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Genetic Testing for Wilson Disease (WD)

Information for Patients and Families

What do I need to know about testing for Wilson disease?

Wilson disease (WD) is caused by a change in someone's DNA. People with WD can have liver problems, abnormal movements, seizures, and psychological problems. This blood test may prove that you have WD. However, some people with WD will have a negative result, so this test will not rule out WD. There is also a chance that the test will find something that we do not understand. Thus, we may need to test other family members to learn more. This information sheet will provide more details about WD and this testing. Please talk to a genetic counselor, if you have more questions about testing.

What is Wilson disease?

Wilson disease is a rare genetic condition that affects how copper is used in our body. It leads to three main types of symptoms:

- Liver problems—jaundice, hepatitis, cirrhosis, enlarged liver, and anemia
- Neurological problems—tremors, poor coordination, muscle spasms, and seizures
- Psychiatric problems—depression, mood changes, personality changes, and psychosis

Kayser-Fleischer rings are copper deposits that form a ring surrounding the cornea of the eyes. These are only found in patients with WD and are diagnostic for this disease. Patients will start showing signs of WD at 3 to 45 years of age. The symptoms may also vary from one individual to another.

What causes Wilson disease?

WD is caused by a change (mutation) in a gene called *ATP7B*. Genes hold our genetic information and instruct our bodies how to develop and function. We have two copies of every gene. One copy of our genes comes from our mother and the other copy from our father. If there is a change in the gene, the gene may not work properly. A change in the *ATP7B* gene leads to build-up of copper in the liver. Once there is build-up of copper in the liver, it distributes the extra copper throughout our body (especially in the nervous system and eyes).

WD is an autosomal recessive condition. This means that both copies of the gene have a change. A person with only one copy of the changed gene is a carrier of the disease, but has no symptoms.

How can a person be diagnosed with Wilson disease?

A person showing symptoms of WD should be evaluated. A series of tests can confirm this diagnosis.

- Low ceruloplasmin in blood
- High copper in urine
- High copper in liver

It is important to remember that individuals that are carriers for the WD gene do not have symptoms, but they may have some abnormal test results.

Reasons for genetic testing for Wilson disease:

- confirm the diagnosis
- test family members that might be at risk and offer reassurance that other family members are not affected
- test family members that may be carriers of WD
- allow the start of treatment before symptoms manifest
- provide information and counseling for future pregnancies

Is genetic testing available for Wilson disease?

The University of Chicago is currently offering genetic testing for WD. The genetic testing looks at the *ATP7B* gene to find a change in the gene that could lead to the symptoms of WD. This is like reading the sentences of a

book and looking for “typos”. You may read the whole book and miss the “typo,” however when you do find it, then it is easy to test other family members (i.e. you know that the change is on page 875 in the second paragraph). Once a change is found in the person with WD, testing other family members, even during a pregnancy, is easy and fast because we know where to look.

What does it mean if they find a mutation? What does it mean for our family?

Finding a mutation will confirm a diagnosis of WD. Once a change has been identified in an affected individual then it allows for easy testing of other family members, who may choose to be tested.

What if the test is negative?

If an individual was diagnosed with WD based on other tests and the genetic test is negative, it does not rule out WD. A negative test means that no change in the gene sequence was found by the available test methods.

What does it mean if they find a variant of unknown significance?

A small number of patients will be found to have a change in the gene that is not reported in other patients. In this case, we are not sure whether the change in the gene causes WD. If this happens, we recommend testing other family members to get more information.

How can a family member or I be tested?

We recommend that a genetic doctor or genetic counselor help you order the test for your child. If you think that you may have WD, you should make an appointment with someone that works in genetics. This can be made through your local physician or hospital. They can order the testing for WD. If there are any questions about ordering the testing, please ask them to contact The University of Chicago Genetics Services Laboratory. A blood sample is required for testing.

How much does the test cost and will my health insurance cover it?

Cost for sequencing of *ATP7B* is \$1800. Deletion/duplication testing of *ATP7B* costs \$1000. All insurance companies are different, but most of them should cover at least part of the cost of testing. We recommend that you contact your insurance company to learn more about your specific coverage prior to testing. You will want to ask your insurance company what your coverage is for the following CPT (Current Procedural Terminology) codes: 81406 for sequencing and 81405 for deletion/duplication analysis. Insurance companies use these codes to define the method of testing. The University of Chicago or your hospital or referring laboratory will bill your child’s insurance company. You may receive a bill for any amount not covered by your insurance company, i.e. copayment, deductible, etc. If you do not have private medical insurance, we will require payment from you by check or credit card before beginning testing.

When/how will I get the results?

Testing takes approximately 4 weeks. Your physician will be informed of the results as soon as it has been completed. Results, along with a report, will be faxed and mailed to your physician.

Additional Resources:

Wilson’s Disease Association International

Phone: 888-264-1450

www.wilsonsisease.org