CONDITION

Mucopolysaccharidosis type II

Mucopolysaccharidosis type II is an inherited condition that causes certain complex sugars to build up throughout the body.

WHAT IT IS
Inherited lysosomal condition causing complex sugar buildup

BODY SYSTEM
Whole body, especially airways, bones, heart, and brain

MAIN SYMPTOMS
Joint stiffness, breathing problems, hearing loss, enlarged organs

WHAT CAUSES IT
Inherited change in the IDS gene

HOW IT IS DIAGNOSED
Enzyme activity testing and IDS genetic testing

MAIN TREATMENT
Enzyme replacement and organ-specific supportive care

CAN IT BE CURED?
No - treatment manages effects, not inheritance

IS IT AN EMERGENCY?
Sometimes - severe breathing trouble or new weakness

LONG-TERM OUTLOOK
Lifelong condition; outlook improves with coordinated specialist care

Overview

Mucopolysaccharidosis type II, or MPS II, is a rare inherited condition. It is also called Hunter syndrome.

MPS II affects lysosomes, which are small recycling structures inside cells. A person with MPS II does not make enough working iduronate-2-sulfatase, an enzyme that breaks down certain complex sugars. These sugars, called glycosaminoglycans or GAGs, gradually collect in cells and tissues.

This buildup can affect the airways, ears, heart, liver, spleen, joints, bones, eyes, and nervous system. Signs often develop gradually, so they may not be obvious at birth.

MPS II has a wide range of effects. Some people develop learning, behaviour, and movement

changes because the brain is involved. This is often called neuronopathic MPS II. Other people have little or no decline in thinking skills, sometimes described as non-neuronopathic or attenuated MPS II. These are not completely separate forms; people can fall anywhere along a spectrum.

Early recognition matters because treatment and monitoring may reduce some physical effects and help specialists manage complications. Current treatment does not remove the underlying genetic change or reliably treat effects within the brain.

Symptoms

Symptoms and their timing differ from person to person. Children with more extensive nervous system involvement often develop signs in early childhood. People with an attenuated form may not be diagnosed until later childhood or adulthood.

Possible early signs include:

Bone and joint changes may affect the spine, ribs, hips, knees, hands, and feet. Unlike many forms of arthritis, stiff joints in MPS II usually are not caused by inflammation. Some people develop carpal tunnel syndrome, in which pressure on a wrist nerve causes hand weakness or numbness.

Airway tissues can become thickened, while the chest may be less able to expand. This can contribute to sleep apnoea, repeated infections, and breathing difficulty.

Heart effects can include thickened or leaky heart valves, heart muscle changes, and narrowing of blood vessels. These changes may not cause symptoms at first.

When the nervous system is involved, a child may have delayed speech, difficulty learning new skills, impulsive or highly active behaviour, sleep problems, or a gradual loss of skills already learned. Seizures can occur but are not present in everyone.

People with MPS II usually do not have the marked clouding of the clear front part of the eye that occurs in some other mucopolysaccharidoses. However, other eye and vision problems can still develop.

- Frequent ear, sinus, or chest infections
- Noisy breathing, snoring, or pauses in breathing during sleep
- Hearing loss
- Umbilical or groin hernias
- A larger-than-usual head, tongue, liver, or spleen

- Thick hair, firm skin, and gradually coarser facial features
- Stiff joints, limited movement, or curled fingers
- Slower growth after an initially typical growth pattern

Causes and risk factors

MPS II is caused by a harmful change in the IDS gene. This gene provides instructions for making iduronate-2-sulfatase.

The IDS gene is on the X chromosome. Because males usually have one X chromosome, a harmful change in their only copy usually causes MPS II. Females usually have two X chromosomes, so a working copy often provides enough enzyme. However, females can rarely develop symptoms, sometimes because the working X chromosome is inactive in many cells.

A female carrying an IDS change has a chance of passing it on during each pregnancy. A male with MPS II passes his altered X chromosome to all his daughters and none of his sons. The exact implications for a family are best established through genetic testing and genetic counselling.

Sometimes the genetic change is new in the affected person rather than inherited from a parent. Having no known family history therefore does not rule out MPS II.

Nothing a parent did before or during pregnancy causes the IDS gene change.

Diagnosis

MPS II may be considered when a person has a pattern of airway, hearing, joint, bone, organ, or developmental changes. Because these signs overlap with more common conditions, diagnosis may take time.

A urine test can measure GAG levels or identify the types of GAGs being released. An abnormal result supports the possibility of a mucopolysaccharidosis, but urine testing alone does not confirm MPS II.

Diagnosis is confirmed by finding very low iduronate-2-sulfatase activity in blood cells or another suitable tissue. Laboratories may test another sulfatase enzyme at the same time to rule out a different condition that affects several enzymes.

Genetic testing can identify a harmful change in the IDS gene. It helps confirm the diagnosis, test relatives, and provide information for future pregnancies. In a small number of cases, specialist interpretation or further testing is needed.

After diagnosis, assessments usually examine hearing, breathing during sleep, heart function, vision, joint movement, mobility, growth, and development. Imaging may be used to check the brain, spine, hips, or airway when clinically needed.

Treatment and management

There is currently no cure for MPS II. Care aims to reduce GAG buildup where possible, manage individual symptoms, and preserve movement, breathing, hearing, communication, and independence.

Idursulfase is an enzyme replacement therapy. It supplies a manufactured form of the missing enzyme through an intravenous infusion. It can reduce liver and spleen enlargement and may improve walking capacity, breathing measures, and some other physical features.

Standard intravenous enzyme replacement does not cross the blood-brain barrier in meaningful amounts. The blood-brain barrier protects the brain from substances in the bloodstream. As a result, this treatment is not expected to prevent neurological decline in people with neuronopathic MPS II. It also may not reverse bone, joint, valve, or other changes that are already established.

Infusion reactions can occur, including serious allergic reactions. Infusions are therefore given with appropriate monitoring. The treatment team explains how reactions are prevented, recognised, and managed.

Supportive care is tailored to the organs affected and may include:

Anaesthesia can be higher risk because of a narrow airway, limited neck movement, spinal changes, and heart or lung problems. Planned procedures should involve a team familiar with MPS II and difficult airway management.

Research is continuing into treatments that may reach the brain, including modified enzymes and gene-based approaches. Availability and approval differ by country.

- Hearing aids, ear procedures, or communication support
- Treatment for sleep apnoea and other breathing problems
- Physiotherapy and activities that maintain comfortable movement
- Heart monitoring and treatment of heart complications
- Dental care adapted for limited mouth opening or airway concerns
- Educational, behavioural, and developmental support
- Surgery for selected hernias, joint problems, or nerve compression

Complications

GAG buildup can lead to progressive narrowing of the upper airway, sleep apnoea, reduced lung function, and repeated respiratory infections. Severe airway obstruction is possible.

Heart valve disease, heart muscle thickening, abnormal heart rhythms, and narrowing of coronary arteries may develop. Regular heart reviews can identify changes before they cause obvious symptoms.

Pressure on the spinal cord or nerves can cause pain, weakness, reduced hand function, walking changes, or loss of bladder and bowel control. These problems need prompt assessment.

Other possible complications include significant hearing loss, vision problems, hip damage, reduced mobility, swallowing difficulty, and hydrocephalus. Hydrocephalus is a buildup of fluid around the brain that can increase pressure inside the skull.

People with neurological involvement may gradually lose language, learning, self-care, and movement skills. Behavioural and sleep difficulties can place a heavy practical and emotional burden on the person and their family.

Prevention

MPS II cannot be prevented through diet, exercise, pregnancy care, or lifestyle changes because it is caused by an inherited genetic change.

Genetic counselling can help families understand inheritance and testing options. If the family's IDS gene change is known, carrier testing, testing during pregnancy, or testing embryos created through in vitro fertilisation may be possible. These are personal choices, and availability differs by location.

Early diagnosis cannot prevent the genetic condition, but it may allow treatment and monitoring to begin before some complications become advanced.

Living with it and what to expect

MPS II is lifelong and usually changes over time. The pace and pattern are not the same for everyone.

Physical features such as joint stiffness, reduced growth, hearing loss, and heart or airway changes may progress over years. Treatment may slow or improve some physical effects, but

regular monitoring remains important because not all organs respond equally.

People without significant brain involvement may continue learning, attend mainstream or supported education, work, build relationships, and live into adulthood. They may still need ongoing treatment, mobility support, hearing care, or help with fatigue and pain.

For people with neuronopathic MPS II, development may slow and skills may later be lost. Support often shifts over time toward communication, safe movement, comfort, sleep, nutrition, and help with daily activities. Families may benefit from respite services, psychological support, and palliative care alongside active treatment. Palliative care focuses on comfort and quality of life and is not limited to end-of-life care.

Regular follow-up commonly involves metabolic specialists along with heart, breathing, ear, bone, rehabilitation, dental, and developmental teams. Reviews may include heart scans, hearing tests, sleep studies, movement assessments, and checks for nerve or spinal cord compression.

When to get medical help

Arrange a routine medical review for new or worsening snoring, hearing difficulty, reduced joint movement, hand numbness, pain, falls, changes in walking, or loss of skills. Changes in behaviour, sleep, school performance, swallowing, or exercise tolerance also deserve assessment.

Contact the specialist team promptly if an infusion causes hives, swelling, wheezing, vomiting, dizziness, or unusual breathing symptoms. Follow the treatment centre's instructions for reactions that begin after leaving.

Seek urgent medical help for:

Call emergency services for severe breathing difficulty, blue or grey lips, inability to wake normally, a prolonged seizure, or signs of a severe allergic reaction such as throat swelling or collapse.

- New weakness, difficulty walking, or loss of bladder or bowel control
- Markedly increased sleepiness with headache or repeated vomiting
- Breathing that is becoming difficult or noisy at rest
- Chest pain, fainting, or a new sustained racing heartbeat

Common questions

Is MPS II the same as Hunter syndrome?

Yes. Mucopolysaccharidosis type II, MPS II, and Hunter syndrome are names for the same condition.

Can a girl have MPS II?

Yes, but it is rare. Females may develop MPS II if they have unusual X-chromosome patterns, two affected gene copies, or if the X chromosome carrying the working IDS gene is inactive in many cells. Females with possible symptoms need enzyme and genetic testing rather than assumptions based on sex.

Does MPS II affect intelligence?

It can, but not in everyone. Some people have progressive effects on learning, language, behaviour, and other brain functions. Others have typical intelligence or mild learning needs. Physical severity alone does not always show how much the brain is affected.

Can enzyme replacement therapy cure MPS II?

No. It replaces some missing enzyme and can improve certain physical effects, but it does not correct the IDS gene change. Standard intravenous treatment also has limited access to the brain and may not reverse established tissue damage.

How long can someone with MPS II live?

Life expectancy is highly individual. Progressive brain involvement, airway obstruction, and heart complications can shorten life, sometimes substantially. People with attenuated MPS II may live well into adulthood. Earlier diagnosis and modern respiratory, cardiac, and enzyme treatment have improved care, but they do not remove all risks.

Should relatives be tested?

Testing may be useful for biological relatives, especially females who could carry the family's IDS change and males with possible symptoms. A genetics service can identify which relatives might benefit and which test is most informative.

Sources

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