CONDITION

Lesch-nyhan syndrome

Lesch-Nyhan syndrome is an inherited condition causing excess uric acid, movement problems, and compulsive self-injury.

WHAT IT IS
Inherited condition affecting movement, behavior, and uric acid

BODY SYSTEM
Nervous system, kidneys, joints, and urinary system

MAIN SYMPTOMS
Movement disorder, self-injury, developmental delay, excess uric acid

WHAT CAUSES IT
Inherited HPRT1 gene change causing HPRT enzyme deficiency

HOW IT IS DIAGNOSED
HPRT enzyme testing and HPRT1 genetic testing

MAIN TREATMENT
Uric acid control, symptom treatment, protective multidisciplinary care

CAN IT BE CURED?
No - the inherited gene change remains

IS IT AN EMERGENCY?
Sometimes - with choking or serious injury

LONG-TERM OUTLOOK
Lifelong disability; many survive into adulthood with comprehensive care

Overview

Lesch-Nyhan syndrome is a rare inherited condition that affects how the body recycles substances called purines. Purines are natural parts of cells and food. When the body breaks them down, it produces uric acid.

People with Lesch-Nyhan syndrome lack enough of an enzyme called hypoxanthine-guanine phosphoribosyltransferase, usually shortened to HPRT. An enzyme is a protein that helps a chemical process happen. Without enough HPRT, the body produces too much uric acid.

The condition also affects brain function. It causes severe problems with movement and muscle control, developmental disability, and a distinctive pattern of compulsive self-injury. These neurological and behavioral features are not simply caused by high uric acid, so lowering uric acid does not correct them.

Lesch-Nyhan syndrome mainly affects boys and men because it is linked to the X chromosome. Symptoms usually become clear during infancy or early childhood. The condition is lifelong and requires coordinated medical, dental, therapy, behavioral, and caregiving support.

Some people have partial HPRT deficiency. They may have excess uric acid and movement problems without the full pattern of Lesch-Nyhan syndrome.

Symptoms

Babies with Lesch-Nyhan syndrome may appear healthy at birth. Early signs often include unusually low muscle tone, delayed head control, or difficulty reaching movement milestones.

Movement problems become more noticeable over time. They can include dystonia, meaning sustained muscle tightening that twists the body into difficult positions. Other movements may resemble those seen in cerebral palsy, including stiffness, sudden jerks, or uncontrolled writhing movements.

Common features include:

Self-injury often begins between ages 2 and 4, although timing differs. Lip and finger biting are especially common. A person may also strike their head or limbs against nearby objects.

This behavior is not a sign that the person wants to be hurt. People may be distressed by it and feel relieved when safely prevented from injuring themselves. Anxiety, excitement, illness, unfamiliar situations, and reduced physical protection can make the behavior harder to control.

Orange or rust-colored crystals may appear in a baby's diaper because of uric acid in the urine. Excess uric acid can later form stones in the kidneys or urinary tract. It can also collect in joints and cause gout, which produces sudden pain, swelling, warmth, and redness.

Understanding may be better than speech or physical abilities suggest. Movement and communication difficulties can make it hard for a person to show what they know.

- Delayed sitting, crawling, walking, or speech
- Severe difficulty controlling voluntary movement
- Muscle stiffness, spasms, or twisting movements
- Difficulty swallowing, eating, or speaking
- Compulsive biting, hitting, scratching, or other self-injury
- Kidney stones, blood in the urine, or gout caused by excess uric acid

Causes and risk factors

Lesch-Nyhan syndrome is caused by a harmful change in the HPRT1 gene. This gene provides instructions for making the HPRT enzyme.

The condition follows an X-linked inheritance pattern. Boys and men have one X chromosome, so one altered copy of HPRT1 can cause the condition. Girls and women have two X chromosomes and are usually unaffected carriers because their other copy often produces enough enzyme.

When a mother carries an altered HPRT1 gene, each pregnancy has a separate chance of inheriting that gene. A son who inherits it is expected to be affected. A daughter who inherits it will usually be a carrier, although females with symptoms have rarely been reported.

Some cases result from a new genetic change rather than one inherited from a parent. Having no known family history therefore does not rule it out.

The condition is not caused by parenting, diet, infection, or anything done during pregnancy. Diet may influence uric acid levels, but it does not cause the underlying enzyme deficiency or neurological symptoms.

Diagnosis

A clinician may suspect Lesch-Nyhan syndrome when a child has delayed development or abnormal movements together with unusually high uric acid. Self-biting or another typical pattern of self-injury can strengthen that suspicion.

Blood and urine tests can measure uric acid. A urine uric acid-to-creatinine ratio may help show excessive uric acid production. These findings alone do not confirm the condition because uric acid levels can be abnormal for other reasons.

Confirmation usually involves one or both of the following:

Kidney imaging may be used to look for stones or kidney damage. Other testing may assess swallowing, nutrition, movement, communication, hearing, dental injuries, and learning needs.

Once the family's genetic change is known, carrier testing and testing during a future pregnancy may be possible. A genetics professional can explain the available options and what each result can and cannot show.

- An enzyme test showing very low HPRT activity in blood cells or other tissue
- Genetic testing finding a disease-causing change in the HPRT1 gene

Treatment and management

There is no treatment that replaces HPRT throughout the body or removes the underlying genetic change. Care focuses on controlling excess uric acid, reducing injury, managing movement symptoms, and supporting daily life.

Allopurinol is commonly used to reduce the production of uric acid. It helps prevent gout, kidney stones, and kidney damage, but it does not improve the neurological or behavioral features of the condition. Treatment requires monitoring because lowering uric acid too far can encourage a different type of crystal, called xanthine, to form.

Adequate fluid intake may help reduce urinary crystals and stones. Nutrition and hydration plans may need adjustment when swallowing is difficult or fluid intake is limited. Kidney or urinary stones sometimes need specialist procedures.

Movement symptoms may be treated with medicines aimed at muscle stiffness, spasms, anxiety, or dystonia. Physical and occupational therapy can support positioning, comfort, joint movement, equipment use, and safer ways to take part in daily activities. Speech and language therapy can help with communication and swallowing.

Managing self-injury usually requires several approaches. These may include:

Physical restriction is sometimes needed to prevent immediate injury, but it should be planned carefully, monitored, and use the least restrictive safe approach. Removing teeth has been used in severe cases when other protection has failed, but this is a major and irreversible decision.

Communication devices can help someone express choices, discomfort, and emotions even when speech is difficult. Educational plans should not assume that severe movement problems mean a person cannot understand.

- Padded surroundings and carefully designed protective equipment
- Dental guards or other oral protection
- Consistent behavioral support and predictable routines
- Medicines for associated anxiety or severe behavior
- Close attention to pain, illness, sleep, and environmental triggers

Complications

Persistently high uric acid can cause kidney stones, urinary blockage, kidney damage, and gout. Good uric acid control greatly reduces these risks, although monitoring remains

important.

Dystonia and reduced mobility can lead to painful joint stiffness, abnormal spinal curvature, hip problems, pressure injuries, and difficulty with personal care. Poor coordination of swallowing can cause weight loss, dehydration, choking, or aspiration, in which food or fluid enters the airway.

Repeated biting or striking may cause wounds, infection, scarring, damaged teeth, or injuries to the eyes and head. Protection needs may change as a child grows or movement patterns change.

Breathing infections, including pneumonia related to aspiration, can become serious. Reduced mobility and feeding difficulties may add to health risks over time.

Prevention

Lesch-Nyhan syndrome cannot be prevented through diet, exercise, vaccination, or pregnancy care because it is caused by a genetic change.

Genetic counseling can help people with a personal or family history understand inheritance. If the family's HPRT1 change is known, options may include carrier testing, prenatal testing, or testing embryos created through in vitro fertilization. These are personal choices rather than ways to prevent symptoms in someone who already has the condition.

Complications can often be reduced through uric acid treatment, hydration support, kidney monitoring, safe swallowing plans, skin and dental care, appropriate equipment, and early attention to injuries.

Living with it and what to expect

Lesch-Nyhan syndrome is lifelong. Movement disability is usually severe, and most affected people need help with mobility, eating, dressing, hygiene, and protection from injury. A wheelchair and customized seating are often needed.

The urge to self-injure may remain over many years. Its frequency and form can change with age, health, stress, surroundings, and the protection available. It should not be treated as deliberate misconduct. Calm, consistent support is usually more helpful than punishment.

Communication may improve with therapy and assistive technology. Some people understand much more than they can express through speech or movement. Access to education, social contact, recreation, and choices in daily life remains important.

Regular follow-up commonly includes checks of uric acid, kidney function, urine, nutrition, swallowing, dental health, skin, joints, posture, and protective equipment. Routine appointments are also a chance to review pain, sleep, caregiver needs, and changes in behavior.

The condition can shorten life because of complications such as aspiration, infection, or kidney problems. However, many people live into adulthood, especially with effective uric acid control and comprehensive support. No single timeline can predict one person's course.

Families often coordinate care across neurology, kidney care, genetics, rehabilitation, dentistry, nutrition, mental health, and primary care. Respite services and practical support can also be important because supervision needs may be continuous.

When to get medical help

Arrange routine medical follow-up for changes in movement, behavior, eating, sleep, urine, or comfort. New self-injury may be a sign of pain, illness, a poorly fitting device, or another source of distress.

Seek prompt medical advice for:

Get emergency help for choking, severe breathing difficulty, blue or gray lips, loss of consciousness, or an injury with heavy bleeding. Emergency assessment is also appropriate for a serious head or eye injury, inability to pass urine with severe pain, or signs of severe dehydration.

- New joint pain, redness, or swelling
- Blood in the urine or pain during urination
- Repeated vomiting, poor fluid intake, or fewer wet diapers
- Coughing during meals, weight loss, or worsening swallowing
- Wounds showing spreading redness, warmth, swelling, or discharge
- A sudden increase in self-injury or muscle spasms

Common questions

Is Lesch-Nyhan syndrome a type of cerebral palsy?

No. Its movement problems can resemble cerebral palsy, but Lesch-Nyhan syndrome has a specific genetic cause and causes excess uric acid. Genetic or HPRT enzyme testing can

confirm it.

Why do people with Lesch-Nyhan syndrome hurt themselves?

Self-injury is a compulsive neurological symptom of the condition. It is not attention-seeking, poor behavior, or a wish to be harmed. Many affected people are upset by the behavior and welcome safe protection.

Can lowering uric acid stop the movement and behavior symptoms?

No. Uric acid treatment can prevent gout, stones, and kidney damage, but it does not correct the brain changes responsible for dystonia and self-injury.

Can girls have Lesch-Nyhan syndrome?

It is much more common in boys because the altered gene is on the X chromosome. Girls are usually carriers without the full condition, but symptomatic females have rarely been reported.

Can someone with Lesch-Nyhan syndrome understand what is happening?

Often, yes. Understanding may be stronger than speech, hand control, or facial expression suggests. Communication aids and enough response time can help show a person's abilities and preferences.

Is there a cure or gene therapy?

There is currently no established cure. Research is exploring the brain mechanisms and possible genetic or cell-based treatments, but present care focuses on preventing complications and supporting safety, comfort, communication, and participation.

Sources

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