CONDITION

Wilson disease

Wilson disease is an inherited condition that causes copper to build up, mainly in the liver and brain.

WHAT IT IS
Inherited copper buildup affecting the liver and brain

BODY SYSTEM
Liver, nervous system, eyes, blood, and kidneys

MAIN SYMPTOMS
Jaundice, tremor, speech changes, mood or behavior changes

WHAT CAUSES IT
Inherited ATP7B gene changes impairing copper removal

HOW IT IS DIAGNOSED
Copper studies, eye examination, liver or genetic testing

MAIN TREATMENT
Lifelong copper-removing medicines or zinc therapy

CAN IT BE CURED?
No - lifelong copper control remains necessary

IS IT AN EMERGENCY?
Sometimes - sudden liver failure or severe confusion

LONG-TERM OUTLOOK
Usually good with early, continuous treatment

Overview

Wilson disease is a rare inherited condition in which the body cannot remove extra copper properly. It is also called hepatocerebral degeneration.

Copper is a mineral needed in small amounts. It helps the body make energy, maintain nerves, and form connective tissue. Most extra copper normally passes from the liver into bile, a digestive fluid, and then leaves the body in stool.

In Wilson disease, this process does not work correctly. Copper first builds up in the liver. Over time, it can enter the bloodstream and collect in the brain, eyes, kidneys, and other tissues.

This buildup may cause liver disease, movement problems, speech or swallowing difficulties, and changes in mood or behavior. Some people have no clear symptoms when the condition is

found through family testing.

Wilson disease can cause serious organ damage if it is untreated. However, treatment can remove extra copper and limit further buildup. Treatment usually needs to continue for life, even when symptoms and test results improve.

Symptoms

Symptoms differ according to where copper has collected. They can begin in childhood or adulthood, and liver and nervous system symptoms may occur together.

Liver-related symptoms may include:

Some people first develop fatty liver, ongoing liver inflammation, or cirrhosis. Cirrhosis means permanent scarring that changes the liver's structure and function. Occasionally, Wilson disease causes acute liver failure, in which liver function worsens quickly.

Brain and nerve symptoms may include tremor, muscle stiffness, slow or uncontrolled movement, poor coordination, and problems with balance. Handwriting may change. Speech can become quiet, slurred, or difficult to understand. Some people have trouble chewing, swallowing, or controlling saliva.

Mood, thinking, and behavior may also change. Possible symptoms include depression, anxiety, irritability, impulsive behavior, difficulty concentrating, or a drop in school or work performance. These symptoms have many possible causes and do not by themselves mean that someone has Wilson disease.

Copper may form a greenish-brown ring around the colored part of the eye. This is called a Kayser-Fleischer ring. It is usually detected during a specialist eye examination and may not be visible in an ordinary mirror.

Less common effects include anemia caused by the rapid breakdown of red blood cells, kidney problems, joint or bone symptoms, and menstrual changes.

- Tiredness, poor appetite, nausea, or abdominal discomfort
- Yellow skin or eyes, called jaundice
- Dark urine or unusually pale stool
- Swelling in the abdomen or legs
- Easy bruising or bleeding

Causes and risk factors

Wilson disease is caused by harmful changes in a gene called ATP7B . This gene gives the body instructions for moving copper out of liver cells and into bile.

The condition is inherited in an autosomal recessive pattern. This means a person usually has a harmful ATP7B change in both copies of the gene, one inherited from each biological parent.

Parents who each carry one changed copy usually do not have Wilson disease. When both parents are carriers, each pregnancy has:

These chances are the same for each pregnancy. They do not mean that exactly one child in every four-child family will have the condition.

Brothers and sisters of someone with Wilson disease have an increased chance of being affected, including before symptoms appear. Screening close biological relatives is therefore an important part of care.

Wilson disease is not caused by eating an ordinary diet containing copper. Food may affect copper intake after the condition develops, but it does not cause the inherited problem.

- A 1 in 4 chance of a child having Wilson disease
- A 1 in 2 chance of a child being an unaffected carrier
- A 1 in 4 chance of a child inheriting neither changed copy

Diagnosis

No single routine test identifies every case. Diagnosis usually combines medical history, examination findings, copper-related tests, and sometimes genetic or liver testing.

Blood tests may measure ceruloplasmin , a protein that carries copper. It is often low in Wilson disease, but it can be normal in some affected people. Low levels can also occur for other reasons. Total blood copper can be difficult to interpret because much of it is attached to ceruloplasmin.

A 24-hour urine test measures how much copper leaves the body in urine over a full day. The result can support the diagnosis and may later help monitor treatment.

An eye specialist may use a slit lamp, a microscope with a bright light, to look for Kayser-Fleischer rings. These rings are common when the brain and nervous system are affected, but their absence does not rule out Wilson disease.

Other testing may include:

A liver biopsy involves removing a small tissue sample with a needle. It is not needed for

everyone.

Clinicians may use a scoring system that combines these findings. Testing close relatives can identify affected family members before organ damage or symptoms develop.

- Liver blood tests and a full blood count
- Genetic testing for harmful ATP7B changes
- A liver biopsy to measure copper in liver tissue when the diagnosis remains unclear
- Brain MRI when movement, speech, mood, or thinking symptoms are present

Treatment and management

Treatment has two goals: remove excess copper already in the body and prevent it from building up again. It is usually coordinated by a liver specialist, sometimes with a neurologist and other professionals.

Chelating medicines bind to copper so that more of it can leave through urine. Examples include penicillamine and trientine. They may be used as initial treatment, particularly when there are symptoms or organ injury.

Zinc therapy reduces the amount of copper absorbed from food in the intestine. It may be used in some people without symptoms, as maintenance treatment after copper levels have fallen, or when selected by the specialist for another clinical reason.

The choice of medicine depends on symptoms, liver function, previous treatment, pregnancy plans, side effects, and test results. These medicines have different timing and monitoring requirements. They should be taken exactly as directed.

Neurological symptoms can sometimes worsen when copper-removing treatment begins, even though the treatment is lowering body copper. New or worsening movement, speech, or swallowing problems should be reported promptly. A specialist can assess whether treatment needs to be adjusted.

Regular monitoring may include blood counts, liver and kidney tests, and urine copper measurements. Monitoring helps show whether treatment is working and checks for side effects or excessive copper removal. Too little copper can also cause health problems, including anemia and nerve symptoms.

Diet is an added support, not a replacement for medicine. A specialist or dietitian may advise limiting foods that contain large amounts of copper, especially early in treatment. These can include liver, shellfish, some nuts, chocolate, and certain mushrooms. The aim is not to remove all copper from the diet.

If household water may contain excess copper, such as from private wells or copper plumbing, the care team may recommend testing it.

Liver transplantation may be needed for acute liver failure or severe liver disease that does not respond to medical treatment. A transplanted liver can restore normal copper handling, but transplantation involves major surgery and lifelong medicines to prevent organ rejection.

Physical therapy, occupational therapy, speech and swallowing therapy, and mental health care can help manage lasting symptoms and support everyday activities.

Complications

Untreated copper buildup can lead to cirrhosis, liver failure, or lasting damage to areas of the brain that control movement and coordination.

Cirrhosis may cause portal hypertension , meaning increased pressure in veins that carry blood through the liver. This can lead to abdominal fluid, an enlarged spleen, or swollen veins in the digestive tract that may bleed.

Acute liver failure can also cause anemia from rapid red blood cell breakdown, abnormal bleeding, kidney injury, and changes in alertness.

Neurological complications may include persistent tremor, stiffness, difficulty walking, speech problems, swallowing problems, and reduced independence. Depression, anxiety, and behavior changes can affect relationships, education, work, and treatment adherence.

Treatment reduces these risks, but it can also cause side effects. The specific risks differ between medicines. Regular blood, urine, liver, and kidney testing is therefore part of long-term management.

Prevention

Wilson disease cannot be prevented because it is inherited.

Early family screening can prevent or reduce organ damage by finding the condition before symptoms appear. Biological siblings of an affected person should usually be offered testing. Parents, children, and other close relatives may also need assessment based on the family pattern.

Genetic counseling can explain test results, carrier status, and the chance of passing an ATP7B change to a child. It can also discuss reproductive testing options without directing a personal decision.

For someone who has Wilson disease, continuous treatment helps prevent copper from building up again. Stopping medicine because symptoms have improved does not prevent recurrence.

Living with it and what to expect

Wilson disease is a lifelong condition, but many people can study, work, have relationships, and remain active when treatment is started early and continued.

Liver tests may improve over months after treatment begins. Jaundice, swelling, and other liver symptoms may also improve, although established cirrhosis may not fully reverse. People with cirrhosis usually need ongoing liver monitoring even when copper control is good.

Movement, speech, and coordination symptoms often improve more slowly. Recovery may continue over months or years. Some neurological effects may remain, especially when there was substantial damage before treatment. Rehabilitation can help with function and safety.

Follow-up remains necessary when you feel well. Appointments are used to review symptoms, test copper control, look for medicine side effects, and discuss whether doses or treatment types need adjustment. Missing treatment for a prolonged period can allow copper to build up again.

Do not stop or change treatment on your own, including during pregnancy. Wilson disease treatment usually continues during pregnancy because uncontrolled copper buildup can harm the pregnant person and pregnancy. A specialist may adjust the treatment plan and monitoring.

Mood or behavior changes are part of the condition for some people, not a personal failing. Mental health treatment and practical support can be used alongside copper-lowering therapy.

When to get medical help

Call emergency services or go to an emergency department for:

These symptoms can occur with liver failure, internal bleeding, aspiration, or a mental health emergency.

Contact your medical team promptly for new jaundice, dark urine, increasing abdominal or leg swelling, worsening tremor, new balance problems, or changes in speech or swallowing. Also seek prompt advice for a severe rash, fever, mouth sores, unusual bruising, or marked weakness after starting or changing medicine.

If medicine has been missed, cannot be kept down, or is difficult to take as directed, contact the prescribing team rather than waiting for the next routine visit.

Routine follow-up includes planned blood and urine tests, medicine reviews, and monitoring of liver or neurological symptoms. A new Wilson disease diagnosis in the family is also a reason to arrange non-emergency screening.

- New severe confusion, extreme sleepiness, collapse, or inability to wake normally
- Vomiting blood, passing black tar-like stool, or heavy unexplained bleeding
- Rapidly worsening jaundice with abdominal swelling or confusion
- Sudden severe difficulty swallowing or breathing
- Thoughts of suicide with an immediate risk of acting on them

Common questions

Is Wilson disease curable?

There is no simple cure that removes the inherited gene changes. Lifelong treatment can control copper levels and prevent further buildup. Liver transplantation corrects copper handling by replacing the affected liver, but it is reserved for severe liver failure and does not always reverse existing brain injury.

Can I stop treatment when my copper tests are normal?

Usually not. Normal or improved results often mean that treatment is working. Copper can build up again if treatment is stopped. Any change should be planned with the specialist overseeing your care.

What foods should I avoid with Wilson disease?

You do not usually need a copper-free diet. Your team may recommend limiting especially copper-rich foods, such as liver and shellfish, particularly early in treatment. Advice should account for your medicine, copper test results, and nutritional needs.

Can I drink alcohol?

Alcohol can add to liver injury. Avoiding it is especially important if there is active liver disease or cirrhosis. Your liver team can explain what is safest for your situation.

Will my children inherit Wilson disease?

A child would need to inherit a harmful ATP7B change from both biological parents to have Wilson disease. A person with Wilson disease will pass one changed copy to each biological child, but the child's risk also depends on the other biological parent's genes. Genetic

counseling and carrier testing can clarify this.

Can someone with Wilson disease have a normal life?

Many people have a usual or near-usual life expectancy and remain active with early diagnosis, continuous treatment, and monitoring. The outlook is less favorable when severe liver failure or advanced neurological damage is already present.

Sources

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