ABHD5 gene

abhydrolase domain containing 5, lysophosphatidic acid acyltransferase

Normal Function

The *ABHD5* gene provides instructions for making a protein that turns on (activates) an enzyme called adipose triglyceride lipase (ATGL). The ATGL enzyme plays a role in breaking down fats called triglycerides, which are a major source of stored energy in cells. Cells primarily store triglycerides in structures called lipid droplets (also called adiposomes). The ABHD5 protein and the ATGL enzyme are found on the surface of lipid droplets. Once activated, the ATGL enzyme breaks down triglycerides in these structures to provide energy for the body.

Health Conditions Related to Genetic Changes

Chanarin-Dorfman syndrome

Several variants (also called mutations) in the *ABHD5* gene have been found to cause Chanarin-Dorfman syndrome, a condition in which triglycerides build up in the body. These variants impair the ABHD5 protein's ability to activate the ATGL enzyme. If there is not enough activated ATGL enzyme to break down triglycerides, these fats can accumulate in various organs and tissues throughout the body. Over time, the buildup of triglycerides can damage cells and tissues, leading to the signs and symptoms of Chanarin-Dorfman syndrome.

Other Names for This Gene

- CGI58
- comparative gene identification 58
- IECN2
- NCIE2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ABHD5 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=51099[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28ABHD5%5BTIAB%5D%29+OR+%28%28CGI58%5BTIAB%5D%29+OR+%28CGI-58%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ABHYDROLASE DOMAIN-CONTAINING PROTEIN 5, LYSOPHOSPHATIDIC ACID ACYLTRANSFERASE; ABHD5 (https://omim.org/entry/604780)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/51099)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ABHD5[gene])

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Rastelli E, Santorelli FM, Tasca E, Tavian D, Toscano A, Angelini C; Italian NLSDGroup. Neutral Lipid Storage Diseases: clinical/genetic features and naturalhistory in a large cohort of Italian patients. Orphanet J Rare Dis. 2017 May12; 12(1):90. doi: 10.1186/s13023-017-0646-9. Citation on PubMed (https://www.ncbi.nlm.nih.gov/pubmed/28499397)

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Genomic Location

The *ABHD5* gene is found on chromosome 3 (https://medlineplus.gov/genetics/chromosome/3/).

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