Primary Hyperoxaluria Type 2, *GRHPR*-Related

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of primary hyperoxaluria type 2, *GRHPR*-related. Carriers typically show no symptoms. Risk for 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 for 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 and their own personal clinical management.

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 or donor are carriers for primary hyperoxaluria type 2, *GRHPR*-related, each of your children has a 1 in 4 (25%) chance to have the condition.

Primary Hyperoxaluria Type 2, *GRHPR*-Related Explained

What is Primary Hyperoxaluria Type 2, *GRHPR*-Related?

Primary hyperoxaluria type 2, *GRHPR*-related (PH2) is a rare inherited condition that leads to kidney damage or injury to other organs due to excess oxalate in the body. Individuals with PH2 have excess oxalate because their bodies do not produce enough of the enzyme that is normally responsible for preventing this accumulation. This oxalate leads to a buildup of insoluble calcium salts in the kidneys and other organs. If untreated, it can result in life-threatening kidney failure.

People with PH2 are prone to recurrent kidney stones that can lead to kidney failure. The disease has similar symptoms to primary hyperoxaluria type 1 (PH1), but PH2 tends to be a less aggressive form of the disease, even when symptoms start early in life. PH1 and PH2 are caused by different missing liver enzymes.

In addition to the kidneys, PH2 also leaves insoluble calcium deposits in other body tissues, causing problems with bones, eyes, teeth, nerves, and the heart.

Symptoms typically begin between the ages of one and 25, with roughly 80% showing signs of the disease in late childhood or early adolescence. Another 10% of people with PH2 show symptoms in early infancy (before the age of six months) while the final 10% do not show symptoms until their forties or fifties.

Prognosis

Prognosis is variable. If left untreated, PH2 can lead to serious illness and even death. With appropriate treatment, including organ transplantation for those with end stage renal disease, individuals with PH2 can have improved prognosis. Individuals with PH2 typically have a better prognosis than those with PH1 due to fewer kidney stones and a lower incidence of end stage renal disease. Those individuals with PH2 who have kidney transplants must also face the challenges of lifelong use of immunosuppressive medications and other challenges of organ transplants.

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Treatment

Several treatments are available to manage the symptoms of PH2. One essential therapy for individuals with PH is the consumption of large volumes of water regularly throughout the day and night; this helps prevent accumulation of calcium oxalate. Individuals should also avoid a high intake of vitamins C because large amounts can cause stone formation. It is also recommended that individuals take certain medications or supplements, such as magnesium and potassium citrate, to inhibit calcium oxalate stone formation. After a certain point, individuals with PH2 may require dialysis to manage the calcium oxalate build-up in their body, though this can eventually lead to oxalate deposits in other major organs. Another therapy considered for individuals with PH2 is kidney transplantation; unlike with PH1, combined liver-kidney transplantation is not used for PH2, as the *GRHPR* enzyme is present throughout the body (not solely in the liver). Individuals with PH2 should also regularly monitor the other major organs that can be affected by calcium oxalate deposits. Furthermore, pregnant women with PH2 should be closely monitored by an obstetrician and a nephrologist because of a high risk for kidney stones during pregnancy or after delivery.

Resources
Oxalosis and Hyperoxaluria Foundation
http://ohf.org/
National Society of Genetic Counselors
https://www.nsgc.org/

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