Clarity Genetics

Genetics Uncoded:

Facts about Achromatopsia

What Your Test Results Mean

Carriers show no symptoms of achromatopsia and are not at risk to develop symptoms of the disorder. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Achromatopsia Explained

Achromatopsia is an inherited disorder caused by abnormal eye photoreceptor cell function that leads to vision loss. Affected individuals are intolerant of bright light, have uncontrollable eye movements, and color blindness. Dark-tinted glasses improve the visual acuity of affected individuals, and low-vision and occupational aids can assist them in everyday living. No cures are available, but clinical trials are underway to determine whether an intraocular implant can improve visual acuity or color vision and reduce sensitivity to light.

How the Genetics Work

Achromatopsia is an autosomal recessive disorder. Approximately 40-50% of achromatopsia cases can be explained by pathogenic variants in the *CNGB3* gene. In general, individuals have two copies of the *CNGB3* gene. Carriers of achromatopsia have a single variant in one copy of the *CNGB3* gene while individuals with achromatopsia have variants in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

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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