Genetics Uncoded:

NxGen MDx

Facts about Primary Hyperoxaluria Type 1

What Your Test Results Mean

Carriers typically show no symptoms of primary hyperoxaluria type 1; however, carriers are at an increased risk of having a child with primary hyperoxaluria type 1. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Primary Hyperoxaluria Type 1 Explained

Primary hyperoxaluria type 1 is an inherited condition that causes an excess accumulation of oxalate, the main component of kidney stones. Deposits of calcium oxalate can lead to kidney damage, kidney failure, and injury to other organs. Primary hyperoxaluria has no cure, but the condition can be managed through diet and medication. Individuals with primary hyperoxaluria should drink plenty of water and avoid foods high in oxalate, such as rhubarb and chocolate. Some patients also respond well to treatment with vitamin B6 or pyridoxine. Kidney and/or liver transplantation may be recommended depending on the severity of the disorder.

How the Genetics Work

Primary hyperoxaluria type 1 is an autosomal recessive disorder caused by variants in the *AGXT* gene. In general, individuals have two copies of the *AGXT* gene. Carriers of primary hyperoxaluria type 1 have a single variant in one copy of the *AGXT* gene while individuals with primary hyperoxaluria type 1 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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