Dihydrolipoamide Dehydrogenase Deficiency

NxGen MDx

What Your Results Mean

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

Lipoamide Dehydrogenase Deficiency Explained

What is Dihydrolipoamide Dehydrogenase Deficiency?

Dihydrolipoamide dehydrogenase deficiency, which is also known as maple syrup urine disease type III, is an inherited metabolic disorder in which the body is unable to process certain amino acids. Infants typically present with a buildup of lactic acid (lactic acidosis), which can cause feeding and breathing problems, vomiting, fatigue, decreased muscle tone, seizures, and developmental delay. The signs and symptoms of dihydrolipoamide dehydrogenase deficiency typically occur periodically during episodes that may be triggered by fever, injury, or other stresses on the body. Affected individuals are usually symptom-free between episodes. Infants typically do not survive past the first few years of life due to the severity of the episodes. Individuals who do survive often have delayed growth and neurological problems, including intellectual disability, muscle stiffness (spasticity), difficulty coordinating movements (ataxia), and seizures.

Prognosis

Prognosis is typically poor for those with symptoms that begin in infancy, and these individuals do not usually survive past the first few years of life. Those who do survive past early childhood have a slightly better prognosis but still experience symptoms and intellectual disability. Individuals with late onset dihydrolipoamide dehydrogenase deficiency have the best prognosis but still have liver complications.

Treatment

There is no cure for dihydrolipoamide dehydrogenase deficiency, and treatment is usually supportive.

Resources

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/dihydrolipoamide-dehydrogenase-deficiency
Genetic and Rare Diseases Information Center
https://rarediseases.info.nih.gov/diseases/3263/dihydrolipoamide-dehydrogenase-deficiency
National Society of Genetic Counselors
https://www.nsgc.org/

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