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. Individuals with ataxia with vitamin D deficiency have a deficiency of the alpha-tocopherol transfer protein, which helps the body distribute vitamin E. As an antioxidant, vitamin E helps to protect cells in the body from damage by free radicals. Symptoms are due to free radical damage to tissues and cells and may include progressive ataxia, areflexia, vibratory and proprioceptive sensory loss, dysarthria, and decreased visual acuity.

Ataxia with vitamin E deficiency is also known as Friedreich-like ataxia.

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. Symptoms may include:

- 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.

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. 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?


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

References