AGPAT2 gene

1-acylglycerol-3-phosphate O-acyltransferase 2

Normal Function

The *AGPAT2* gene provides instructions for making an enzyme that is found in many of the body's cells and tissues. It plays a critical role in the growth and development of adipocytes, which are cells that store fats for energy. Adipocytes are the major component of the body's fatty (adipose) tissue.

The AGPAT2 enzyme is part of a chemical pathway in many cells that produces two important types of fats (lipids): glycerophospholipids and triacylglycerols. Glycerophospholipids are the major component of cell membranes and are involved in chemical signaling within cells. Triacylglycerols (also known as triglycerides) are fat molecules that are stored in adipocytes for later conversion to energy.

The AGPAT2 enzyme is responsible for a particular chemical reaction in the production of these two types of lipids. Specifically, the enzyme helps convert a molecule called lysophosphatidic acid (LPA) to another molecule, phosphatidic acid (PA). Additional reactions convert phosphatidic acid to glycerophospholipids and triacylglycerols.

Health Conditions Related to Genetic Changes

Congenital generalized lipodystrophy

At least 26 mutations in the *AGPAT2* gene have been identified in people with congenital generalized lipodystrophy (also called Berardinelli-Seip congenital lipodystrophy) type 1. This rare condition is characterized by an almost total absence of adipose tissue and a very muscular appearance. A shortage of adipose tissue leads to multiple health problems, including high levels of triglycerides circulating in the bloodstream (hypertriglyceridemia) and diabetes mellitus.

The *AGPAT2* gene mutations that cause congenital generalized lipodystrophy type 1 greatly reduce or eliminate the activity of the AGPAT2 enzyme. Studies suggest that a loss of this enzyme's activity reduces the production and storage of triacylglycerols in adipocytes, which prevents these cells from storing fats. A lack of enzyme activity may also reduce the levels of glycerophospholipids in adipocytes, which changes the structure of the cell membrane and disrupts normal signaling within these cells. All of these abnormalities prevent the body from storing fats normally in adipose tissue. The

resulting lack of body fat underlies the varied signs and symptoms of congenital generalized lipodystrophy type 1.

Other Names for This Gene

- 1-acyl-sn-glycerol-3-phosphate acyltransferase beta
- 1-acylglycerol-3-phosphate O-acyltransferase 2 (lysophosphatidic acid acyltransferase, beta)
- 1-AGP acyltransferase 2
- 1-AGPAT2
- BSCL1
- LPAAB
- LPAAT-beta
- lysophosphatidic acid acyltransferase-beta
- PLCB_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of AGPAT2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=10555[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28AGPAT2%5BTIAB%5D%29+OR+%281-acylglycerol-3-phosphate+O-acyltransferase+2%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- 1-ACYLGLYCEROL-3-PHOSPHATE O-ACYLTRANSFERASE 2; AGPAT2 (https://omim.org/entry/603100)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/10555)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=AGPAT2[gene])

References

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Genomic Location

The *AGPAT2* gene is found on chromosome 9 (https://medlineplus.gov/genetics/chromosome/9/).

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