NxGen MDx

Hereditary Fructose Intolerance

What Your Results Mean

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

Hereditary Fructose Intolerance Explained

What is Hereditary Fructose Intolerance?

Hereditary fructose intolerance is an inherited condition that affects a person's ability to digest the sugar fructose, found mostly in fruits. 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.

Prognosis

Early diagnosis and strict fructose restriction are vital for a good prognosis. Disease symptoms usually disappear with a totally fructose-free diet. Once fructose is restricted, infants often develop a self-protective dislike to foods that cause symptoms. Poor management or lack of treatment can lead to poor feeding, growth failure, jaundice, hepatomegaly, hemorrhage, and progressive liver damage, which may be fatal in some cases.

Treatment

Treatment includes dietary restriction of fructose. Early diagnosis and early implementation of diet is ideal.

Resources

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/hereditary-fructose-intolerance
Genetic and Rare Diseases Information Center
https://rarediseases.info.nih.gov/diseases/6622/hereditary-fructose-intolerance
National Society of Genetic Counselors
https://www.nsgc.org/

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