CONDITION

Hunter syndrome

Hunter syndrome is an inherited condition in which certain complex sugars build up and gradually affect many organs.

WHAT IT IS
Inherited lysosomal storage condition affecting many organs

BODY SYSTEM
Airways, joints, heart, hearing, liver, and brain

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

WHAT CAUSES IT
Inherited deficiency of the IDS enzyme

HOW IT IS DIAGNOSED
Enzyme activity testing and genetic testing

MAIN TREATMENT
Enzyme replacement and supportive specialist care

CAN IT BE CURED?
No - treatment can reduce some effects

IS IT AN EMERGENCY?
Sometimes - severe breathing difficulty needs emergency care

LONG-TERM OUTLOOK
Severe forms shorten life; milder forms often reach adulthood

Overview

Hunter syndrome is a rare inherited condition that affects how the body breaks down certain complex sugars. It is a type of lysosomal storage condition. Lysosomes are small parts of cells that recycle substances the body no longer needs.

People with Hunter syndrome have little or none of an enzyme called iduronate-2-sulfatase, or IDS. Without enough IDS, substances called glycosaminoglycans build up inside cells. This buildup gradually affects tissues and organs throughout the body.

The medical term for Hunter syndrome is mucopolysaccharidosis type II, often shortened to MPS II. The condition mainly affects boys and men, although girls and women can rarely have symptoms.

Hunter syndrome covers a wide range. In some children, it affects brain development and leads to a gradual loss of learned skills. In others, thinking and learning are less affected or not affected, while physical problems continue to develop. There is no sharp boundary between these patterns.

Early recognition matters because treatment and planned monitoring may reduce some physical effects, identify complications sooner, and support development, mobility, hearing, breathing, and quality of life.

Symptoms

Babies with Hunter syndrome often appear healthy at birth. Symptoms usually emerge gradually during childhood, although their timing and severity differ.

Early physical features may include:

The liver and spleen may become enlarged, making the abdomen look prominent. This enlargement does not necessarily mean that these organs are failing.

Joint stiffness can affect the shoulders, elbows, hips, fingers, and other joints. Hands may become short, broad, and curved into a claw-like position. Walking may become more difficult as stiffness, hip problems, nerve compression, and reduced stamina develop.

Hearing loss is common. It may result from repeated middle-ear infections, fluid behind the eardrum, or changes in the hearing nerves. Hearing difficulties can affect speech, learning, and behavior if they are not recognized.

Some children develop delayed speech, attention difficulties, sleep problems, or very active behavior. In more severe forms, development may slow and previously learned skills may gradually be lost. Seizures can occur but are not usually an early feature.

Unlike several related storage conditions, Hunter syndrome does not usually cause obvious clouding of the clear front surface of the eye. However, other eye and vision problems may still occur.

- Frequent ear, sinus, or chest infections
- Noisy breathing, snoring, or pauses in breathing during sleep
- Umbilical or groin hernias
- A larger head, tongue, or abdomen
- Thick hair and gradually coarser facial features
- Ongoing runny nose

- Loose stools or diarrhea
- Stiff joints and reduced movement

Causes and risk factors

Hunter syndrome is caused by a harmful change in the IDS gene. This gene gives the body instructions for making the IDS enzyme.

The gene is on the X chromosome. Most males have one X chromosome, so a harmful change in their only copy of the gene causes the condition. Most females have two X chromosomes. If one carries the altered gene, the other copy often produces enough enzyme to prevent symptoms.

A female carrier can still have symptoms in rare cases. This can happen because of how X chromosomes are switched on and off in different cells or because of an unusual chromosome change.

A carrier has a chance of passing the altered gene to each child. Sons who inherit it are generally affected, while daughters who inherit it are generally carriers, although rare exceptions occur. The gene change can also arise for the first time in a child, so there may be no previous family history.

The condition is not caused by diet, parenting, pregnancy activities, infection, or anything a child or parent did.

Diagnosis

The symptoms of Hunter syndrome overlap with those of more common childhood conditions. Diagnosis often involves several steps.

A urine test can measure glycosaminoglycans. A high level supports the diagnosis, but urine testing alone cannot confirm Hunter syndrome. Some affected people, especially those with milder features, may not have a clearly abnormal screening result.

The diagnosis is confirmed by finding very low IDS enzyme activity in an appropriate blood or cell sample. Laboratories may check another enzyme at the same time to distinguish Hunter syndrome from a different condition that affects several enzymes.

Genetic testing can identify the change in the IDS gene. This confirms the genetic cause and can help with testing relatives, carrier testing, and discussions about future pregnancies.

After diagnosis, assessments usually look at hearing, vision, breathing during sleep, heart

function, lung function, joints, mobility, growth, development, and the brain and spinal cord when needed. These tests establish a baseline for future follow-up.

Testing should be interpreted by a team familiar with lysosomal storage conditions. A low enzyme result from a screening sample may need confirmation before a diagnosis is made.

Treatment and management

There is currently no cure for Hunter syndrome. Care aims to reduce the buildup of glycosaminoglycans where possible, manage complications, preserve function, and support daily life.

Intravenous enzyme replacement therapy supplies a manufactured form of the missing enzyme. Idursulfase is an established enzyme replacement treatment for Hunter syndrome. It is given through a vein at regular intervals in a specialist service.

Enzyme replacement can reduce liver and spleen size and may improve walking endurance, breathing measures, or other physical features. The response is not the same for everyone. It cannot reliably reverse damage that has already developed.

Standard intravenous enzyme replacement does not cross into the brain in meaningful amounts. It therefore has limited ability to prevent or treat the learning and behavioral effects of forms that involve the brain. Availability of particular enzyme products and newer approaches differs by country.

Infusion reactions can include rash, fever, headache, changes in blood pressure, wheezing, or more serious allergic reactions. Treatment teams monitor for these reactions and explain how they are managed.

Supportive care is equally important. It may include:

Anesthesia and sedation require special planning. A narrowed airway, stiff neck, enlarged tongue, and heart or lung problems can make these procedures more complex. They should be planned with clinicians experienced in Hunter syndrome whenever possible.

Regular follow-up usually involves a metabolic or genetics service working with heart, breathing, ear, orthopedic, neurological, therapy, and developmental specialists.

- Hearing aids, ear tubes, or treatment of ear infections
- Treatment for obstructive sleep apnea, in which the airway closes during sleep
- Physical and occupational therapy for mobility and daily activities
- Speech, communication, learning, and behavioral support

- Heart monitoring and treatment of heart complications
- Dental care adapted for limited mouth opening or airway concerns
- Surgery for selected hernias, joint problems, or nerve compression

Complications

Glycosaminoglycan buildup can narrow the upper airway and make the windpipe less flexible. Enlarged tonsils and adenoids, a large tongue, and thickened tissues may contribute to snoring, sleep apnea, repeated infections, and breathing difficulty.

Heart complications can include thickened or leaky valves, heart muscle changes, high blood pressure, and narrowing of blood vessels. These problems may progress without causing obvious early symptoms, which is why routine heart checks matter.

Pressure on nerves can cause hand pain, weakness, or numbness, including carpal tunnel syndrome. Changes around the upper spine may compress the spinal cord. Hydrocephalus, a buildup of fluid around the brain, can also occur.

Other possible complications include worsening hearing loss, reduced vision, limited mobility, chronic pain, swallowing difficulty, dental problems, and reduced lung capacity.

In forms involving the brain, developmental progress may slow before skills are gradually lost. Communication, sleep, behavior, and the ability to carry out everyday activities may change over time.

Prevention

Hunter syndrome cannot be prevented through lifestyle changes because it is inherited.

Genetic counseling can help affected people and relatives understand how the condition is passed on. It can also explain carrier testing and reproductive options without directing a family toward a particular choice.

When the family's IDS gene change is known, testing may be possible before or during pregnancy. Newborn screening for Hunter syndrome is available only in some places and does not replace confirmatory enzyme and genetic testing.

Living with it and what to expect

Hunter syndrome is lifelong and usually changes over time. The rate of change is highly individual, even among relatives with the same gene change.

Children whose brain development is affected may initially gain skills, then reach a plateau and later lose some abilities. Physical complications also progress. These children usually need increasing support with communication, movement, eating, breathing, and personal care. Severe forms can substantially shorten life, often because of breathing or heart complications.

People whose thinking and learning are less affected may attend school, work, form relationships, and live well into adulthood. They can still develop significant joint, hearing, airway, heart, spinal, and mobility problems. Adult care remains important rather than ending when childhood services stop.

Enzyme replacement and supportive care may improve some physical outcomes, but they do not remove the need for ongoing monitoring. Changes may be gradual and easier to detect by comparing hearing tests, heart scans, sleep studies, mobility, breathing measurements, and daily function over time.

School or workplace adjustments may help with hearing, fatigue, mobility, writing, communication, and attendance for treatment. Families may also need practical support with equipment, home access, transport, respite care, or the move from pediatric to adult services.

When to get medical help

Routine follow-up is important even when symptoms seem stable. Contact the usual care team if there is a new decline in hearing, walking, hand strength, sleep, swallowing, behavior, development, or ability to manage daily activities.

Arrange prompt medical review for:

Seek emergency help for severe breathing difficulty, blue or gray lips, prolonged pauses in breathing, sudden inability to wake normally, a seizure lasting several minutes, or sudden weakness or loss of movement.

Before any planned operation, dental procedure with sedation, or scan requiring anesthesia, make sure the team knows about Hunter syndrome. This is a safety-planning issue, even if the procedure seems minor.

- Increasing snoring, choking during sleep, or daytime sleepiness
- New hand weakness, numbness, pain, or frequent dropping of objects
- New balance problems, falls, neck pain, or loss of bladder control

- Difficulty swallowing, repeated choking, or recurrent chest infections
- Fainting, chest discomfort, reduced stamina, or a racing heartbeat
- Persistent headache, vomiting, or an unexplained change in alertness

Common questions

Is Hunter syndrome always severe?

No. It forms a spectrum. Some people develop progressive effects on learning and brain function, while others mainly have physical complications. A gene result alone does not always predict the exact course.

Can girls have Hunter syndrome?

Yes, but it is rare. Girls are more often carriers because they have a second X chromosome with a working copy of the gene. Unusual X-chromosome activity or chromosome changes can sometimes cause symptoms.

Is Hunter syndrome contagious?

No. It is caused by an inherited gene change and cannot spread between people.

Does enzyme replacement cure Hunter syndrome?

No. Enzyme replacement may improve some physical features and slow some effects, but it does not remove the genetic cause. Standard intravenous treatment has limited effect on the brain.

Why does Hunter syndrome affect so many organs?

The missing IDS enzyme is normally used by cells throughout the body. When complex sugars are not broken down properly, they build up in many tissues rather than in one organ.

Can someone with Hunter syndrome live into adulthood?

Yes, especially when brain development is less affected. However, ongoing heart, airway, joint, hearing, and spinal problems can be serious. People with more severe neurological involvement often have a shorter lifespan.

Sources

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