

Glycogen storage disease type VI

Description

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

The signs and symptoms of GSDVI typically begin in infancy to early childhood. The first sign is usually an enlarged liver (hepatomegaly). During prolonged periods without food (fasting), affected individuals may have low blood sugar (hypoglycemia) or elevated levels of ketones in the blood (ketosis). Ketones are molecules produced during the breakdown of fats, which occurs when stored sugars are unavailable. Children with GSDVI tend to grow slower than their peers, but they often achieve normal height as adults. Some affected children also have mild delays in the development of motor skills, such as sitting, standing, or walking.

The signs and symptoms of GSDVI tend to improve with age; most adults with this condition do not have any related health problems.

Frequency

The exact prevalence of GSDVI is unknown. At least 11 cases have been reported in the medical literature, although this condition is likely to be underdiagnosed because it can be difficult to detect in children with mild symptoms or adults with no symptoms. GSDVI is more common in the Old Order Mennonite population, with an estimated incidence of 1 in 1,000 individuals.

Causes

Mutations in the *PYGL* gene cause GSDVI. The *PYGL* gene provides instructions for making an enzyme called liver glycogen phosphorylase. This enzyme 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.

PYGL gene mutations prevent liver glycogen phosphorylase from breaking down glycogen effectively. Because liver cells cannot break down glycogen into glucose, individuals with GSDVI can have hypoglycemia and may use fats for energy, resulting in

ketosis. Glycogen accumulates within liver cells, causing these cells to become enlarged and dysfunctional.

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

- PYGL

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

- GSD type VI
- GSD VI
- GSD6
- Hepatic glycogen phosphorylase deficiency
- Hers disease
- Liver phosphorylase deficiency syndrome

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Glycogen storage disease, type VI (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0017925/>)

Genetic and Rare Diseases Information Center

- Glycogen storage disease due to liver glycogen phosphorylase deficiency (<https://rarediseases.info.nih.gov/diseases/6529/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 dise](https://clinicaltrials.gov/search?cond=%22Glycogen+storage+dise)

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Catalog of Genes and Diseases from OMIM

- GLYCOGEN STORAGE DISEASE VI; GSD6 (<https://omim.org/entry/232700>)

Scientific Articles on PubMed

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

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