ABHD5 gene
abhydrolase domain containing 5

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.

Chromosomal Location

Cytogenetic Location: 3p21.33, which is the short (p) arm of chromosome 3 at position 21.33

Molecular Location: base pairs 43,690,870 to 43,734,371 on chromosome 3 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)
Other Names for This Gene
• ABHD5_HUMAN
• CDS
• CGI-58
• CGI58
• CGI58 protein
• IECN2
• MGC8731
• NCIE2

Additional Information & Resources

Scientific Articles on PubMed
• PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?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+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 5
  http://omim.org/entry/604780

Research Resources
• Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_ABHD5.html
• ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ABHD5%5Bgene%5D
• HGNC Gene Family: Abhydrolase domain containing
  https://www.genenames.org/cgi-bin/genefamilies/set/4
• HGNC Gene Symbol Report
  https://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=21396
• Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:51099
Sources for This Summary

- OMIM: ABHYDROLASE DOMAIN-CONTAINING 5
  http://omim.org/entry/604780

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16679289

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11590543
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1274347/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16679288

Reprinted from Genetics Home Reference:

Reviewed: November 2008
Published: August 28, 2018

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