What Your Test Results Mean

Testing results indicate that you are a carrier of hereditary fructose intolerance. Carriers typically show no symptoms of hereditary fructose intolerance; however, carriers are at an increased risk of having a child with hereditary fructose intolerance. Risk for the current or future pregnancies is dependent on your partner’s carrier status. Carrier testing of your partner is recommended in addition to consultation with a genetic counselor.

Hereditary Fructose Intolerance Explained

Hereditary fructose intolerance is an inherited condition that affects fructose metabolism. A reduced level of the enzyme Aldolase B prevents the body from breaking down fructose. After ingesting fructose, individuals with hereditary fructose intolerance may experience nausea, bloating, abdominal pain, diarrhea, vomiting, and low blood sugar. Affected infants may fail to grow and gain weight at the expected rate, and repeated ingestion of fructose-containing foods can lead to liver and kidney damage. Continued exposure to fructose may result in seizures, coma, and ultimately death from liver and kidney failure. However, most affected individuals can live without these symptoms by following a diet free of fructose, sucrose, and sorbitol.

How the Genetics Work

Hereditary fructose intolerance is an autosomal recessive disorder caused by mutations in the ALDOB gene. In general, individuals have two copies of the ALDOB gene. Carriers of hereditary fructose intolerance have a single mutation in one copy of the ALDOB gene while individuals with hereditary fructose intolerance have mutations in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%. 

Recommended Next Steps

Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

Questions?

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