

Phosphoglycerate mutase deficiency

Description

Phosphoglycerate mutase deficiency is a disorder that primarily affects muscles used for movement (skeletal muscles). Beginning in childhood or adolescence, affected individuals experience muscle aches or cramping following strenuous physical activity. Some people with this condition also have recurrent episodes of myoglobinuria. Myoglobinuria occurs when muscle tissue breaks down abnormally and releases a protein called myoglobin, which is processed by the kidneys and released in the urine. If untreated, myoglobinuria can lead to kidney failure.

In some cases of phosphoglycerate mutase deficiency, microscopic tube-shaped structures called tubular aggregates are seen in muscle fibers. It is unclear how tubular aggregates are associated with the signs and symptoms of the disorder.

Frequency

Phosphoglycerate mutase deficiency is a rare condition; about 15 affected people have been reported in the medical literature. Most affected individuals have been African American.

Causes

Phosphoglycerate mutase deficiency is caused by mutations in the *PGAM2* gene. This gene provides instructions for making an enzyme called phosphoglycerate mutase, which is involved in a critical energy-producing process in cells known as glycolysis. During glycolysis, the simple sugar glucose is broken down to produce energy.

The version of phosphoglycerate mutase produced from the *PGAM2* gene is found primarily in skeletal muscle cells. Mutations in the *PGAM2* gene greatly reduce the activity of phosphoglycerate mutase, which disrupts energy production in these cells. This defect underlies the muscle cramping and myoglobinuria that occur after strenuous exercise in affected individuals.

[Learn more about the gene associated with Phosphoglycerate mutase deficiency](#)

- [PGAM2](#)

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the *PGAM2* gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition. However, people who carry one altered copy of the *PGAM2* gene may have some features of phosphoglycerate mutase deficiency, including episodes of exercise-induced muscle cramping and myoglobinuria.

Other Names for This Condition

- Deficiency mutase phosphoglycerate
- Glycogen storage disease X
- GSD X
- GSD10
- GSDX
- Myopathy due to phosphoglycerate mutase deficiency
- PGAM deficiency
- PGAMM deficiency

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Glycogen storage disease type X (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0268149/>)

Genetic and Rare Diseases Information Center

- Glycogen storage disease due to phosphoglycerate mutase deficiency (<https://rarediseases.info.nih.gov/diseases/9964/index>)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

Clinical Trials

- ClinicalTrials.gov ([https://clinicaltrials.gov/search?cond=%22Phosphoglycerate mutase deficiency%22](https://clinicaltrials.gov/search?cond=%22Phosphoglycerate%20mutase%20deficiency%22))

Catalog of Genes and Diseases from OMIM

- GLYCOGEN STORAGE DISEASE X; GSD10 (<https://omim.org/entry/261670>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28phosphoglycerate+mutase+deficiency%5BTIAB%5D%29+OR+%28glycogen+storage+disease+type+X%5BTIAB%5D%29%29+OR+%28%28PGAM%5BTIAB%5D%29+AND+%28deficiency%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>)

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