NxGen MDx

Glycogen Storage Disease Type IB

What Your Results Mean

Test results indicate that you are a carrier of glycogen storage disease type IB. Carriers are not expected to show symptoms of glycogen storage disease type IB. You and your partner or donor would both have to be carriers of glycogen storage disease type IB 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 IB, each of your children has a 1 in 4 (25%) chance to have the condition.

Glycogen Storage Disease Type IB Explained

What is Glycogen Storage Disease Type IB?

Glycogen storage disease type IB 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 IB do not produce enough of the enzyme glucose-6-phosphatase which os needed to metabolize glucose-6-phosphate into glucose. Symptoms of the condition usually present at 3-4 months old and may include enlarged liver, swollen abdomen, delayed or stunted growth, irritability, and seizures. Children are also at increased risk to develop neutropenia, which makes them prone to recurrent bacterial infections. People with glycogen storage disease type IB may have oral problems such as cavities, inflammation of the gums, chronic gum disease, abnormal tooth development, and open sores in the mouth. Long-term complications of untreated glycogen storage disease type IB include growth retardation resulting in short stature, thinning of the bones, relatively thin arms and legs, delayed puberty, gout, kidney disease, high blood pressure in the lungs, liver tumors, polycystic ovaries, pancreatitis, and changes in brain function.

Prognosis

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

Treatment

Treatment includes a diet low in fructose and sucrose, overnight glucose infusion, cornstarch therapy, and frequent daytime feedings to prevent hypoglycemia. Liver transplantation has been used to restore metabolic balance and reduce the risk for hepatocellular carcinoma. Treatment with human granulocyte colony-stimulating factor for recurrent infections may be beneficial. Treatment of individuals with glycogen storage disease type IB typically includes care by a metabolic team to monitor medical complications and nutrition.

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Resources

Association for Glycogen Storage Disease
https://www.agsdus.org/type-ii.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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