



## 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

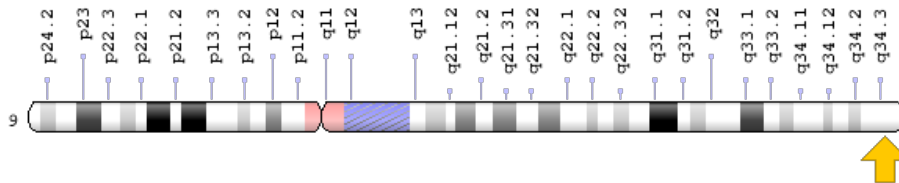
At least 26 mutations in the *AGPAT2* gene have been identified in people with congenital generalized lipodystrophy (also called Berardinelli-Seip congenital lipodystrophy) type 1. This rare condition is characterized by an almost total absence of adipose tissue and a very muscular appearance. A shortage 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.

## Chromosomal Location

Cytogenetic Location: 9q34.3, which is the long (q) arm of chromosome 9 at position 34.3

Molecular Location: base pairs 136,673,143 to 136,687,457 on chromosome 9 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

## 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

### Educational Resources

- Biochemistry (fifth edition, 2002): Phosphatidate Is a Common Intermediate in the Synthesis of Phospholipids and Triacylglycerols  
<https://www.ncbi.nlm.nih.gov/books/NBK22405/>

### Clinical Information from GeneReviews

- Berardinelli-Seip Congenital Lipodystrophy  
<https://www.ncbi.nlm.nih.gov/books/NBK1212>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?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  
<http://omim.org/entry/603100>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_AGPAT2.html](http://atlasgeneticsoncology.org/Genes/GC_AGPAT2.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=AGPAT2%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report#!/hgnc\\_id/HGNC:325](https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:325)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:10555>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/10555>
- UniProt  
<https://www.uniprot.org/uniprot/O15120>

### **Sources for This Summary**

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<https://ghr.nlm.nih.gov/gene/AGPAT2>

Reviewed: January 2016

Published: November 12, 2019

Lister Hill National Center for Biomedical Communications  
U.S. National Library of Medicine  
National Institutes of Health  
Department of Health & Human Services