Genetics Uncoded:

FACTS ABOUT ABETALIPOPROTEINEMIA

AKA | Abetalipoproteinemia neuropathy, acanthocytosis, Apolipoprotein B deficiency, Bassen-Kornzweig Syndrome Betalipoprotein Deficiency Disease, Congenital betalipoprotein deficiency syndrome, Microsomal Triglyceride Transfer Protein Deficiency Disease

What Your Test Results Mean

Testing results indicate that you are a carrier of abetalipoproteinemia. Carriers typically show no symptoms of abetalipoproteinemia; however, carriers are at an increased risk of having a child with abetalipoproteinemia. Risk for the current or future pregnancies is dependent on your partner’s carrier status. Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

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 mutations in the MTTP gene. In general, individuals have two copies of the MTTP gene. Carriers of abetalipoproteinemia have a single mutation in one copy of the MTTP gene while individuals with abetalipoproteinemia have mutations in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Recommended Next Steps

Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

Questions?

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