ABCA1 gene

ATP binding cassette subfamily A member 1

Normal Function

The *ABCA1* gene belongs to a group of genes called the ATP-binding cassette family. These genes provide instructions for making proteins that transport molecules across cell membranes. The ABCA1 protein is produced in many tissues, but high levels of this protein are found in the liver and in immune cells called macrophages.

The ABCA1 protein helps move cholesterol and certain fats called phospholipids across the cell membrane to the outside of the cell. These substances are then picked up by a protein called apolipoprotein A-I (apoA-I), which is produced from the *APOA1* gene. ApoA-I, cholesterol, and phospholipids combine to make high-density lipoprotein (HDL), often referred to as "good cholesterol" because high levels of this substance reduce the chances of developing heart and blood vessel (cardiovascular) disease.

HDL carries cholesterol and phospholipids through the bloodstream from the body's tissues to the liver. Once in the liver, cholesterol and phospholipids are redistributed to other tissues or removed from the body. The process of removing excess cholesterol from cells is extremely important for balancing cholesterol levels and maintaining cardiovascular health.

Health Conditions Related to Genetic Changes

Familial HDL deficiency

Variants (also called mutations) in the *ABCA1* gene can cause a condition called familial HDL deficiency. People with this condition have reduced levels of HDL in their blood and may experience early-onset cardiovascular disease, often before age 50. While one copy of the altered *ABCA1* gene causes familial HDL deficiency, two copies of the altered gene cause a more severe disorder called Tangier disease (described below).

Most *ABCA1* gene variants that cause familial HDL deficiency change single protein building blocks (amino acids) in the ABCA1 protein. Other variants alter the protein in other ways. The changes in the ABCA1 protein prevent the release of cholesterol and phospholipids from cells, decreasing the amount of these substances available to form HDL. As a result, the levels of HDL in the blood are low. A shortage (deficiency) of HDL is believed to increase a person's risk of cardiovascular disease.

Tangier disease

Several variants in the *ABCA1* gene have been found to cause Tangier disease. Most of these variants change single amino acids in the ABCA1 protein. These variants 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 may increase the risk of cardiovascular disease. All these factors cause the signs and symptoms of Tangier disease.

Other Names for This Gene

- ABC1
- ABCA1_HUMAN
- ATP binding cassette transporter 1
- ATP-binding cassette 1
- ATP-binding cassette, sub-family A (ABC1), member 1
- CERP
- cholesterol efflux regulatory protein
- FLJ14958
- HDLDT1
- high density lipoprotein deficiency, Tangier type, 1
- TGD

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ABCA1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=19[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28ABCA1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ATP-BINDING CASSETTE, SUBFAMILY A, MEMBER 1; ABCA1 (https://omim.org/entry/600046)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/19)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ABCA1[gene])

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Genomic Location

The *ABCA1* gene is found on chromosome 9 (https://medlineplus.gov/genetics/chromosome/9/).

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