Genetics Uncoded:

Facts about Abetalipoproteinemia

What Your Test Results Mean

Carriers of abetalipoproteinemia show no symptoms and are not at risk to develop symptoms of the disorder. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Abetalipoproteinemia Explained

Abetalipoproteinemia is an inherited condition that affects the absorption of dietary fats, cholesterol, and fat-soluble vitamins. People affected by this disorder are not able to make certain lipoproteins, which are particles that carry fats and fat-like substances (such as cholesterol) in the blood. Since certain levels of fats, cholesterol, and vitamins are necessary for normal growth, development, and maintenance of the body, symptoms of abetalipoproteinemia appear during infancy in affected individuals. Infants with abetalipoproteinemia fail to gain weight and grow at the expected rate and have diarrhea, abnormal star-shaped red blood cells, and fatty, foul-smelling stools. Individuals with abetalipoproteinemia may live into adulthood, but have severe health problems and reduced life expectancy. A low fat diet and administration of fat-soluble vitamins may be used to manage symptoms, but there is no cure for abetalipoproteinemia.

How the Genetics Work

Abetalipoproteinemia is an autosomal recessive disorder that can be explained by pathogenic variants in the *MTTP* gene. In general, individuals have two copies of the *MTTP* gene. Carriers of abetalipoproteinemia have a single pathogenic variant in one copy of the *MTTP* gene while individuals with abetalipoproteinemia have pathogenic variants in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Questions?

Contact us at **1-855-776-9436** to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.

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Revised 12/15/16