Tyrosinemia Type I

What Your Results Mean

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

Tyrosinemia Type I Explained

What is Tyrosinemia Type I?

Tyrosinemia type I is an inherited metabolic disorder that often presents in infancy. The condition is characterized by failure to thrive, diarrhea, vomiting, jaundice, and an increased tendency to bleed. Infants with this condition may also have a cabbage-like odor. Tyrosinemia type I can potentially lead to liver and kidney failure and affect the nervous system. Affected individuals also have an increased risk for liver cancer.

Prognosis

Prognosis is poor if the disease is left untreated, and typically fatal by age 10. However, with prompt and appropriate treatment and management, prognosis can be good. Treated individuals can have a normal life span with typical growth, bone structure, and improved liver and kidney function.

Treatment

Treatment consists of dietary restriction of tyrosine and daily nitisinone (an FDA-approved drug) intake. A metabolic specialist and dietitian typically manage care of individuals with the disorder. The earlier the disease is recognized and treated, the less damage is done to the body and the better the prognosis.

Resources

National Organization for Rare Disorders
https://rarediseases.org/rare-diseases/tyrosinemia-type-1/
Genetics Home Reference
https://ghr.nlm.nih.gov/condition/tyrosinemia
National Society of Genetic Counselors
https://www.nsgc.org/

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