

FUCA1 gene

alpha-L-fucosidase 1

Normal Function

The *FUCA1* gene provides instructions for making an enzyme called alpha-L-fucosidase. This enzyme is found in lysosomes, which are compartments in the cell that digest and recycle different types of molecules. Within lysosomes, this enzyme plays a role in the breakdown of sugar molecules (oligosaccharides) that are attached to certain fats (glycolipids) and proteins (glycoproteins). Alpha-L-fucosidase is responsible for cutting off (cleaving) a sugar molecule called fucose toward the end of the breakdown process.

Health Conditions Related to Genetic Changes

Fucosidosis

Many variants (also called mutations) in the *FUCA1* gene have been found to cause fucosidosis. This condition can cause severe health problems and early death in affected individuals. Most of the *FUCA1* variants either shorten the alpha-L-fucosidase enzyme or change a single protein building block (amino acid) in the enzyme. These changes mean that the enzyme cannot break down glycolipids and glycoproteins properly. These compounds then accumulate in the lysosomes and cause cells throughout the body to malfunction. Brain cells are particularly sensitive to the buildup of glycolipids and glycoproteins. Damage to brain cells is thought to cause the neurological symptoms of fucosidosis. Glycolipids and glycoproteins also accumulate in organs such as the liver, spleen, and skin. This contributes to the other symptoms of fucosidosis.

Other Names for This Gene

- a-L-fucosidase 1
- alpha-L-fucosidase 1
- FUCA
- fucosidase, alpha-L, tissue
- fucosidase, alpha-L-1, tissue
- tissue fucosidase
- α -L-fucosidase 1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of FUCA1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2517\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2517[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28FUCA1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D%29>)

Catalog of Genes and Diseases from OMIM

- FUCOSIDASE, ALPHA-L, 1; FUCA1 (<https://omim.org/entry/612280>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/2517>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=FUCA1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=FUCA1[gene]))

References

- Cragg H, Williamson M, Young E, O'Brien J, Alhadeff J, Fang-Kircher S, Paschke E, Winchester B. Fucosidosis: genetic and biochemical analysis of eight cases. *J Med Genet*. 1997 Feb;34(2):105-10. doi: 10.1136/jmg.34.2.105. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/9039984>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1050861/>)
- Puente-Ruiz N, Ellis I, Bregu M, Chen C, Church HJ, Tylee KL, Gladston S, Hackett R, Oldham A, Virk S, Hendriksz C, Morris AAM, Jones SA, Stepień KM. Long-term outcomes in two adult siblings with Fucosidosis - Diagnostic odyssey and clinical manifestations. *Mol Genet Metab Rep*. 2023 Sep 27;37:101009. doi:10.1016/j.ymgmr.2023.101009. eCollection 2023 Dec. Citation on PubMed (<https://www.ncbi.nlm.nih.gov/pubmed/38053939>)
- Stepień KM, Ciara E, Jezela-Stanek A. Fucosidosis-Clinical Manifestation, Long-Term Outcomes, and Genetic Profile-Review and Case Series. *Genes (Basel)*. 2020 Nov 22;11(11):1383. doi: 10.3390/genes11111383. Citation on PubMed (<https://www.ncbi.nlm.nih.gov/pubmed/33266441>)
- Willems PJ, Gatti R, Darby JK, Romeo G, Durand P, Dumon JE, O'Brien JS. Fucosidosis revisited: a review of 77 patients. *Am J Med Genet*. 1991 Jan;38(1):111-31. doi: 10.1002/ajmg.1320380125. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/2012122>)

- Willems PJ, Seo HC, Coucke P, Tonlorenzi R, O'Brien JS. Spectrum of mutations in fucosidosis. Eur J Hum Genet. 1999 Jan;7(1):60-7. doi:10.1038/sj.ejhg.5200272. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/10094192>)

Genomic Location

The *FUCA1* gene is found on chromosome 1 (<https://medlineplus.gov/genetics/chromosome/1/>).

Last updated March 14, 2024