AGL gene

amylo-alpha-1, 6-glucosidase, 4-alpha-glucanotransferase

Normal Function

The *AGL* gene provides instructions for making the glycogen debranching enzyme. This enzyme is involved in the breakdown of a complex sugar called glycogen, which is a major source of stored energy in the body. Glycogen is made up of several molecules of a simple sugar called glucose. Some glucose molecules are linked together in a straight line, while others branch off and form side chains. The glycogen debranching enzyme is involved in the breakdown of these side chains. The branched structure of glycogen makes it more compact for storage and allows it to break down more easily when it is needed for fuel.

The *AGL* gene provides instructions for making several different versions (isoforms) of the glycogen debranching enzyme. These isoforms vary by size and are active (expressed) in different tissues.

Health Conditions Related to Genetic Changes

Glycogen storage disease type III

Approximately 100 mutations in the *AGL* gene have been found to cause glycogen storage disease type III (also called GSDIII or Cori disease). Most of these mutations lead to a premature stop signal in the instructions for making the glycogen debranching enzyme, resulting in a nonfunctional enzyme. As a result, the side chains of glycogen molecules cannot be removed and abnormal, partially broken down glycogen molecules are stored within cells. A buildup of abnormal glycogen damages organs and tissues throughout the body, particularly the liver and muscles, leading to the signs and symptoms of GSDIII.

Mutations in the *AGL* gene can affect different isoforms of the enzyme, depending on where the mutations are located in the gene. For example, mutations that occur in a part of the *AGL* gene called exon 3 affect the isoform that is primarily expressed in the liver. These mutations almost always lead to GSD type IIIb, which is characterized by liver problems.

Other Names for This Gene

- amylo-1, 6-glucosidase, 4-alpha-glucanotransferase
- GDE
- GDE_HUMAN
- glycogen debrancher
- glycogen debranching enzyme

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of AGL (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=178[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28AGL%5BTIAB%5D%29+OR+%28glycogen+debranching+enzyme%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+2880+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- AMYLO-1,6-GLUCOSIDASE, 4-ALPHA-GLUCANOTRANSFERASE; AGL (https://omim.org/entry/610860)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/178)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=AGL[gene])

References

- Cheng A, Zhang M, Okubo M, Omichi K, Saltiel AR. Distinct mutations in theglycogen debranching enzyme found in glycogen storage disease type III lead toimpairment in diverse cellular functions. Hum Mol Genet. 2009 Jun1;18(11):2045-52. doi: 10.1093/hmg/ddp128. Epub 2009 Mar 19. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/19299494) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2678930/)
- Endo Y, Horinishi A, Vorgerd M, Aoyama Y, Ebara T, Murase T, Odawara M, Podskarbi T, Shin YS, Okubo M. Molecular analysis of the AGL gene: heterogeneityof mutations in patients with glycogen storage disease type III from

Germany,Canada, Afghanistan, Iran, and Turkey. J Hum Genet. 2006;51(11):958-963. doi:10.1007/s10038-006-0045-x. Epub 2006 Sep 19. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17047887)

- Frisbie JH, O'Connell DJ, Tow DE, Sasahara AA, Belko JS. Autologousradioiodinated fibrinogen, simplified. J Nucl Med. 1975 May;16(5):393-401. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/1194993)
- Lucchiari S, Pagliarani S, Salani S, Filocamo M, Di Rocco M, Melis D, RodolicoC, Musumeci O, Toscano A, Bresolin N, Comi GP. Hepatic and neuromuscular forms ofglycogenosis type III: nine mutations in AGL. Hum Mutat. 2006 Jun;27(6):600-1.doi: 10.1002/humu.9426. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16705713)

Genomic Location

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

Last updated September 1, 2010