

GAA gene

alpha glucosidase

Normal Function

The *GAA* gene provides instructions for producing an enzyme called acid alpha-glucosidase (also known as acid maltase). This enzyme is active in lysosomes, which are structures that serve as recycling centers within cells. Lysosomes use digestive enzymes to break down complex molecules into simpler ones that can be used by cells. Acid alpha-glucosidase normally breaks down a complex sugar called glycogen into a simpler sugar called glucose. Glucose is the main energy source for most cells.

Health Conditions Related to Genetic Changes

Pompe disease

More than 200 mutations in the *GAA* gene have been identified in people with Pompe disease. Many of these mutations change one of the protein building blocks (amino acids) used to make acid alpha-glucosidase. Other mutations insert or delete genetic material in the *GAA* gene. Mutations in this gene significantly reduce the activity of acid alpha-glucosidase, preventing the enzyme from breaking down glycogen effectively. As a result, this complex sugar can build up to toxic levels in lysosomes. The abnormal buildup of glycogen damages organs and tissues throughout the body, particularly the muscles, leading to progressive muscle weakness, heart problems, and the other features of Pompe disease.

Other Names for This Gene

- acid alpha-glucosidase
- acid alpha-glucosidase preproprotein
- acid maltase
- Aglucosidase alfa
- Alpha-1,4-glucosidase
- Amyloglucosidase
- Glucoamylase
- glucosidase, alpha; acid
- glucosidase, alpha; acid (Pompe disease, glycogen storage disease type II)

- LYAG
- LYAG_HUMAN
- lysosomal alpha-glucosidase

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of GAA ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2548\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2548[geneid]))

Scientific Articles on PubMed

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

Catalog of Genes and Diseases from OMIM

- GLUCOSIDASE, ALPHA, ACID; GAA (<https://omim.org/entry/606800>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/2548>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=GAA\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=GAA[gene]))

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Genomic Location

The GAA gene is found on chromosome 17 (<https://medlineplus.gov/genetics/chromosome/17/>).

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