GLA gene

galactosidase alpha

Normal Function

The *GLA* gene provides instructions for making an enzyme called alpha-galactosidase A. This enzyme is active in lysosomes, which are structures that act as recycling centers within cells. Lysosomes use digestive enzymes to process worn-out cell components and recycle usable parts.

Alpha-galactosidase A breaks down a molecule called globotriaosylceramide, which consists of three sugars attached to a fatty substance. This molecule is degraded as part of the normal recycling of old red blood cells (erythrocytes) and other types of cells.

Health Conditions Related to Genetic Changes

Fabry disease

More than 370 mutations in the *GLA* gene have been identified in people with Fabry disease. Most of these genetic changes are unique to single families. The most common type of mutation changes a single protein building block (amino acid) in alpha-galactosidase A. Other mutations delete part of the *GLA* gene, insert extra genetic material into the gene, or insert a premature stop signal in the gene's instructions for making alpha-galactosidase A. Alterations in the *GLA* gene produce an abnormal version of the enzyme that is unable to break down globotriaosylceramide effectively. As a result, this substance builds up in the body's cells, particularly cells lining blood vessels in the skin and cells in the kidneys, heart, and nervous system. The progressive accumulation of globotriaosylceramide damages these cells, leading to the varied signs and symptoms of Fabry disease.

Mutations that eliminate the activity of the alpha-galactosidase A enzyme lead to the severe, classic form of Fabry disease, which typically begins in childhood. Mutations that reduce but do not completely eliminate the enzyme's activity usually cause milder, late-onset forms of the disorder.

Other Names for This Gene

- AGAL_HUMAN
- Agalsidase alfa

- Alpha-D-galactosidase A
- alpha-D-galactoside galactohydrolase
- Alpha-galactosidase
- alpha-Galactosidase A
- ceramidetrihexosidase
- GALA
- galactosidase, alpha
- Melibiase

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of GLA (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2717[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28Agalsidase+alfa%5BTIAB%5D%29+OR+%28Alpha-D-galactosidase+A%5BTIAB%5D%29+OR+%28alpha-D-galactoside+galactohydrolase%5BTIAB%5D%29+OR+%28alpha-Galactosidase%5BTIAB%5D%29+OR+%28Melibiase%5BTIAB%5D%29+OR+%28ceramidetrihexosidase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- GALACTOSIDASE, ALPHA; GLA (https://omim.org/entry/300644)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/2717)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=GLA[gene])

References

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Genomic Location

The *GLA* gene is found on the X chromosome (https://medlineplus.gov/genetics/chromosome/x/).

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