Genetics Uncoded:

Facts about Retinitis Pigmentosa 59

What Your Test Results Mean

Carriers typically show no symptoms of retinitis pigmentosa 59; however, carriers are at an increased risk of having a child with retinitis pigmentosa 59. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Retinitis Pigmentosa 59 Explained

Retinitis pigmentosa 59 is an inherited condition that characterized by progressive vision loss. Affected individuals usually have normal vision in infancy and childhood, but are diagnosed with retinitis pigmentosa in their teenage years due to impaired night and peripheral vision. The photoreceptors on the retina continue to degenerate over time, and individuals eventually lose most or all of their vision and are declared legally blind by their thirties or forties. There is no cure for retinitis pigmentosum, but nutritional supplements such as vitamin A can help slow the progression of the disease, and researchers are working on experimental treatments such as retinal transplants and implants and drug therapy.

How the Genetics Work

Retinitis pigmentosa 59 is an autosomal recessive disorder caused by pathogenic variants in the *DHDDS* gene. In general, individuals have two copies of the *DHDDS* gene. Carriers of retinitis pigmentosa 59 have a single variant in one copy of the *DHDDS* gene while individuals with retinitis pigmentosa 59 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%.

Questions? Contact us at **1-855-776-9436** to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.

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