

PYGM gene

glycogen phosphorylase, muscle associated

Normal Function

The *PYGM* gene provides instructions for making an enzyme called myophosphorylase. This enzyme breaks down a complex sugar called glycogen. Myophosphorylase is one of three related enzymes called glycogen phosphorylases that break down glycogen in cells. Myophosphorylase is found only in muscle cells, where it breaks down glycogen into a simpler 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.

Health Conditions Related to Genetic Changes

Glycogen storage disease type V

Approximately 130 mutations in the *PYGM* gene have been found to cause glycogen storage disease type V (GSDV). One mutation that is common in North American and European populations is written as Arg50Ter or R50X. This mutation creates a premature stop signal in the instructions for making myophosphorylase, which decreases the production of the enzyme. A shortage of myophosphorylase impairs the normal breakdown of glycogen. Other mutations that cause GSDV may severely reduce enzyme activity or change the way the enzyme folds into a 3-dimensional shape. The defective enzyme is unable to break down glycogen. As a result, muscle cells cannot produce enough energy, so muscles become easily fatigued. Reduced energy production in muscle cells leads to the major features of GSDV.

Other Names for This Gene

- glycogen phosphorylase, muscle form
- myophosphorylase
- phosphorylase, glycogen, muscle
- PYGM_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PYGM ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5837\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5837[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28PYGM%5BTIAB%5D%29+OR+%28myophosphorylase%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+1800+days%22%5Bdp%5D%29%29%29>)

Catalog of Genes and Diseases from OMIM

- GLYCOGEN PHOSPHORYLASE, MUSCLE; PYGM (<https://omim.org/entry/608455>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/5837>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=PYGM\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=PYGM[gene]))

References

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Genomic Location

The *PYGM* gene is found on chromosome 11 (<https://medlineplus.gov/genetics/chromosome/11/>).

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