MTTP gene

microsomal triglyceride transfer protein

Normal Function

The *MTTP* gene provides instructions for making a protein called microsomal triglyceride transfer protein. This protein helps produce beta-lipoproteins, which are molecules that are made up of proteins (including one called apolipoprotein B), cholesterol, and particular types of fats called phospholipids and triglycerides.

Different types of beta-lipoproteins are made in the intestine and liver. In the intestine, beta-lipoproteins include chylomicrons, which are formed as food is digested after a meal to carry dietary fats and cholesterol from the intestine to the bloodstream. Chylomicrons are also necessary for the absorption of certain fat-soluble vitamins, such as vitamins E, A, and K. In the liver, beta-lipoproteins include low-density lipoproteins (LDL) and very low-density lipoproteins (VLDL). These lipoproteins transport fats, cholesterol, and fat-soluble vitamins in the bloodstream to tissues throughout the body. Sufficient levels of fats, cholesterol, and vitamins are necessary for normal growth, development, and maintenance of the body's cells and tissues.

Health Conditions Related to Genetic Changes

Abetalipoproteinemia

More than 60 mutations in the *MTTP* gene have been found to cause abetalipoproteinemia. This condition impairs the normal absorption of fats and fat-soluble vitamins from the diet and primarily affects the gastrointestinal system, eyes, nervous system, and blood.

Most *MTTP* gene mutations lead to the production of microsomal triglyceride transfer protein with reduced or absent function, preventing the formation of beta-lipoproteins. One particular mutation is common in affected individuals of Ashkenazi (eastern and central European) Jewish descent; this mutation replaces the protein building block (amino acid) glycine with a premature stop signal at position 865 (written as Gly865Ter or G865X) in the instructions used to make the microsomal triglyceride transfer protein. As a result of this change, an abnormally small, nonfunctional version of the protein is made. All *MTTP* gene mutations that cause abetalipoproteinemia impair beta-lipoprotein formation and result in a severe shortage of chylomicrons, LDLs, and VLDLs. A lack of these lipoproteins prevents dietary fats and fat-soluble vitamins from being absorbed

from the diet and transported through the bloodstream to the body's tissues. These nutritional deficiencies lead to health problems in people with abetalipoproteinemia.

Other Names for This Gene

- ABL
- microsomal TG transfer protein
- microsomal triglyceride transfer protein (large polypeptide, 88kD)
- microsomal triglyceride transfer protein large subunit
- MTP
- MTP triglyceride carrier
- MTP_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MTTP (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4547[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28MTTP%5BTIAB%5D%29+OR+%28microsomal+triglyceride+transfer+protein%5BTIAB%5D%29%29+AND+%28Genes%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- MICROSOMAL TRIGLYCERIDE TRANSFER PROTEIN; MTTP (https://omim.org/entry/157147)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/4547)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=MTTP[gene])

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Genomic Location

The *MTTP* gene is found on chromosome 4 (https://medlineplus.gov/genetics/chromosome/4/).

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