Inheritest®
CARRIER SCREEN

Ataxia with vitamin E deficiency

What is ataxia with vitamin E deficiency?

Ataxia with vitamin E deficiency is a neurodegenerative inherited disease with variable severity and onset typically in childhood and adolescence.[1] Individuals with ataxia with vitamin D deficiency have a deficiency of the alpha-tocopherol transfer protein, which helps the body distribute vitamin E.[2] As an antioxidant, vitamin E helps to protect cells in the body from damage by free radicals.[1] Symptoms are due to free radical damage to tissues and cells and may include progressive ataxia, areflexia, vibratory and proprioceptive sensory loss, dysarthia, and decreased visual acuity.[3,4] Ataxia with vitamin E deficiency is also known as Friedreich-like ataxia.[1]

What are the symptoms of ataxia with vitamin E deficiency and what treatment is available?

Symptoms are variable and can start in childhood; however, most affected individuals show symptoms between 5 and 15 years of age.[1,4] Symptoms may include[1,3,4]

- Progressive ataxia (loss of muscle coordination)
- Dysarthria (slurred speech)
- Lower limb areflexia (absence of reflexes)
- Peripheral nerve dysfunction
- Retinitis pigmentosa (degenerative eye disease)
- Cardiomyopathy (abnormal heart muscle)
- Scoliosis (curvature of the spine)
- Reduction in sensitivity to pain or touch
- Impaired swallowing
- Impaired movement or muscle control

Treatment is primarily dietary, involving therapeutic vitamin E supplementation.[3]

How is ataxia with vitamin E deficiency inherited?

Ataxia with vitamin E deficiency is an autosomal recessive disease caused by mutations in the *TTPA* gene.[1] An individual who inherits one copy of a *TTPA* gene mutation is a carrier and is not expected to have related health problems. An individual who inherits two mutations in the *TTPA* gene, one from each parent, is expected to be affected with ataxia with vitamin E deficiency.

If both members of a couple are carriers of mutations in the same gene, the risk for an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

Who is at risk for ataxia with vitamin E deficiency?

Ataxia with vitamin E deficiency is a rare condition and its prevalence is unknown.

Having a relative who is a carrier or who is affected can increase an individual's risk to be a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetics health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

Where can I get more information?

Genetics Home Reference: http://ghr.nlm.nih.gov/condition/ataxia-with-vitamin-e-deficiency

National Organization for Rare Disorders (NORD): http://rarediseases.org/rare-diseases/ataxia-with-vitamin-e-deficiency/

National Ataxia Foundation: http://www.ataxia.org/

References

1. Ataxia with vitamin E deficiency. *Genetics Home Reference.* Available at: http://ghr.nlm.nih.gov/condition/ataxia-with-vitamin-e-deficiency. Accessed February 12, 2016.
2. *TTPA. Genetics Home Reference.* Available at: http://ghr.nlm.nih.gov/gene/TTPA. Accessed February 12, 2016.
3. Ataxia with Vitamin E Deficiency. *National Organization for Rare Disorders.* Available at: http://rarediseases.org/rare-diseases/ataxia-with-vitamin-e-deficiency/. Accessed February 12,2016.
4. Schuelke, M. Ataxia with Vitamin E Deficiency. *GeneReviews* Available at: http://www.ncbi.nlm.nih.gov/books/NBK1241/. Accessed February 12, 2016.