

GYS1 gene

glycogen synthase 1

Normal Function

The *GYS1* gene provides instructions for making an enzyme called muscle glycogen synthase. Muscle glycogen synthase is produced in most cells but is most abundant in heart (cardiac) muscle and muscles used for movement (skeletal muscles). Muscle glycogen synthase helps link together molecules of the simple sugar glucose to form the complex sugar glycogen, which is a major source of stored energy in the body. Most glucose that is taken in from food is stored as glycogen in muscle cells. During contractions of the cardiac muscle or rapid or sustained movement of skeletal muscle, glycogen stored in muscle cells is broken down to supply the cells with energy.

Health Conditions Related to Genetic Changes

Glycogen storage disease type 0

At least four mutations in the *GYS1* gene have been found to cause a form of glycogen storage disease type 0 (GSD 0) that affects cardiac and skeletal muscle. Most *GYS1* gene mutations that cause this condition lead to a lack of functional muscle glycogen synthase, resulting in a complete absence of glycogen in muscle 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 sugar is released by the body. As a result, people with muscle GSD 0 do not have any stored energy, which leads to muscle pain, weakness, or episodes of fainting following moderate physical activity. Since there is no glycogen in cardiac muscle, affected individuals are also at an increased risk of cardiac arrest and sudden death, particularly after physical activity.

Other Names for This Gene

- glycogen [starch] synthase, muscle
- glycogen synthase 1 (muscle)
- GSY
- GYS
- *GYS1_HUMAN*
- muscle glycogen synthase

- muscle glycogen synthase 1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of GYS1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2997\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2997[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28GYS1%5BTIAB%5D%29+OR+%28glycogen+synthase+1%5BTIAB%5D%29%29+OR+%28%28muscle+glycogen+synthase%5BTIAB%5D%29+OR+%28glycogen+synthase+1%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%29%29>)

Catalog of Genes and Diseases from OMIM

- GLYCOGEN SYNTHASE 1; GYS1 (<https://omim.org/entry/138570>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/2997>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=GYS1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=GYS1[gene]))

References

- Cameron JM, Levandovskiy V, MacKay N, Utgikar R, Ackerley C, Chiasson D, Halliday W, Raiman J, Robinson BH. Identification of a novel mutation in GYS1(muscle-specific glycogen synthase) resulting in sudden cardiac death, that isdiagnosable from skin fibroblasts. *Mol Genet Metab.* 2009 Dec;98(4):378-82. doi: 10.1016/j.ymgme.2009.07.012. Epub 2009 Jul 26. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19699667>)
- Fredriksson J, Anevski D, Almgren P, Sjogren M, Lyssenko V, Carlson J, IsomaaB, Taskinen MR, Groop L, Orho-Melander M; Botnia Study Group. Variation in GYS1interacts with exercise and gender to predict cardiovascular mortality. *PLoS One.*2007 Mar 14;2(3):e285. doi: 10.1371/journal.pone.0000285. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/17356695>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1805686/>)
- Kollberg G, Tulinius M, Gilljam T, Ostman-Smith I, Forsander G, Jotorp P, Oldfors A, Holme E. Cardiomyopathy and exercise intolerance in muscle glycogenstorage disease 0. *N Engl J Med.* 2007 Oct 11;357(15):1507-14. doi:10.1056/

NEJMoa066691. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/17928598>)

- Orho M, Nikula-Ijas P, Schalin-Jantti C, Permutt MA, Groop LC. Isolation and characterization of the human muscle glycogen synthase gene. *Diabetes*. 1995 Sep;44(9):1099-105. doi: 10.2337/diab.44.9.1099. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/7657035>)
- Sukigara S, Liang WC, Komaki H, Fukuda T, Miyamoto T, Saito T, Saito Y, Nakagawa E, Sugai K, Hayashi YK, Sugie H, Sasaki M, Nishino I. Muscle glycogen storage disease 0 presenting recurrent syncope with weakness and myalgia. *Neuromuscul Disord*. 2012 Feb;22(2):162-5. doi: 10.1016/j.nmd.2011.08.008. Epub 2011 Sep 29. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/21958591>)

Genomic Location

The *GYS1* gene is found on chromosome 19 (<https://medlineplus.gov/genetics/chromosome/19/>).

Last updated January 1, 2014