic crisis, affected individuals should avoid fasting and may be placed on a special diet. In the event of a metabolic crisis, affected individuals must undergo prompt and comprehensive emergency treatment. Diet and medications are typically managed by a metabolic physician and dietitian.

Resources

National Organization for Rare Disorders
https://rarediseases.org/rare-diseases/glutaricaciduria-i/

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/glutaric-acidemia-type-i#genes

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

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