PYGL gene

glycogen phosphorylase L

Normal Function

The *PYGL* gene provides instructions for making an enzyme called liver glycogen phosphorylase. This enzyme breaks down a complex sugar called glycogen. Liver glycogen phosphorylase is one of three related enzymes that break down glycogen in cells; the other glycogen phosphorylases are found in the brain and in muscles. Liver glycogen phosphorylase is found only in liver cells, where it breaks down glycogen into a type of sugar called glucose-1-phosphate. Additional steps convert glucose-1-phosphate into glucose, a simple sugar that is the main energy source for most cells in the body.

Health Conditions Related to Genetic Changes

Glycogen storage disease type VI

At least 17 mutations in the *PYGL* gene have been found to cause glycogen storage disease type VI (GSDVI). Most mutations change single protein building blocks (amino acids) in liver glycogen phosphorylase, affecting the normal function of the enzyme. In the Old Order Mennonite population, a common mutation (written as 1620+1G>A) disrupts the way the *PYGL* gene's instructions are used to make the enzyme. A defective liver glycogen phosphorylase enzyme impairs the normal breakdown of glycogen. As a result, liver cells cannot use glycogen for energy, so liver function becomes impaired. A lack of glycogen breakdown within liver cells leads to the major features of GSDVI.

Other Names for This Gene

- glycogen phosphorylase, liver form
- GSD6
- phosphorylase, glycogen, liver
- PYGL_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PYGL (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5836[geneid])

Scientific Articles on PubMed

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

Catalog of Genes and Diseases from OMIM

- GLYCOGEN PHOSPHORYLASE, LIVER; PYGL (https://omim.org/entry/613741)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/5836)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=PYGL[gene])

References

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- Burwinkel B, Bakker HD, Herschkovitz E, Moses SW, Shin YS, Kilimann MW. Mutations in the liver glycogen phosphorylase gene (PYGL) underlying glycogenosistype VI. Am J Hum Genet. 1998 Apr;62(4):785-91. doi: 10.1086/301790. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/9529348) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1377030/)
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Genomic Location

The *PYGL* gene is found on chromosome 14 (https://medlineplus.gov/genetics/chromosome/14/).

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