Genetics Uncoded:

NxGen MDx

Facts about

Glycogen Storage Disease Type 1a

What Your Test Results Mean

Carriers typically show no symptoms of glycogen storage disease type 1A; however, carriers are at an increased risk of having a child with glycogen storage disease type 1A. Risk for the current or future pregnancies is dependent on your partner's carrier status.

Glycogen Storage Disease Type 1a Explained

Glycogen storage disease type 1A is an inherited metabolic disorder in which harmful amounts of glycogen and fat accumulate in the liver and kidneys. Individuals with glycogen storage disease type 1A do not produce enough of one of the enzymes, glucose-6-phosphatase. This enzyme is needed to metabolize glucose-6-phosphate into glucose. Lack of this enzyme causes severe hypoglycemia that can lead to seizures and brain damage. Some infants with glycogen storage disease type 1A present with severe hypoglycemia while others present at three to four months of age with hepatomegaly, lactic acidosis, and/or seizures. Long-term complications of untreated glycogen storage disease include neurocognitive symptoms, anemia, short stature, osteoporosis, delayed puberty, gout, renal disease, pancreatitis, and risk for hepatocellular carcinoma later in life. Many affected individuals live into adulthood with appropriate treatment with normal growth and puberty.

Treatment of individuals with glycogen storage disease type 1A typically includes care by a metabolic team, specifically to monitor medical complications and nutrition. A diet low in fructose and sucrose, overnight glucose infusion, cornstarch therapy, and frequent daytime feedings may be recommended in an effort to prevent hypoglycemia. Liver transplantation has been used in individuals with glycogen storage disease type 1A to restore metabolic balance and reduce the risk for hepatocellular carcinoma.

How the Genetics Work

The clinical features of glycogen storage disease type 1A can be explained by pathogenic variants in the *G6PC* gene. In general, individuals have two copies of the *G6PC* gene. Carriers of glycogen storage disease type 1A have a single variant in one copy of the *G6PC* gene while individuals with glycogen storage disease type 1A have variants in both copies of their *G6PC* genes, one inherited from each parent.

Questions? Contact us at **1-855-776-9436** to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.

nxgenmdx.com

801 Broadway Ave NW, Suite 203
Grand Rapids, MI, 49504

Revised 12/15/16