Wilson Disease

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of Wilson disease. Carriers are not expected to show symptoms. You and your partner would both have to be carriers of Wilson disease 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 Wilson disease, each of your children has a 1 in 4 (25%) chance to have the condition.

Wilson Disease Explained

What is Wilson Disease?

Wilson disease is an inherited disorder caused by the inability or decreased ability to transport copper from the liver to other parts of the body or eliminate it from the body. Copper build-up causes damage to many organs and tissues in the body, specifically the brain and liver.

Signs and symptoms can first appear in childhood or may not appear until adulthood. Most individuals with Wilson disease are symptomatic by their teenage years. Liver disease is the most prominent symptom of this disorder. Jaundice (yellowing of the skin), fatigue, loss of appetite, and even swelling of the abdomen are also common. In adulthood, symptoms are more common in the nervous system as opposed to the liver; symptoms can include tremors, difficulty walking and speaking, impaired ability to think, depression, anxiety, and mood swings.

Many individuals with Wilson disease will also experience copper build-up in their eyes, causing a Kayser-Fleischer ring to form. A Kayser-Fleischer ring is a green to brownish ring that surrounds the colored part of the eye, but it does not interfere with vision.

Prognosis

With proper treatment, prognosis for Wilson disease is fair. It is possible to manage this condition for many years after diagnosis. The effect on lifespan is unclear.

Treatment

Medications can help manage Wilson disease. Medications called chelating agents help to release the copper in the body so it can be excreted properly. The next course of action is to help prevent copper from building up again. This is typically done by limiting the amount of copper consumed. Copper is present in multivitamins, liver, shellfish, and nuts. Affected individuals should also avoid copper cooking utensils. If liver damage is extreme, a liver transplant may be necessary.

Resources

Wilson Disease Association
http://www.wilsonsdisease.org/
Genetics Home Reference
https://ghr.nlm.nih.gov/condition/wilson-disease
National Society of Genetic Counselors
https://www.nsgc.org/

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