Wilson disease

Summary

- Wilson disease is a relatively rare genetic disorder that prevents the body from eliminating copper.
- The build-up of copper damages certain structures including the liver, nervous system, brain, kidneys and eyes.
- Wilson disease is fatal without medical treatment.
- There is no cure, but the condition can be managed.
- Treatment options include medications, chelation therapy and avoiding foods high in copper.

Metabolism refers to the countless and ongoing chemical processes inside the body that allow life and normal functioning. Wilson disease is a rare genetic disorder, an ‘inborn error of metabolism’ that prevents the body from eliminating copper.

The build-up of copper in the body damages certain organs including the liver, nervous system, brain, kidneys and eyes. In around half of cases, only the liver is affected. The copper begins to accumulate at birth, but symptoms appear most commonly during the teenage years. Without treatment, the copper poisoning is fatal. There is no cure, but Wilson disease can be managed.

Around one in 30,000 people have Wilson disease. It is caused by faults in a gene called ATP7B, which must be inherited from both parents for the disease to be expressed. Wilson disease is also known as hepatolenticular degeneration.

Symptoms of Wilson disease

Symptoms of Wilson disease can appear in individuals ranging from three years to over 50 years of age. Symptoms depend on which structures of the body are affected by copper poisoning, but can include:

- enlarged abdomen
- abdominal pain
- loss of appetite
- vomiting of blood
- splenomegaly (enlarged spleen)
- jaundice (yellowed eyes and skin)
- hand tremors
- stiffness and reduced movement of the extremities
- the movement of the body is jerky, slow and difficult to control
- speech difficulties
- personality changes
- symptoms of mental illness, such as depression or homicidal tendencies
- lack of menstruation (amenorrhoea)
- confusion
- dementia.

Cause of Wilson disease

Enzymes are special proteins that help to spark chemical reactions in the body. The removal of excess copper relies on a particular enzyme, but this enzyme is defective in people with Wilson disease. Copper deposits then

build up inside the body and cause damage.

Wilson disease is inherited in an autosomal recessive manner. This means that in order for a person to be affected by Wilson disease, they need to inherit two faulty genes, one from each parent. If both parents carry a faulty gene for Wilson disease, each child has a one in four chance of inheriting both faulty genes and being affected by Wilson disease. If only one faulty gene is inherited, the child is a carrier but won't develop any symptoms.

Most people with Wilson disease have no family history of the disorder.

Effects of copper on the body

Copper is common to a wide range of foods, but the human body needs only tiny amounts. The rest is excreted. In people with Wilson disease, the excess copper leaves the bloodstream and settles in various organs and structures, including the brain, spinal cord, eyes, liver and kidneys.

Copper is a toxic substance in large amounts, and causes damage to your tissues. The damaged tissues die and are replaced by scar tissue. As more and more tissue is replaced by scars, the affected organ loses its ability to function until it eventually fails.

Complications of Wilson disease

Without medical treatment, complications of Wilson disease can include:

- liver disease, such as hepatitis, cirrhosis or necrosis (death of the tissue)
- increased susceptibility to infections
- disorders of the spleen
- anaemia
- muscle atrophy
- increased susceptibility to bone fractures
- permanent physical disability
- permanent intellectual disability
- death.

Diagnosis of Wilson disease

Wilson disease is diagnosed using a number of tests, including:

- physical examination
- medical history
- eye examination to check for Kayser-Fleischer rings (brown rings outside the iris)
- blood tests
- urine tests
- **genetic testing** to detect mutations in the ATP7B gene
- magnetic resonance imaging (MRI)
- biopsy of affected organs, particularly the liver.

Treatment of Wilson disease

Without treatment, Wilson disease is fatal. The longer the copper poisoning continues, the harder it is to successfully treat, so early diagnosis is important. The aims of treatment are to reduce the amount of copper in the body and control the symptoms. Treatment must be lifelong. Death can occur in a matter of months if the treatment is stopped. Options may include:

- vitamin B6 to bolster the nervous system
- potassium supplements, taken before eating, to reduce the absorption of dietary copper
- zinc therapy to prevent the absorption of copper in the small intestine
- switching to a diet low in copper
- chelation therapy, which is the use of medications (such as penicillamine) that bind to copper and allow it to

be excreted in the urine
- regular blood and urine tests to check copper levels so that treatment can be adjusted if necessary
- liver transplant in severe cases
- genetic counselling and testing for the family.

Low copper diet

Management of Wilson disease is lifelong. It is important to switch to a low copper diet. Some of the foods to avoid include:

- chocolate
- dried beans
- dried fruits
- mushrooms
- nuts
- offal such as liver
- peas
- shellfish
- whole wheat products.

Where to get help

- Your **GP (doctor)**
- **Victorian Clinical Genetic Services**
- **Genetic counsellor**

This page has been produced in consultation with and approved by:

Victorian Clinical Genetics Services (VCGS)