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 the main source of stored energy in cells. Triglycerides are the major component of cell 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

At least 20 mutations in the *ABHD5* gene have been found to cause Chanarin-Dorfman syndrome. These mutations impair the ABHD5 protein's ability to activate the ATGL enzyme. Without an active ATGL enzyme, triglycerides within lipid droplets cannot be broken down. As a result, these fats accumulate in various organs and tissues throughout the body, causing the signs and symptoms of Chanarin-Dorfman syndrome.

Other Names for This Gene

- ABHD5_HUMAN
- CDS
- CGI-58
- CGI58
- CGI58 protein
- IECN2
- MGC8731
- 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])

References

- Lass A, Zimmermann R, Haemmerle G, Riederer M, Schoiswohl G, Schweiger M, Kienesberger P, Strauss JG, Gorkiewicz G, Zechner R. Adipose triglyceridelipase-mediated lipolysis of cellular fat stores is activated by CGI-58 anddefective in Chanarin-Dorfman Syndrome. Cell Metab. 2006 May;3(5):309-19. doi:10.1016/j.cmet.2006.03.005. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16679289)
- Lefevre C, Jobard F, Caux F, Bouadjar B, Karaduman A, Heilig R, Lakhdar H, Wollenberg A, Verret JL, Weissenbach J, Ozguc M, Lathrop M, Prud'homme JF,Fischer J. Mutations in CGI-58, the gene encoding a new protein of theesterase/lipase/thioesterase subfamily, in Chanarin-Dorfman syndrome. Am J HumGenet. 2001 Nov;69(5):1002-12. doi: 10.1086/324121. Epub 2001 Oct 2. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/11590543) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1274347/)
- Yen CL, Farese RV Jr. Fat breakdown: a function for CGI-58 (ABHD5) provides anew piece of the puzzle. Cell Metab. 2006 May;3(5):305-7. doi:10.1016/j.cmet.2006.04.001. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16679288)

Genomic Location

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

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