NxGen MDx

Ataxia with Vitamin E Deficiency

What Your Results Mean

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

Ataxia with Vitamin E Deficiency Explained

What is Ataxia with Vitamin E Deficiency?

Ataxia with vitamin E deficiency (AVED) is an inherited disorder where individuals are unable to properly retain and use dietary vitamin E. This can cause damage to the body's cells, especially nerve cells in the brain and spinal cord. Individuals typically begin to exhibit symptoms between the ages of 5 to 15. Signs and symptoms can include neurological problems such as difficulty coordinating movements (ataxia) and speech, loss of reflexes in the legs, and a loss of sensation in the extremities. Affected individuals may also develop retinitis pigmentosa, an eye disorder that causes vision loss. Most individuals with AVED begin to experience symptoms before adulthood and if untreated, may require use of a wheelchair by adulthood.

Prognosis

Prognosis is considered favorable as long as individuals start treatment prior to signs and symptoms. This can prevent or improve symptoms. If untreated, individuals become wheelchair-dependent between 11 to 50 years old.

Treatment

Treatment for AVED consists of vitamin E supplementation in high doses. This can prevent or improve many of the symptoms associated with AVED.

Resources
National Ataxia Foundation
https://ataxia.org/
Genetics Home Reference
https://ghr.nlm.nih.gov/condition/ataxia-with-vitamin-e-deficiency
National Organization for Rare Disorders (NORD)
https://rarediseases.org/rare-diseases/ataxia-with-vitamin-e-deficiency/
National Society of Genetic Counselors
https://www.nsgc.org/

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