CONDITION

Menkes disease

Menkes disease is an inherited condition that prevents the body from using copper normally, mainly affecting the brain and connective tissues.

WHAT IT IS
Inherited disorder of copper transport

BODY SYSTEM
Nervous system, connective tissue, hair, and blood vessels

MAIN SYMPTOMS
Unusual hair, seizures, poor growth, developmental delay

WHAT CAUSES IT
Inherited changes in the ATP7A gene

HOW IT IS DIAGNOSED
ATP7A genetic testing and copper-related blood tests

MAIN TREATMENT
Early injected copper and supportive care

CAN IT BE CURED?
No - treatment can limit some harm

IS IT AN EMERGENCY?
Sometimes - prolonged seizures or breathing difficulty

LONG-TERM OUTLOOK
Often severe; early copper treatment may improve outcomes

Overview

Menkes disease is a rare inherited condition that affects how the body moves and uses copper. Copper is a mineral needed in small amounts for brain development, energy production, healthy blood vessels, strong connective tissue, and normal hair structure.

The condition is caused by a change in the ATP7A gene. This gene gives the body instructions for moving copper out of intestinal cells and delivering it to tissues. When ATP7A does not work properly, the brain and other organs do not receive enough usable copper.

Most affected babies appear healthy at birth. Symptoms often become noticeable during the first few months of life. These may include poor feeding, slow growth, seizures, loss of early developmental skills, and sparse or unusually twisted hair.

Menkes disease is serious because a lack of copper-dependent enzymes can interfere with early brain development. Treatment with an injected form of copper works best when started very early, sometimes before clear symptoms appear. It can improve the course for some children, but it does not fully correct the genetic change.

Symptoms

The symptoms and their severity are not the same in every child. Classic Menkes disease usually begins in early infancy and affects several parts of the body.

Early signs may include:

The hair may break easily and feel coarse. Under a microscope, individual hairs may appear twisted. This finding is called pili torti , but it is not unique to Menkes disease and cannot confirm the condition by itself.

As the condition progresses, some children develop stiff muscles, unusual movements, or difficulty controlling the head and limbs. Feeding and swallowing may become harder. Developmental disability can be profound in the classic form.

Changes in connective tissue can cause loose skin, weak bones, hernias, and unusually flexible joints. Blood vessels may become twisted or fragile. Some children develop bladder outpouchings called diverticula, which can contribute to urinary problems.

There are milder ATP7A-related conditions. These include occipital horn syndrome, which mainly affects connective tissue and may cause bony projections at the back of the skull. People with milder forms may have fewer neurological symptoms and survive into adulthood.

- Poor feeding or difficulty gaining weight
- Low muscle tone, meaning the body feels unusually floppy
- Unexplained low body temperature
- Seizures
- Delayed development or loss of skills
- Sparse, short, pale, or wiry hair

Causes and risk factors

Menkes disease is caused by a harmful change, also called a pathogenic variant, in the ATP7A gene.

The condition usually follows an X-linked inheritance pattern. The ATP7A gene is on the X chromosome. Boys generally have one X chromosome, so a harmful variant in their only copy of ATP7A can cause the condition.

Girls generally have two X chromosomes. If one ATP7A copy has a harmful variant, the other copy often provides enough function. Most female carriers have no major symptoms, although some have areas of unusual hair or mild neurological or connective-tissue features. Rarely, a girl can be more significantly affected because of how her X chromosomes are used in different cells.

A child may inherit the gene variant from a carrier mother. The variant can also arise for the first time in the child, meaning there may be no known family history.

Having a relative with Menkes disease or another ATP7A-related condition increases the chance that other family members carry the same variant. A genetics professional can explain inheritance based on the specific family and test results.

Diagnosis

Early diagnosis matters because copper treatment has the greatest chance of helping before extensive brain injury has occurred.

A medical team may suspect Menkes disease from a combination of early symptoms, unusual hair, and family history. Blood tests often show low copper and low ceruloplasmin , a protein that carries copper in the blood.

Low copper and ceruloplasmin do not confirm the diagnosis by themselves. Healthy newborns can naturally have low levels, and other medical conditions can also affect these results.

Genetic testing that identifies a disease-causing ATP7A variant usually confirms the diagnosis. If a specific variant is already known in the family, a baby can be tested soon after birth, before symptoms develop.

Other tests may help assess how the condition is affecting the child. These can include:

Specialist centres may use additional biochemical tests that examine how copper-dependent enzymes are working. These tests are not available everywhere.

- Hair examination under a microscope
- Brain magnetic resonance imaging, or MRI
- An electroencephalogram, or EEG, to examine seizure activity
- Bone imaging when fractures or skeletal changes are suspected

- Tests of feeding, swallowing, hearing, vision, kidneys, or bladder

Treatment and management

The main condition-specific treatment is copper replacement given by injection. Copper histidinate is a form designed to deliver copper without relying on normal intestinal transport.

Timing is important. Treatment started in the newborn period or soon after diagnosis is more likely to help than treatment begun after significant neurological symptoms have developed. Response is also influenced by the ATP7A variant. Some variants allow a small amount of copper transport and may respond better than variants that leave the protein with no function.

Copper treatment may improve seizure control, growth, muscle tone, development, or survival in some children. It is not a cure, and it may not prevent severe disability. It generally cannot reverse brain injury that has already occurred.

Treatment must be managed by a specialist team. Blood tests and clinical assessments are used to monitor copper status, treatment response, and possible effects on organs such as the kidneys. Ordinary oral copper supplements are not an alternative because the underlying problem is copper transport, not simply a low-copper diet.

Other care is based on the child's needs and may include:

Palliative care can also support comfort, symptom control, and family decision-making. It can be provided alongside copper treatment and other active care; it is not limited to the final stage of illness.

- Medicines to control seizures
- Nutrition support and feeding therapy
- A feeding tube if eating is unsafe or does not provide enough nutrition
- Physical and occupational therapy
- Positioning equipment and mobility support
- Communication, vision, and hearing support
- Treatment of reflux, constipation, infections, or urinary problems
- Bone-health assessment and fracture care

Complications

Neurological complications can include difficult-to-control seizures, severe developmental disability, abnormal muscle tone, and loss of previously gained skills.

Weak or unusually formed blood vessels can raise the risk of bleeding around the brain. This is called a subdural haemorrhage . Bleeding can sometimes follow a minor injury, although it may also occur without an obvious cause.

Weak connective tissue and reduced bone strength may lead to fractures, joint problems, hernias, or changes in the shape of the chest. Twisted arteries can sometimes complicate medical procedures or affect blood flow.

Swallowing problems may allow food or liquid to enter the airway. This is known as aspiration and can contribute to pneumonia. Poor feeding can also cause dehydration and inadequate growth.

Bladder diverticula may prevent the bladder from emptying normally and can increase the risk of urinary infections. Regular monitoring can identify these complications before they cause more serious problems.

Prevention

Menkes disease cannot be prevented through diet, lifestyle, or care during pregnancy. It results from an inherited or newly occurring genetic change.

Genetic counselling can help families understand the chance of the condition occurring again. When a family's ATP7A variant is known, options may include carrier testing for relatives, testing during pregnancy, or testing embryos created through in vitro fertilisation.

Testing an at-risk newborn does not prevent the genetic condition. It can, however, allow copper treatment to begin before symptoms appear, when treatment is most likely to be useful.

Living with it and what to expect

Classic Menkes disease is usually progressive, meaning its effects can become more noticeable over time. Without effective early treatment, many children develop severe neurological disability and have a shortened life expectancy, often because of seizures, infections, breathing problems, or blood-vessel complications.

Early copper treatment can change this course for some children. A child with a partially functioning ATP7A protein may make meaningful developmental progress and live longer than

would otherwise be expected. Other children continue to have major medical and developmental needs despite early treatment.

Regular follow-up commonly involves neurology, genetics, nutrition, rehabilitation, and other specialists. Appointments may focus on seizures, growth, swallowing, movement, communication, kidney function, bladder health, and the effects of copper treatment.

Daily life may include medicines, tube feeding, therapy, mobility equipment, and help with personal care. Families may also need practical support with home nursing, schooling, transport, respite care, and financial services. A care plan shared with emergency services, school, and other caregivers can explain the child's usual symptoms and what to do during a seizure or illness.

The milder ATP7A-related forms have a different course. People with occipital horn syndrome may reach adulthood but can have ongoing problems involving joints, muscles, bladder function, blood vessels, and the autonomic nervous system, which controls automatic functions such as blood pressure.

When to get medical help

Arrange prompt medical assessment if a baby has unusual hair together with poor feeding, slow growth, low muscle tone, developmental concerns, or possible seizures. These signs have many possible causes and do not by themselves mean that the baby has Menkes disease.

If Menkes disease runs in the family, contact the baby's medical or genetics team as soon as possible after birth. Testing before symptoms develop may affect treatment options.

During routine follow-up, report new or worsening feeding problems, vomiting, fewer wet nappies, changes in alertness, increased seizures, possible urinary infection, or signs of pain. New loss of skills should also be assessed.

Seek emergency help for:

Follow the child's existing emergency seizure plan if one has been provided.

- A seizure lasting five minutes or longer
- Repeated seizures without recovery between them
- Trouble breathing, blue or grey lips, or choking that does not clear
- Sudden unresponsiveness or extreme difficulty waking
- A significant head injury or possible bleeding
- Sudden weakness or a major change in movement or awareness

Common questions

Is Menkes disease caused by not eating enough copper?

No. It is caused by a genetic problem with copper transport. The body cannot move copper normally from the intestine to the brain and other tissues. A low-copper diet does not cause the condition.

Can ordinary copper supplements treat it?

Ordinary oral supplements do not correct the underlying transport problem and should not replace specialist treatment. Menkes disease is treated with an injected form of copper under close medical supervision.

Why does Menkes disease change the hair?

Copper is needed by enzymes involved in hair strength, structure, and colour. When these enzymes do not work normally, hair may become sparse, pale, brittle, or twisted.

Can girls have Menkes disease?

They can, but it is uncommon. Girls with one altered ATP7A copy are usually carriers with few or no symptoms. Some develop mild features, and rare girls are more significantly affected.

Is Menkes disease the same as Wilson disease?

No. Both involve copper, but they have opposite problems. Menkes disease prevents enough copper from reaching tissues. Wilson disease causes copper to build up, especially in the liver and brain.

How long can someone live with Menkes disease?

Classic Menkes disease often shortens life, but there is no single timeline. Outlook is influenced by the ATP7A variant, how early treatment begins, treatment response, and complications. Milder ATP7A-related conditions can be compatible with survival into adulthood.

Sources

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