NxGen MDx

X-Linked Juvenile Retinoschisis

What Your Results Mean

Test results indicate that you are a carrier of X-linked juvenile retinoschisis. Female carriers typically show no symptoms; however, carriers are at an increased risk of having a child with X-linked juvenile retinoschisis. Risk for the current or future pregnancies is sex dependent. Consultation with a genetic counselor for a more detailed risk assessment is recommended.

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

Risk to have a female carrier is 50% and risk to have a male with X-linked juvenile retinoschisis is 50%. Prenatal testing for X-linked juvenile retinoschisis is available, as well as testing shortly after birth to rule out a diagnosis in a child, particularly if the child is a male. In the absence of clinical symptoms, reflexive testing for male partners of X-linked juvenile retinoschisis carriers is typically not indicated due to the X-linked inheritance pattern. Follow-up with your provider to discuss your testing options and consultation with a genetic counselor for a more detailed risk assessment is recommended.

X-Linked Juvenile Retinoschisis Explained

What is X-Linked Juvenile Retinoschisis?

X-linked juvenile retinoschisis is an inherited condition that occurs almost exclusively in males. This condition is characterized by damage to the retina of the eye that causes progressive vision loss during childhood and adolescence. This condition is typically diagnosed when affected boys start school and poor vision and difficulty with reading become apparent. Vision usually stabilizes in an affected individual's twenties but may deteriorate again in middle age and eventually give way to legal blindness.

Prognosis

X-linked juvenile retinoschisis does not affect lifespan. However, affected individuals will have vision problems that may result in legal blindness after middle age.

Treatment

There is no cure for X-linked juvenile retinoschisis, but tools such as eyeglasses, magnifying glasses, high-contrast reading material, and adaptive software can help affected individuals cope with poor vision. All affected individuals should be followed by an ophthalmologist throughout their life, beginning in childhood. Additionally, affected individuals should avoid high contact sports and other activities that might cause a hard blow to the head to minimize the risk of retinal detachment or bleeding in the eye.

Resources

Foundation Fighting Blindness
http://www.blindness.org/x-linked-retinoschisis-xlrs-0

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/x-linked-juvenile-retinoschisis

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

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