Inheritest® CARRIER SCREEN

Abetalipoproteinemia

What is abetalipoproteinemia?

Abetalipoproteinemia is a rare inherited disease of lipoprotein metabolism characterized by hypocholesterolemia and malabsorption of lipid-soluble vitamins.[1,3] Production of the microsomal triglyceride transfer protein is impaired, which leads to an absence of beta-lipoproteins.[2] Without these lipoproteins, the body has difficulty absorbing cholesterol, dietary fats, and fat-soluble vitamins.[1] Abetalipoproteinemia is also known as Bassen-Kornzweig syndrome.[3]

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

Clinical features can be evident in infancy or childhood and may include:[1,3]

- 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.[3]

How is abetalipoproteinemia inherited?

Abetalipoproteinemia is an autosomal recessive disease caused by mutations in the *MTTP* gene.[1] 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.

LabCorp

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Where can I get more information?

Genetics Home Reference: http://ghr.nlm.nih.gov/condition/abetalipoproteinemia

National Organization for Rare Disorders (NORD): http://rarediseases.org/rare-diseases/abetalipoproteinemia/

References

1. Abetalipoproteinemia. *Genetics Home Reference.* Available at http://ghr.nlm.nih.gov/condition/abetalipoproteinemia. Accessed February 3, 2016.
2. *MTTP. Genetics Home Reference.* Available at: http://ghr.nlm.nih.gov/gene/MTTP. Accessed February 3, 2016.
3. Abetalipoproteinemia. *National Organization for Rare Disorders*. Available at http://rarediseases.org/rare-diseases/abetalipoproteinemia/. Accessed February 3,2016.