Clarity Genetics

Genetics Uncoded:

Facts about Wilson Disease

Wilson Disease Explained

Wilson disease is an inherited disorder caused by the inability or decreased ability for transporting copper from the liver to other parts of the body or eliminating 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 generally seen 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.

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 cooper from building up again. This is typically done by limiting the amount of copper consumed. Copper is present in multivitamins, liver, shellfish, and nuts. If liver damage is extreme, a liver transplant may be necessary.

How the Genetics Work

Wilson disease is an autosomal recessive disorder caused by pathogenic variants in the *ATP7B* gene. In general, individuals have two copies of the *ATP7B* gene. Carriers of Wilson disease have a variant in one copy of the *ATP7B* gene while individuals with the disorder have variants in both copies of *ATP7B* gene, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

What Your Test Results Mean

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

Questions?

Contact us at **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.

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