

PNPLA2 gene

patatin like phospholipase domain containing 2

Normal Function

The *PNPLA2* gene provides instructions for making an enzyme called adipose triglyceride lipase (ATGL). The ATGL enzyme plays a role in breaking down fats called triglycerides, which are the main source of stored energy in cells. Triglycerides are the major component of cell structures called lipid droplets (also called adiposomes). The ATGL enzyme is found on the surface of lipid droplets. When activated, the ATGL enzyme breaks down triglycerides to provide energy for the body.

Health Conditions Related to Genetic Changes

Neutral lipid storage disease with myopathy

At least five mutations in the *PNPLA2* gene have been found to cause neutral lipid storage disease with myopathy. Some of these mutations cause the enzyme to function abnormally. Other mutations prevent the enzyme from ever reaching lipid droplets, so it is unable to interact with triglycerides. Any disruption in the breakdown of triglycerides leads to the accumulation of these fats in muscle and other tissues throughout the body, resulting in the signs and symptoms of neutral lipid storage disease with myopathy.

Other Names for This Gene

- adipose triglyceride lipase
- ATGL
- desnutrin
- FP17548
- patatin-like phospholipase domain containing 2
- PLPL2_HUMAN
- transport-secretion protein 2.2
- triglyceride hydrolase
- TTS-2.2
- TTS2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PNPLA2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=57104\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=57104[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28PNPLA2%5BTIAB%5D%29+OR+%28ATGL%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D%29>)

Catalog of Genes and Diseases from OMIM

- PATATIN-LIKE PHOSPHOLIPASE DOMAIN-CONTAINING PROTEIN 2; PNPLA2 (<https://omim.org/entry/609059>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/57104>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=PNPLA2\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=PNPLA2[gene]))

References

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Genomic Location

The *PNPLA2* gene is found on chromosome 11 (<https://medlineplus.gov/genetics/chromosome/11/>).

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