3-Methylglutaconic Aciduria Type III (Costeff Optic Atrophy)

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of 3-methylglutaconic aciduria type III (Costeff optic atrophy). Carriers are not expected to show symptoms. You and your partner would both have to be carriers of Costeff optic atrophy 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 for Costeff optic atrophy, each of your children has a 1 in 4 (25%) chance to have the condition.

3-Methylglutaconic Aciduria Type III (Costeff Optic Atrophy) Explained

What is 3-Methylglutaconic Aciduria Type III (Costeff Optic Atrophy)?

Costeff optic atrophy is an inherited condition characterized by vision loss, delayed development, and movement problems. Vision loss is primarily caused by degeneration (atrophy) of the optic nerves, which carry information from the eyes to the brain. This typically begins in infancy or early childhood and results in vision impairment that worsens over time.

Development of motor skills such as walking is often delayed in people with Costeff optic atrophy. Affected individuals may also have speech difficulties (dysarthria). While some people with Costeff optic atrophy have mild to moderate intellectual disability, many have normal intelligence.

Movement problems in people with Costeff optic atrophy develop in late childhood and include muscle stiffness, impaired muscle coordination (ataxia), and involuntary jerking movements. As a result of these movement difficulties, individuals with Costeff optic atrophy may require wheelchair assistance.

Prognosis

While some individuals with this condition have been known to live into their thirties, life expectancy beyond that is unknown.

Treatment

Treatment is supportive. A multidisciplinary team including a neurologist, orthopedic surgeon, ophthalmologist, geneticist, and physical therapist is required for the care of affected individuals.

Resources

Organic Acidemia Association
https://www.oaanews.org/
Genetics Home Reference
https://ghr.nlm.nih.gov/condition/costeff-syndrome
National Society of Genetic Counselors
https://www.nsgc.org/

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