

Choroideremia

What Your Results Mean

Test results indicate that you are a carrier of choroideremia. Female carriers typically show no symptoms of choroideremia; however, some female carriers may show small areas of cell loss within the retina, which can be observed during a thorough eye examination. These changes may impair vision later in life. Female carriers are at an increased risk to have a child with choroideremia, but risk is dependent on the sex of the child--this is known as X-linked inheritance. Consultation with a genetic counselor for a more detailed risk assessment is recommended.



We recommend you share and discuss this information with all of your health care providers. Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning and their own personal clinical management.

Recommended Next Steps

Risk for a female child to be a carrier is 50% and risk for a male child to have choroideremia is 50%. In the absence of clinical symptoms, reflexive testing for male partners of choroideremia carriers is typically not indicated due to the X-linked inheritance pattern. Consultation with a genetic counselor for a more detailed risk assessment is recommended.

Choroideremia Explained

What is Choroideremia?

Choroideremia is an inherited condition that is characterized by progressive vision loss. This vision loss usually begins before an individual reaches 20 years of age. Night vision is the first to be affected, followed by peripheral vision. Field of vision gradually decreases, affecting a person's central vision by about age 40-50 and eventually leading to blindness.

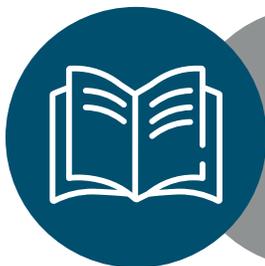


Prognosis

Prognosis is considered favorable. This condition does not affect an individual's life expectancy.

Treatment

Treatment is symptomatic as there currently is no cure for choroideremia. Low vision aids and services are recommended.



Resources

Choroideremia Research Foundation

<http://curechm.org/>

Genetics Home Reference

<https://ghr.nlm.nih.gov/condition/choroideremia#statistics>

National Organization for Rare Disorders (NORD)

<https://rarediseases.org/rare-diseases/choroideremia/>

National Society of Genetic Counselors

<https://www.nsgc.org/>