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What is Glycogen Storage Disease Type Ia?
Glycogen storage disease type 1 (GSD I), also called von Gierke disease[1], 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). Symptoms associated with GSD I are attributed to low blood glucose levels and excessive accumulation of glycogen and fat in the liver and kidneys.[2] GSD I occurs in two forms GSD-Ia and GSD-Ib. GSD-Ia, which accounts for about 80% of all GSD-I cases, is caused by a deficiency of an enzyme called glucose-6-phosphatase (G6Pase).[1,2]

What are the Symptoms of Glycogen Storage Disease Type Ia and What Treatments are Available?
Signs and symptoms of GSD 1a typically begin around three to four months of age. Initial signs include low blood sugar and enlarged liver. GSD-Ia is characterized by[2]:

- Low blood sugar levels (hypoglycemia)
- Enlarged liver and kidneys (hepatomegaly and renomegaly)
- High levels of lactic acid and uric acid in the blood (lactic acidemia and hyperuricemia)
- 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 (high blood pressure in the lungs causing shortness of breath, chest pain, and rapid heart rate)
- Decreased bone density (osteoporosis)

There is no permanent cure for GSD-Ia, 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; dialysis for kidney disease; kidney and liver transplantation are also available. Normal growth and puberty may be expected in treated children.[2]

How is Glycogen Storage Disease Type Ia Inherited?
GSD-Ia is an autosomal-recessive disease most commonly caused by mutations in the *G6PC* gene.[2] An individual who inherits one gene mutation in the *G6PC* gene is a "carrier" of GSD-Ia and is not expected to have related health problems. An individual who inherits two mutations in the *G6PC* gene, one from each parent, is expected to be affected with GSD-Ia.

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 Ia?
GSD-Ia can occur in individuals of any ethnic background. The overall incidence of GSD-Ia is estimated to be 1 in 125,000 with a carrier frequency of 1 in 177.[1] It is known to be more common in individuals of Ashkenazi Jewish ancestry with a calculated incidence of 1 in 16,000 based on a carrier frequency of 1 in 64.[3]

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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

References

1. Chou JY, et al. Type I glycogen storage diseases: Disorders of the glucose-6-phosphatase complex. *Curr Mol Med*. 2002 Mar; 2(2): 121-143.
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3. Scott SA, et al. Experience with carrier screening and prenatal diagnosis for 16 Ashkenazi Jewish genetic diseases. *Hum Mut.* 2010 Nov; 31(11): 1240-1250.

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