What is Wilson’s disease?
Wilson’s disease is an inherited disease caused by a defective gene. It causes excessive levels of copper to be stored in the brain, liver and other organs.

How does Wilson’s disease first appear and what are the common symptoms?
Some Wilson’s disease symptoms may start in childhood or early adulthood. These neurological symptoms often include:
- Involuntary movements, including abnormal posturing (like dystonia) or tremor
- Difficulty with balance or hand coordination
- Difficulty with speech or swallowing
- Cognitive or behavioral changes
Liver problems are also common in Wilson’s disease, as well as low blood count (anemia), kidney problems, or heart problems.

What causes Wilson’s disease?
Wilson’s disease is inherited. It is caused by the mutation of a specific gene that makes a protein needed to removes excess copper from the body. This causes copper to accumulate in the body, especially the brain, liver and eyes. The damage caused by the excess copper produces the symptoms of Wilson’s disease.

How can Wilson’s disease be diagnosed?
It is important for doctors to watch carefully for symptoms of Wilson’s disease, as it is easily missed. It is mainly diagnosed by blood and urine testing.
Ceruloplasmin is a protein made in your liver that stores and carries copper through your body. A blood test that measures that protein may be used to diagnose Wilson’s disease. But, because that test can sometimes miss Wilson’s, a urine test that measures copper levels is recommended. This test involves collecting urine for an entire 24-hour period. Having this urine test done properly is very important to diagnose Wilson’s disease.
There are other tests often associated with a Wilson’s disease diagnosis, including:
- Eye exams to detect abnormal copper in the cornea
- Liver biopsy to confirm the diagnosis
- MRI brain scan to detect changes associated with Wilson’s disease
- Genetic testing to make a final diagnosis.

Is Wilson’s disease treatable?
Yes. Wilson’s disease is very treatable. Early diagnosis and treatment are important to prevent irreversible damage to the brain and body.

Should I change my diet?
Many common foods include copper and most people get more than they need. If you have Wilson’s disease, it is best to avoid or eat fewer copper-rich foods, including:
- Chocolate
- Nuts
- Shellfish
- Liver

What treatments are available?
There are several types of treatment available for Wilson’s disease. These include:
- Medicines that decrease copper absorption from the gut. These include preparations of zinc. Medications like these take some time to work.
- Medicines that bind to copper in the body. These include d-penicillamine, trientine, and ammonium tetrathio-molybdate. These medications may have serious side effects and are not available in every country.
- Liver transplant may be used in severe cases to normalize copper metabolism.
Medications may also be used to improve Wilson’s disease symptoms, including abnormal movements. Talk with your doctor about which treatment or treatments may be appropriate for you.

Should other family members be tested?
Because Wilson’s disease is a genetic disorder, family members may be at risk. Wilson’s is an autosomal recessive condition. This means that two copies of the abnormal gene are needed; one from the mother and one from the father. The mother and father should be fine, since they have one normal copy. However, brothers and sisters may be at risk. If you are diagnosed with Wilson’s and have siblings, they should be screened for the disease. In rare instances, other family members may also need to be screened. Be sure to ask your doctor or a genetics counselor if your family members should be screened.