



Canadian Liver Foundation  
Fondation canadienne du foie

*Bringing liver research to life*  
*Donner vie à la recherche sur le foie*

## Wilson Disease

### What is Wilson disease?

Wilson disease is a hereditary disease in which excessive amounts of copper accumulate in the body, mainly in the liver. The disease affects approximately one in every 30,000 Canadians. Small amounts of copper are essential to good health. One of the liver's jobs is to maintain the balance of copper in the body. The liver is also the main organ to store copper. In Wilson disease, when its storage capacity is full, copper is released into the blood stream. It then accumulates in various organs such as the brain and the cornea of the eye. This copper overload damages these organs. Left untreated, Wilson disease can be fatal.

### What causes the disease?

Wilson disease is inherited. In order to have the disease, a person must get two defective genes, one from each parent. The liver begins to retain copper at birth and it may take years before symptoms manifest themselves. Having only one defective gene does not lead to Wilson disease.

### What are the symptoms of Wilson disease?

Wilson disease is sometimes difficult to diagnose. Affected individuals may have no symptoms for years. When symptoms develop, they can be subtle. Sometimes symptoms of Wilson disease resemble hepatitis. Alternatively, some individuals have an enlarged liver and spleen and liver test abnormalities. Copper accumulation in the brain can present itself in two ways: (1) as physical symptoms such as slurred speech, failing voice, drooling, tremors or difficulty in swallowing or (2) as psychiatric disorders such as depression, manic behaviour or suicidal impulses.

Very rarely, Wilson disease may cause the liver to fail. This liver failure may be the first sign of having Wilson disease. These individuals require liver transplantation.

### How is the disease diagnosed?

Most people with Wilson disease show signs of liver or neuropsychiatric disease while fairly young. Anyone between 3 and 45 years of age who shows signs of any of the previously mentioned symptoms should be screened for Wilson disease. Any child or teenager with unexplained liver disease should be tested for Wilson disease. Occasionally Wilson disease is not evident until well past middle-age. The diagnosis can be confirmed with blood tests measuring serum copper and ceruloplasmin, a blood protein that contains copper. Measurements of how much copper is excreted in the urine over a 24-hour period are also useful for making the diagnosis. Another diagnostic test for this disease is finding Kayser-Fleischer rings due to accumulation of copper in the cornea, sometimes visible as brown rings around the iris, but usually requiring special eye examination. Despite the copper accumulation, eyesight is not affected. Copper in the bloodstream may cause red blood cells to breakdown, causing anemia or gallstones.



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The gene responsible for Wilson disease has been discovered. Conclusive genetic testing is now available to diagnose affected brothers and sisters before they show disease symptoms. Individuals who are found to have the gene defect can be treated before problems arise.

## **How is the disease treated?**

Once diagnosed, effective treatments are available to treat Wilson disease, even in advanced stages. The toxic concentration of copper in the body must be removed. Reaccumulation of copper must be prevented. This is done with the use of a decoppering agent such as D-penicillamine or with high-dose zinc. These medications are pills, taken 2-3 times per day.

Except for those individuals with liver failure, most people affected by Wilson disease do not require liver transplantation.

## **Can Wilson disease be completely cured?**

The treatment does not correct the fundamental flaw in liver function. Therefore, to prevent reaccumulation of copper in the body, treatment must be continued throughout the person's entire lifetime. Individuals who tolerate the treatment and take it consistently generally enjoy good health and normal life expectancy.

## **What precautions should be taken against this disease?**

Since the gene transmitting the disease is recessively inherited, siblings of an affected individual have a 25 per cent chance of being affected. Therefore, when a new case is diagnosed, all siblings must be screened for the disease. Parents and children of affected individuals should also be screened.

## **What else can we learn about it?**

The Canadian Liver Foundation is proud to have supported research that helped in the discovery of the "Wilson disease gene". Now that the gene defect has been identified, more research is needed to identify all the ways changes in the gene can cause Wilson disease.

*This information is current for September 2014.*