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CARRIER SCREEN

Wilson disease

What is Wilson disease?

Wilson disease is an inherited disorder of copper metabolism characterized by liver disease, movement disorders, and psychiatric problems.[1,2] Individuals with Wilson disease have abnormalities in the enzyme copper-transporting ATPase 2, which removes excess copper from the body.[1,2] The symptoms associated with Wilson disease are due to the buildup of excess copper in the liver, brain, and other tissues. Wilson disease is also known as hepatolenticular degeneration.[1,2]

What are the symptoms of Wilson disease and what treatment is available?

Wilson disease varies in severity and age at presentation, even within families. Onset occurs anytime from early childhood through late adulthood, but it is most common in the teenage years.[1] Symptoms may include:[2]

- Jaundice (yellow color of the eyes and skin)
- Chronic liver disease and/or liver failure
- Dystonia (sustained muscle contractions causing twisting movements and abnormal posture)
- Coordination and walking problems
- Tremors
- Lack of facial movement
- Difficulty swallowing
- Depression, anxiety, and other psychiatric issues
- Intellectual deterioration
- Kayser-Fleischer rings (golden-colored ring around the cornea of the eye)
- Infertility in women, if untreated

There is no cure for Wilson disease and affected individuals require lifelong clinical management. Treatment may include medications to remove copper from the body, nutrition supplements, and dietary restrictions. If individuals do not respond well to treatments, a liver transplant may be considered.[2]

How is Wilson disease inherited?

Wilson disease is an autosomal recessive disease caused by mutations in the *ATP7B* gene.[1] An individual who inherits one copy of an *ATP7B* gene mutation is a carrier and is not expected to have related health problems. An individual who inherits two *ATP7B* mutations, one from each parent, is expected to be affected with Wilson disease.

If both members of a couple are carriers of a mutation in the same gene, the risk of having an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

Who is at risk for Wilson disease?

Wilson disease can occur in individuals of all races and ethnicities. In the Caucasian population, Wilson disease is estimated to affect 1 in 30,000 individuals, with a calculated carrier frequency of approximately 1 in 90.[2] It is more common in individuals of Chinese and Japanese ancestry, affecting approximately 1 in 10,000.[2]

Having a relative who is a carrier or is affected can increase an individual's risk to be a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetics health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

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What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

Where can I get more information?

Wilson Disease Association: http://www.wilsonsdisease.org/

American Liver Foundation: http://www.liverfoundation.org/abouttheliver/info/wilson/

References

1. Wilson disease. *Genetics Home Reference.* Available at: http://ghr.nlm.nih.gov/condition/wilson-disease. Accessed May 8, 2012.
2. Cox, D and Roberts, E. Wilson disease. *GeneReviews.* Available at: http://www.ncbi.nlm.nih.gov/books/NBK1512/. Accessed on: May 8, 2012.