

What is Glycogen Storage Disease Type Ib?

Glycogen storage disease type I (GSD-I), also called von Gierke disease¹, is an inherited disease caused by a defect in the body's ability to break down glycogen (the form in which the body stores sugar) to glucose (a free form of sugar and the body's main source of energy)². Symptoms associated with GSD-I are attributed to low blood glucose levels and excessive storage of glycogen in the liver and kidneys. GSD I occurs in two forms: GSD-Ia and GSD-Ib. GSD-Ib is caused by a deficiency of the enzyme glucose-6-phosphate transporter (G6PT) whose function is to help maintain normal blood glucose levels.²

What are the symptoms of glycogen storage disease type Ib and what treatment is available?

Signs and symptoms of GSD-Ib typically begin around three to four months of age. Initial signs include low blood sugar and enlarged liver. GSD-Ib is characterized by^{1, 3}:

- Low blood sugar levels (hypoglycemia)
- Enlarged liver and kidneys (hepatomegaly and renomegaly)
- High levels of lactic acid and uric acid in the blood
- High levels of fat in the blood (hyperlipidemia)
- Growth retardation and short stature
- Delayed puberty
- Kidney disease (proteinuria, hypertension, kidney stones, renal failure)
- Gout (red, tender, hot, swollen joints)
- Liver tumors (typically non-cancerous)
- Pulmonary hypertension
- Decreased bone density (osteoporosis)
- Recurrent bacterial and fungal infections
- Oral (mouth) and intestinal ulcers

There is no cure for GSD-Ib, but with treatment, many affected individuals live into adulthood. Long term complications may be minimized or delayed with early intervention and ongoing care. Treatment measures include dietary therapy to maintain normal blood sugar levels and provide optimal nutrition for growth and development; prescription medications for complications such as gout, hyperlipidemia, kidney disease, and infections; and dialysis or transplant for severe kidney disease. Normal growth and puberty may be expected in treated children³.

How is glycogen storage disease type 1b inherited?

GSD-Ib is an autosomal recessive disease caused by mutations in the *SLC37A4* gene³. An individual who inherits one gene mutation in the *SLC37A4* gene is a "carrier" of GSD-Ib and is not expected to have related health problems. An individual who inherits two mutations in the *SLC37A4* gene, one from each parent, is expected to be affected with GSD-Ib.

If both members of a couple are carriers, the risk for an affected child is 25% in each pregnancy. Therefore, it is especially important that the reproductive partner of a carrier be offered testing.

Who is at risk for glycogen storage disease type Ib?

GSD type Ib can occur in individuals of any ethnic background. The incidence of GSD-Ib is estimated to be 1 in 500,000 with a carrier frequency of 1 in 354^{1,3}.



What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetics professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

Where can I get more information?

- Association for Glycogen Storage Disease (AGSD): www.agsdus.org
- Children Living with Inherited Metabolic Diseases (CLIMB): www.climb.org.uk

References

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2. Chen Y-T. Glycogen storage diseases. In: Scriver CR, Beaudet AL, Sly WS, Valle D, eds. Childs B, Kinzler KW, Vogelstein B, assoc. eds. *The Metabolic and Molecular Bases of Inherited Disease*. 8 ed. New York: McGraw-Hill, 2001:1521–51.
3. GeneReviews: <http://www.ncbi.nlm.nih.gov/books/NBK1312/>; Accessed February 20, 2012.

