Genetics Uncoded:

Clarity Genetics

Facts about
Ataxia with Vitamin E Deficiency

What Your Test Results Mean

Carriers typically show no symptoms of ataxia with vitamin E deficiency (AVED). Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Ataxia with Vitamin E Deficiency Explained

Ataxia with vitamin E deficiency is an inherited disorder characterized by neurological problems, such as difficulty coordinating movements (ataxia) and speech, loss of reflexes in the legs, and a loss of sensation in the extremities. Individuals with AVED are unable to properly retain and use dietary vitamin E, causing damage to the body's cells, especially nerve cells in the brain and spinal cord. Most individuals with AVED begin to experience symptoms before adulthood, and if untreated may require use of a wheelchair by adulthood. If treated early and consistently with vitamin E, symptoms of the disease can be avoided.

How the Genetics Work

Ataxia with vitamin E deficiency is an autosomal recessive disorder caused by variants in the *TTPA* gene. In general, individuals have two copies of the *TTPA* gene. Carriers of AVED have a variant in one copy of the *TTPA* gene while individuals with AVED have variants in both copies of the *TTPA* gene, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Questions? Contact us at **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.

Clarity Genetics, LLC
5231 NW 33rd Ave
Ft. Lauderdale, FL 33309
Revised 12/15/16