PGAM2 gene

phosphoglycerate mutase 2

Normal Function

The *PGAM2* gene provides instructions for making an enzyme called phosphoglycerate mutase. The version of phosphoglycerate mutase produced from this gene is found predominantly in skeletal muscle cells. (Skeletal muscles are the muscles used for movement.) Another version of this enzyme, which is produced from a different gene, is found in many other cells and tissues.

Phosphoglycerate mutase is involved in a critical energy-producing process known as glycolysis. During glycolysis, the simple sugar glucose is broken down to produce energy. Phosphoglycerate mutase helps carry out a chemical reaction that converts a molecule called 3-phosphoglycerate, which is produced during the breakdown of glucose, to another molecule called 2-phosphoglycerate.

Health Conditions Related to Genetic Changes

Phosphoglycerate mutase deficiency

At least five mutations in the *PGAM2* gene have been found to cause phosphoglycerate mutase deficiency. The most common of these mutations, written as Trp78Ter or W78X, replaces the protein building block (amino acid) tryptophan with a premature stop signal in the instructions for making phosphoglycerate mutase. This mutation results in the production of an abnormally short, nonfunctional version of the enzyme. Other mutations change single amino acids in phosphoglycerate mutase.

Mutations in the *PGAM2* gene greatly reduce the activity of phosphoglycerate mutase, which disrupts energy production in skeletal muscle cells. This defect underlies the muscle cramping, muscle breakdown, and related signs and symptoms that occur following strenuous exercise in affected individuals.

Other Names for This Gene

- BPG-dependent PGAM 2
- GSD10
- MGC88743
- muscle-specific phosphoglycerate mutase

- PGAM-M
- PGAM2_HUMAN
- PGAMM
- phosphoglycerate mutase 2 (muscle)
- phosphoglycerate mutase isozyme M

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PGAM2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5224[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28PGAM2%5BTIAB%5D%29+OR+%28phosphoglycerate+mutase+2%5BTIAB%5D%29%29+OR+%28PGAM-M%5BTIAB%5D%29+OR+%28%28phosphoglycerate+mutase%5BTIAB%5D%29+AND+%28muscle%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

Catalog of Genes and Diseases from OMIM

- PHOSPHOGLYCERATE MUTASE 2; PGAM2 (https://omim.org/entry/612931)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/5224)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=PGAM2[gene])

References

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Genomic Location

The *PGAM2* gene is found on chromosome 7 (https://medlineplus.gov/genetics/chromosome/7/).

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