Genetics Uncoded:

Facts about

X-Linked Juvenile Retinoschisis

What Your Test Results Mean

Carriers typically show no symptoms of X-linked juvenile retinoschisis; however, carriers are at an increased risk of having a child with X-linked juvenile retinoschisis. Because this disease is X-linked, risk for the current or future pregnancies is sex-dependent.

X-Linked Juvenile Retinoschisis Explained

X-linked juvenile retinoschisis is an inherited condition that causes the retina of the eye to split or develop cysts or lesions over time. This causes progressive vision loss during childhood and adolescence. Vision usually stabilizes in an affected individual's twenties, but may deteriorate again in middle age and eventually give way to legal blindness. 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. Lifespan is not affected.

How the Genetics Work

The clinical features of X-linked juvenile retinoschisis can be explained by pathogenic variants in the *RS1* gene. Women have two copies of the *RS1* gene and can be unaffected carriers of X-linked juvenile retinoschisis by possessing a single variant in one copy of the *RS1* gene. Men have one copy of the X chromosome, so they have one copy of the *RS1* gene. If they inherit a variant in the *RS1*, they will have X-linked juvenile retinoschisis. Sons of carrier women have a 50% chance of having with X-linked juvenile retinoschisis, while daughters will not have this disease unless their father also has the disease. As a result of this inheritance pattern, this disease primarily affects males.

Questions? Contact us at **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.

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