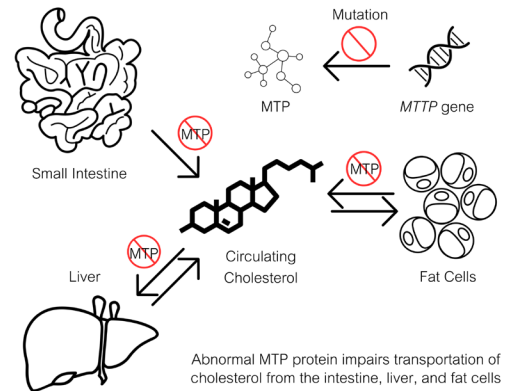


What is Abetalipoproteinemia (MTP Deficiency)?

Abetalipoproteinemia (ABL) is a very rare, inherited, autosomal recessive disorder with a prevalence of less than one in a million. ABL is caused by a mutation in the MTP gene which codes for microsomal triglyceride protein (MTP). Abnormal MTP function leads to compromised absorption and transportation of fat (and fat-soluble vitamins) in the intestine and the liver. Therefore, individuals with ABL typically have very low blood cholesterol, particularly LDL-cholesterol (LDL-C), and VLDL cholesterol (VLDL-C). This leads to malnutrition, nervous system problems, and abnormal red blood cells count.



What are the symptoms of Abetalipoproteinemia?

Most babies with ABL start showing symptoms when they are very young because they can't properly absorb fats and certain vitamins from their food.

Early Symptoms

- » Fatty diarrhea
- » Vomiting
- » Failure to thrive
- » Abnormal red blood cells and anemia



Late Symptoms

- » Muscle weakness
- » Numbness in hands and feet (Peripheral neuropathy)
- » Vision problems
- » Fatty liver and liver enlargement
- » Bleeding disorder

How is Abetalipoproteinemia diagnosed?

The diagnosis of ABL is made based on symptoms and blood tests. Genetic testing is required for a definite diagnosis. Other tests are sometimes obtained to monitor for damage to other organs (heart, liver, eye, and nervous system).



Medical Care

As ABL can affect multiple organ systems, multidisciplinary care is the key to management for ABL patients. The team usually involves a lipidologist, gastroenterologist, hematologist, neurologist, ophthalmologist, and a physical and occupational therapist. The patient's family should discuss the risks of ABL with a genetic counselor. After diagnosis, the patient should undergo routine evaluation by specialists. physical examination, blood tests, and other specific studies are obtained periodically to ensure normal growth and development and surveillance for complications.



Treatment Options

- » Reduction of dietary fat to prevent diarrhea.
- » Essential fatty acid supplement. Medium-chain triglyceride is sometimes administered in infants.
- » Fatty acid and triglyceride supplementation are given should neurological or visual symptoms develop.
- » Fat-soluble vitamin supplementation including vitamin A, D, E, and K.
- » Exercise recommendations by physical and occupational therapists.
- » Gene therapy may be a potential treatment option in the future.