

Wilson's disease

Metabolism refers to the countless and ongoing chemical processes inside the body that allow life and normal functioning. Wilson's disease is a relatively rare genetic disorder, called an 'inborn error of metabolism', that prevents the body from eliminating copper. The build-up of copper 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 usually appear during the teenage years. Without treatment, the copper poisoning is fatal. There is no cure, but the condition can be managed.

Around one in 30,000 people have the disorder, with Southern Italians and Eastern Europeans at slightly increased risk. The cause is a faulty gene that must be inherited from both parents. Sometimes, the disorder is caused by a spontaneous gene mutation. Wilson's disease is also known as hepatolenticular degeneration.

Symptoms

Symptoms of Wilson's disease 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.

Flawed genes and defective enzymes

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's disease. Copper deposits then build up inside the body and cause damage. The faulty gene that causes the defective enzyme must be inherited from both parents for the child to develop Wilson's disease. If only one faulty gene is inherited, the child is a carrier but won't develop any symptoms. In many cases, Wilson's disease is caused by a spontaneous gene mutation without any family history of the disorder.

Effects 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 Wilson's 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. 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

Without medical treatment, complications of Wilson's 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 methods

Wilson's 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
- X-rays
- Computerised tomography (CT) scans
- Magnetic resonance imaging (MRI)
- Biopsy of affected organs, particularly the liver.

Treatment options

Without treatment, Wilson's 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's 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 doctor

Things to remember

- Wilson's 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's 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.

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

Royal Children's Hospital - Clinical support services

Content on this website is provided for education and information purposes only. Information about a therapy, service, product or treatment does not imply endorsement and is not intended to replace advice from your doctor or other registered health professional. Content has been prepared for Victorian residents and wider Australian audiences, and was accurate at the time of publication. Readers should note that, over time, currency and completeness of the information may change. All users are urged to always seek advice from a registered health care professional for diagnosis and answers to their medical questions.

For the latest updates and more information, visit www.betterhealth.vic.gov.au

Copyright © 1999/2011 State of Victoria. Reproduced from the Better Health Channel (www.betterhealth.vic.gov.au) at no cost with permission of the Victorian Minister for Health. Unauthorised reproduction and other uses comprised in the copyright are prohibited without permission.