Tyrosinemia Type II

What Your Results Mean

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

Tyrosinemia Type II Explained

What is Tyrosinemia Type II?

Tyrosinemia type II is an inherited metabolic disorder that often presents in early childhood. This condition can affect the eyes, skin, and intellectual development. Symptoms include excessive tearing, abnormal sensitivity to light (photophobia), eye pain and redness, poor vision, and painful skin lesions on the palms and soles (palmoplantar hyperkeratosis). About half of individuals have intellectual disability.

Prognosis

Treatment may not be effective for all individuals. The effect on life expectancy remains unknown, and survival into adulthood has been reported.

Treatment

There is no cure for tyrosinemia type II. Treatment consists of a low-protein diet and medical food. 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. Treatment may not be effective for all individuals.

Resources

National Organization for Rare Disorders
https://rarediseases.info.nih.gov/diseases/3105/tyrosinemia-type-2

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/tyrosinemia

National Society of Genetic Counselors
https://www.nsgc.org/

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