NxGen MDx

Glycogen Storage Disease Type IA

What Your Results Mean

Test results indicate that you are a carrier of glycogen storage disease type IA. Carriers are not expected to show symptoms. You and your partner or donor would both have to be carriers of glycogen storage disease type Ia for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment.

Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning.

Recommended Next Steps

Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner or donor are carriers for glycogen storage disease type IA, each of your children has a 1 in 4 (25%) chance to have the condition.

Glycogen Storage Disease Type IA Explained

What is Glycogen Storage Disease Type IA?

Glycogen storage disease type IA is an inherited metabolic disorder that causes harmful amounts of glycogen and fat to accumulate in the liver and kidneys. Individuals with glycogen storage disease type IA do not produce enough glucose-6-phosphatase, an enzyme needed to metabolize glucose-6-phosphate into glucose. This causes severe hypoglycemia that can lead to seizures and brain damage. Some infants with glycogen storage disease type IA present with severe hypoglycemia, while others present at 3-4 months of age with hepatomegaly, lactic acidosis, and/or seizures. Long-term complications of untreated glycogen storage disease type IA include neurocognitive symptoms, anemia, short stature, osteoporosis, delayed puberty, gout, renal disease, pancreatitis, and risk for hepatocellular carcinoma later in life.

Prognosis

With early diagnosis and appropriate treatment and management, prognosis is typically favorable. Many affected individuals live into adulthood with appropriate treatment, including normal growth and puberty. Early treatment may also decrease the development of gout, kidney failure, life-threatening low blood sugar, and liver tumors.

Treatment

A diet low in fructose and sucrose, overnight glucose infusion, cornstarch therapy, and frequent daytime feedings are treatment options to prevent hypoglycemia. Liver transplantation has been used to restore metabolic balance and reduce the risk for hepatocellular carcinoma. Treatment typically includes care by a metabolic team to monitor medical complications and nutrition.

Resources

Association for Glycogen Storage Disease
https://www.agsdus.org/type-i.php
Genetics Home Reference
https://ghr.nlm.nih.gov/condition/glycogen-storage-disease-type-i
National Society of Genetic Counselors
https://www.nsgc.org/

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Revised: 4/2022