Tangier disease

Description

Tangier disease is an inherited disorder characterized by significantly reduced levels of high-density lipoprotein (HDL) in the blood. HDL transports cholesterol and certain fats called phospholipids from the body's tissues to the liver, where they are removed from the blood. HDL is often referred to as "good cholesterol" because high levels of this substance reduce the chances of developing heart and blood vessel (cardiovascular) disease. Because people with Tangier disease have very low levels of HDL, they have a moderately increased risk of cardiovascular disease.

Additional signs and symptoms of Tangier disease include a slightly elevated amount of fat in the blood (mild hypertriglyceridemia); disturbances in nerve function (neuropathy); and enlarged, orange-colored tonsils. Affected individuals often develop atherosclerosis, which is an accumulation of fatty deposits and scar-like tissue in the lining of the arteries. Other features of this condition may include an enlarged spleen (splenomegaly), an enlarged liver (hepatomegaly), clouding of the outermost layer of the eye (corneal clouding), and type 2 diabetes.

Frequency

Tangier disease is a rare disorder, with approximately 100 cases identified worldwide. More cases are likely undiagnosed. This condition is named after an island off the coast of Virginia where the first affected individuals were identified.

Causes

Variants (also called mutations) in the *ABCA1* gene cause Tangier disease. This gene provides instructions for making a protein that releases cholesterol and phospholipids from cells. These substances are used to make HDL. HDL then transports cholesterol and phospholipids to the liver.

Variants in the *ABCA1* gene prevent the release of cholesterol and phospholipids from cells. As a result, these substances accumulate within cells, causing certain body tissues to enlarge and the tonsils to acquire a yellowish-orange color. A buildup of cholesterol can be toxic to cells, leading to impaired cell function or cell death. In addition, the inability to transport cholesterol and phospholipids out of cells results in very low HDL levels, which increases a person's risk of cardiovascular disease. All

these factors cause the signs and symptoms of Tangier disease.

Learn more about the gene associated with Tangier disease

- ABCA1

Inheritance

Tangier disease is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell must have a variant to cause the disorder. The parents of an individual with an autosomal recessive condition each carry one copy of the altered gene, but they typically do not show signs and symptoms of the condition; however, they may have a related condition called familial HDL deficiency.

Other Names for This Condition

- A-alphalipoprotein neuropathy
- Alpha high density lipoprotein deficiency disease
- Analphalipoproteinemia
- Cholesterol thesaurismosis
- Familial high density lipoprotein deficiency disease
- Familial hypoalphalipoproteinemia
- HDL lipoprotein deficiency disease
- Lipoprotein deficiency disease, HDL, familial
- Tangier disease neuropathy
- Tangier hereditary neuropathy

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Tangier disease (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0039292/)

Genetic and Rare Diseases Information Center

- Tangier disease (https://rarediseases.info.nih.gov/diseases/7731/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Tangier disease%22)

Catalog of Genes and Diseases from OMIM

- TANGIER DISEASE; TGD (https://omim.org/entry/205400)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28Tangier+Disease%5BMAJR%5D%29+AND+%28Tangier+disease%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

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