Isovaleric Acidemia

NxGen MDx

What Your Results Mean

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

Isovaleric Acidemia Explained

What is Isovaleric Acidemia?

Isovaleric acidemia is an inherited metabolic condition characterized by an inability to break down the amino acid leucine due to having inadequate levels of the enzyme isovaleryl-CoA dehydrogenase. Due to the lack of this enzyme, proteins cannot be processed properly and organic acids, including isovaleric acid, build-up in the body, which can cause a variety of health issues.

Initial symptoms may be present shortly after birth and include vomiting, poor feeding, and lethargy. These symptoms can progress to serious medical concerns such as seizures, coma, and potentially death. The build-up of isovaleric acid causes a distinct odor similar to sweaty feet during acute illness. Other medical problems that characterize this disorder are failure to grow and gain weight at a normal rate (failure to thrive), as well as delays in achieving developmental milestones.

Prognosis

Prognosis is only poor when the condition goes undiagnosed and/or untreated. When left untreated, individuals progress into a coma and death. However, when diagnosed early and properly treated, prognosis is good, and a normal lifespan is expected.

Treatment

Treatment consists of restricting protein in the diet. Avoiding protein-rich foods, infection, and fasting (going for long periods without eating) can help prevent symptoms of the disease.

Resources

National Organization for Rare Disorders
https://rarediseases.org/rare-diseases/acidemia-isovaleric/

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/isovaleric-acidemia

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

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