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.
Chromosomal Location

Cytogenetic Location: 14q22.1, which is the long (q) arm of chromosome 14 at position 22.1

Molecular Location: base pairs 50,905,217 to 50,944,483 on chromosome 14 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)

Other Names for This Gene

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

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): Glucose Regulation of Liver Glycogen Metabolism
  https://www.ncbi.nlm.nih.gov/books/NBK22444/?rendertype=figure&id=A2963
  https://www.ncbi.nlm.nih.gov/books/NBK22467/#A2918

Clinical Information from GeneReviews

- Glycogen Storage Disease Type VI
  https://www.ncbi.nlm.nih.gov/books/NBK5941
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?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
  http://omim.org/entry/613741

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=PYGL%5Bgene%5D

- HGNC Gene Symbol Report

- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:5836

- NCBI Gene

- UniProt
  https://www.uniprot.org/uniprot/P06737

Sources for This Summary


