Abetalipoproteinemia

What is abetalipoproteinemia?

Abetalipoproteinemia is a rare inherited disease of lipoprotein metabolism characterized by hypocholesterolemia and malabsorption of lipid-soluble vitamins. Production of the microsomal triglyceride transfer protein is impaired, which leads to an absence of beta-lipoproteins. Without these lipoproteins, the body has difficulty absorbing cholesterol, dietary fats, and fat-soluble vitamins. Abetalipoproteinemia is also known as Bassen-Kornzweig syndrome.

What are the symptoms of abetalipoproteinemia and what treatment is available?

Clinical features can be evident in infancy or childhood and may include:

- Failure to thrive
- Diarrhea
- Vomiting
- Acanthocytosis
- Vitamin E deficiency
- Neuropathy
- Spinocerebellar dysfunction
- Steatorrhea
- Retinitis pigmentosa
- Skeletal abnormalities including lordosis, kyphoscoliosis, and pes cavus

Treatment is primarily dietary and supportive. A low fat diet can reduce gastrointestinal symptoms while high doses of fat-soluble vitamins can help with many of the other associated symptoms.

How is abetalipoproteinemia inherited?

Abetalipoproteinemia is an autosomal recessive disease caused by mutations in the MTTP gene. An individual who inherits one copy of a MTTP gene mutation is a carrier and is not expected to have related health problems. An individual who inherits two MTTP mutations, one from each parent, is expected to be affected with abetalipoproteinemia.

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 abetalipoproteinemia?

Abetalipoproteinemia is rare, but can occur in individuals of all races and ethnicities. Carrier frequencies are 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 Organization for Rare Disorders (NORD): http://rarediseases.org/rare-diseases/abetalipoproteinemia/

References