Genetics Uncoded:

NxGen MDx

Facts about

Costeff Optic Atrophy Syndrome

What Your Test Results Mean

Carriers show no symptoms of Costeff optic atrophy syndrome 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.

Costeff Optic Atrophy Syndrome Explained

Costeff optic atrophy syndrome is an inherited condition that affects visual acuity and muscle control. Children with this disease experience rapid optic atrophy starting in infancy resulting in poor vision. Affected individuals also develop muscle weakness and spasticity, leading many to require a wheelchair. Some patients also have mild cognitive problems. While the disease progresses quickly during childhood, it tends to stabilize over time and individuals survive well into adulthood.

How the Genetics Work

Costeff optic atrophy syndrome is an autosomal recessive disorder caused by pathogenic variants in the *OPA3* gene. In general, individuals have two copies of the *OPA3* gene. Carriers of Costeff optic atrophy syndrome have a single variant in one copy of the *OPA3* gene while individuals with Costeff optic atrophy syndrome 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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