Achromatopsia

What Your Results Mean

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

Achromatopsia Explained

What is Achromatopsia?

Achromatopsia is a non-progressive inherited disorder characterized by partial or complete loss of color vision. Affected individuals can only see black, white, and various shades of gray. Individuals with milder achromatopsia may have only partial loss of color vision that allows for some color. Additional signs and symptoms can include an intolerance to bright light, uncontrollable eye movements, and a reduced sharpness of image (low visual acuity). It is important to note that achromatopsia is different from red-green color blindness, a condition where individuals can perceive color but have difficulties differentiating the colors from each other.

Prognosis

Life expectancy is not affected by a diagnosis of achromatopsia; however, quality of life can be affected. Depending on the severity of visual acuity, individuals with achromatopsia may benefit from preferential seating in classrooms, low-vision aids, or attending a specialized school equipped to assist students that have varying degrees of visual impairments.

Treatment

Dark-tinted glasses improve the visual acuity of affected individuals, but there is no cure. Low-vision and occupational aids can assist individuals with achromatopsia in everyday living.

Resources

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

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

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