NxGen MDx

Abetalipoproteinemia

What Your Results Mean

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

Abetalipoproteinemia Explained

What is Abetalipoproteinemia?

Abetalipoproteinemia is an inherited condition that affects the ability of an individual to absorb dietary fats, cholesterol, and fat-soluble vitamins. Certain levels of fats, cholesterol, and vitamins are necessary for normal growth, development, and maintenance of the body. However, individuals affected by this disorder are not able to make certain lipoproteins, which are particles that carry fats and fat-like substances (such as cholesterol) in the blood. Signs and symptoms of abetalipoproteinemia first show around infancy. These symptoms can include a failure to gain weight and grow at the expected rate, diarrhea, abnormally star-shaped red blood cells (acanthocytosis), and fatty, foul-smelling stools. Additional symptoms may occur later in childhood such as ataxia, poor muscle coordination, and an eye disorder called retinitis pigmentosa which could cause blindness by the fourth decade of life. Adults with abetalipoproteinemia may also have difficulties with balance and walking.

Prognosis

Signs and symptoms for abetalipoproteinemia can vary. As such, prognosis can depend on the severity of brain and nervous system involvement. Early diagnosis and treatment can potentially improve quality of life and reduce neurological and ophthalmological symptoms.

Treatment

A low-fat diet and administration of fat-soluble vitamins may be used to manage symptoms, but there is no cure for abetalipoproteinemia.

Resources

National Organization for Rare Disorders (NORD)
https://rarediseases.org/rare-diseases/abetalipoproteinemia/
National Society of Genetic Counselors
https://www.nsgc.org/

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