

AGL gene

amylo-alpha-1,6-glucosidase and 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 variants (also called mutations) in the *AGL* gene have been found to cause glycogen storage disease type III (also called GSDIII or Cori disease). Most of these variants 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.

Variants in the *AGL* gene can affect different isoforms of the enzyme, depending on where the variants are located in the gene. For example, variants that occur in a part of the *AGL* gene called exon 3 affect the isoform that is primarily expressed in the liver. These variants 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\[geneid\]](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%29>)

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\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=AGL[gene]))

References

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Genomic Location

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

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