Genetics Uncoded:

FACTS ABOUT PRIMARY HYPEROXALURIA TYPE 1

AKA | Serine-Pyruvate Aminotransferase Deficiency, Alanine-Glyoxylate Aminotransferase Deficiency, Hepatic Agt Deficiency, HP1, PH1, Peroxisomal Alanine-Glyoxylate Aminotransferase Deficiency, Oxalosis I, Glycolic Aciduria, Hyperoxaluria, Primary, Type 1

What Your Test Results Mean

Testing results indicate that you are a carrier of primary hyperoxaluria type 1. 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. Risk for the current or future pregnancies is dependent on your partner’s carrier status. Carrier testing of your partner is recommended in addition to consultation with a genetic counselor.

Primary Hyperoxaluria Type 1 Explained

Primary hyperoxaluria type 1 is an inherited condition that affects the body’s metabolism of oxalate. Accumulation of oxalate causes the oxalate ion to combine with calcium and crystallize in body tissues as calcium oxalate. This chemical is the main component of kidney stones, and 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 lots 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 mutations in the AGXT gene. In general, individuals have two copies of the AGXT gene. Carriers of primary hyperoxaluria type 1 have a single mutation in one copy of the AGXT gene while individuals with primary hyperoxaluria type 1 have mutations in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Recommended Next Steps

Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

Questions?

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