

Glycogen storage disease type V

Description

Glycogen storage disease type V (also known as GSDV or McArdle disease) is an inherited disorder caused by an inability to break down a complex sugar called glycogen in muscle cells. A lack of glycogen breakdown interferes with the function of muscle cells.

People with GSDV typically experience fatigue, muscle pain, and cramps during the first few minutes of exercise (exercise intolerance). Exercise such as weight lifting or jogging usually triggers these symptoms in affected individuals. The discomfort is generally alleviated with rest. If individuals rest after brief exercise and wait for their pain to go away, they can usually resume exercising with little or no discomfort (a characteristic phenomenon known as "second wind").

Prolonged or intense exercise can cause muscle damage in people with GSDV. About half of people with GSDV experience breakdown of muscle tissue (rhabdomyolysis). In severe episodes, the destruction of muscle tissue releases a protein called myoglobin, which is filtered through the kidneys and released in the urine (myoglobinuria). Myoglobin causes the urine to be red or brown. This protein can also damage the kidneys, and it is estimated that half of those individuals with GSDV who have myoglobinuria will develop life-threatening kidney failure.

The signs and symptoms of GSDV can vary significantly in affected individuals. The features of this condition typically begin in a person's teens or twenties, but they can appear anytime from infancy to adulthood. In most people with GSDV, the muscle weakness worsens over time; however, in about one-third of affected individuals, the muscle weakness is stable. Some people with GSDV experience mild symptoms such as poor stamina; others do not experience any symptoms.

Frequency

GSDV is a rare disorder; however, its prevalence is unknown. In the Dallas-Fort Worth area of Texas, where the prevalence of GSDV has been studied, the condition is estimated to affect 1 in 100,000 individuals.

Causes

Mutations in the *PYGM* gene cause GSDV. The *PYGM* gene provides instructions for making an enzyme called myophosphorylase. This enzyme 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.

PYGM gene mutations prevent myophosphorylase from breaking down glycogen effectively. 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.

[Learn more about the gene associated with Glycogen storage disease type V](#)

- *PYGM*

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the 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.

Other Names for This Condition

- Glycogen storage disease type 5
- Glycogenosis 5
- GSD type V
- GSD V
- McArdle disease
- McArdle syndrome
- McArdle type glycogen storage disease
- McArdle's disease
- Muscle glycogen phosphorylase deficiency
- Muscle phosphorylase deficiency
- Myophosphorylase deficiency
- *PYGM* deficiency

Additional Information & Resources

[Genetic Testing Information](#)

- Genetic Testing Registry: Glycogen storage disease, type V (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0017924/>)

Genetic and Rare Diseases Information Center

- Glycogen storage disease due to muscle glycogen phosphorylase deficiency (<https://rarediseases.info.nih.gov/diseases/6528/index>)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov ([https://clinicaltrials.gov/search?cond=%22Glycogen storage disease type V%22](https://clinicaltrials.gov/search?cond=%22Glycogen+storage+disease+type+V%22))

Catalog of Genes and Diseases from OMIM

- GLYCOGEN STORAGE DISEASE V; GSD5 (<https://omim.org/entry/232600>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28glycogen+storage+disease+type+V%5BTIAB%5D%29+OR+%28McArdle+disease%5BTIAB%5D%29+OR+%28GSDV%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>)

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