Primary Hyperoxaluria Type 1, *AGXT*-Related

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of primary hyperoxaluria type 1, *AGXT*-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 1, *AGXT*-related, each of your children has a 1 in 4 (25%) chance to have the condition.

Primary Hyperoxaluria Type 1, *AGXT*-Related Explained

What is Primary Hyperoxaluria Type 1, *AGXT*-Related

Primary hyperoxaluria type 1, *AGXT*-related (PH1) is an inherited condition that leads to kidney damage or injury to other organs due to excess oxalate in the body. Individuals with PH1 have excess oxalate because their bodies do not produce enough of an 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 results in life-threatening kidney failure. Among people with PH1, 50% experience kidney failure by the age of 15, and 80% by the age of 30.

In addition to the kidneys, PH1 also leaves insoluble calcium deposits in other body tissues. This can lead to bone pain; vision loss; tingling, numbness, or pain in the extremities; enlargement of the liver and spleen; and problems with the electrical system of the heart (heart block).

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 PH1 show symptoms in early infancy (before the age of six months) while the remaining 10% do not show symptoms until their forties or fifties.

Prognosis

If left untreated, PH1 can lead to serious illness and even death. With appropriate treatment, including organ transplantation for those with end stage renal disease, individuals with PH1 can have improved prognosis. In individuals with PH1, the prognosis is less predictable due to the variability in symptoms and age of onset and the prognosis can often be poor, particularly for those individuals who have symptoms at earlier ages. Although combined liver-kidney transplantation can provide a cure for individuals with PH1, these individuals must then face the challenges of life-long use of immunosuppressive medications and other challenges of organ transplants.

801 Broadway Ave NW, Suite 203, Grand Rapids, MI 49504 • Phone: (855) 776-9436 • Fax: (616) 710-4667 info@nxgenmdx.com
nxgenmdx.com

Revised: 4/2022

Treatment

Several treatments are available to manage the symptoms of PH1. One essential therapy for individuals with PH1 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 or D because large amounts can cause stone formation. Approximately 10-30% of individuals with PH1 also respond to treatment with vitamin B6, which has been shown to reduce calcium oxalate levels in the body, and responsiveness has been linked to certain genetic mutations. After a certain point, individuals with PH1 will require dialysis to manage the calcium oxalate build-up in their body. Another major therapy considered for individuals with PH1 is organ transplantation, with three potential strategies: kidney transplant, liver transplant (to restore production of the *AGXT* enzyme), or combined kidney-liver transplant. In individuals with PH1, kidney transplant is usually supplanted by liver transplant because calcium oxalate deposits can quickly re-accumulate in the new kidney. Individuals with PH1 should also regularly monitor the other major organs that can be affected by calcium oxalate deposits.

Resources

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

801 Broadway Ave NW, Suite 203, Grand Rapids, MI 49504 • Phone: (855) 776-9436 • Fax: (616) 710-4667 info@nxgenmdx.com
nxgenmdx.com

Revised: 4/2022