GYS2 gene

glycogen synthase 2

Normal Function

The *GYS2* gene provides instructions for making an enzyme called liver glycogen synthase. Liver glycogen synthase is produced solely in liver cells, where it helps form the complex sugar glycogen by linking together molecules of the simple sugar glucose. Glucose that is taken in from food is stored in the body as glycogen, which is a major source of energy. Glycogen that is stored in the liver can be broken down rapidly when glucose is needed to maintain normal blood glucose levels between meals.

Health Conditions Related to Genetic Changes

Glycogen storage disease type 0

Approximately 20 mutations in the *GYS2* gene have been found to cause a form of glycogen storage disease type 0 (GSD 0) that affects the liver. Most *GYS2* gene mutations that cause this condition lead to a lack of functional glycogen synthase, resulting in a complete absence of glycogen in liver cells. Normally, glycogen is formed from the leftover glucose that is not immediately used by cells after glucose is consumed during meals. In people with GSD 0, who cannot form glycogen, the extra glucose is released by the body. As a result, people with this condition have no stored energy during long periods without food (fasting). During these periods, affected individuals may develop low blood glucose (hypoglycemia) and nausea as well as other signs and symptoms of GSD 0.

Other Names for This Gene

- glycogen [starch] synthase, liver
- glycogen synthase 2 (liver)
- GYS2_HUMAN
- liver glycogen synthase
- liver glycogen synthase 2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of GYS2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2998[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28GYS2%5BTIAB%5D%29+OR+%28glycogen+synthase+2%5BTIAB%5D%29%29+OR+%28%28liver+glycogen+synthase%5BTIAB%5D%29+OR+%28glycogen+synthase+2%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+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- GLYCOGEN SYNTHASE 2; GYS2 (https://omim.org/entry/138571)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/2998)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=GYS2[gene])

References

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- Orho M, Bosshard NU, Buist NR, Gitzelmann R, Aynsley-Green A, Blumel P, GannonMC, Nuttall FQ, Groop LC. Mutations in the liver glycogen synthase gene inchildren with hypoglycemia due to glycogen storage disease type 0. J Clin Invest. 1998 Aug 1;102(3):507-15. doi: 10.1172/JCI2890. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/9691087) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC508911/)
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- Weinstein DA, Correia CE, Saunders AC, Wolfsdorf JI. Hepatic glycogen synthasedeficiency: an infrequently recognized cause of ketotic hypoglycemia. Mol

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Genomic Location

The *GYS2* gene is found on chromosome 12 (https://medlineplus.gov/genetics/chromosome/12/).

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