CONDITION

Hartnup disorder

Hartnup disorder is an inherited condition that affects how the intestines and kidneys handle certain amino acids.

WHAT IT IS
Inherited difficulty absorbing and reusing certain amino acids

BODY SYSTEM
Digestive system, kidneys, skin, and nervous system

MAIN SYMPTOMS
Sun-sensitive rash, poor coordination, tremor, confusion

WHAT CAUSES IT
Inherited SLC6A19 gene changes

HOW IT IS DIAGNOSED
Urine amino acid testing, confirmed by genetic testing

MAIN TREATMENT
Nutritional support, vitamin replacement, sun protection

CAN IT BE CURED?
No - inherited gene changes remain

IS IT AN EMERGENCY?
Sometimes - sudden severe confusion or loss of coordination

LONG-TERM OUTLOOK
Usually normal life with effective symptom prevention

Overview

Hartnup disorder is a rare inherited condition that changes how the body transports certain protein building blocks called neutral amino acids. It mainly affects their absorption from the intestines and their recovery by the kidneys.

One important affected amino acid is tryptophan. The body uses tryptophan to make niacin, also called vitamin B3. Niacin helps the skin, digestive system, and nervous system work normally. When too little tryptophan is available, some people develop symptoms similar to niacin deficiency.

Possible symptoms include a rash on sun-exposed skin and short episodes of poor balance, shaking, confusion, or changes in mood and behaviour. However, many people with the genetic changes never develop noticeable symptoms. Others have episodes only during childhood or

at times of illness, poor nutrition, or heavy sun exposure.

Hartnup disorder does not usually cause ongoing kidney failure or steadily worsening damage to the nervous system. With adequate nutrition, sun protection, and treatment when needed, the long-term outlook is generally good.

Symptoms

Symptoms often begin in childhood, but they can first appear later. They may come and go rather than being present every day. Some people are diagnosed through testing even though they have never had symptoms.

The characteristic skin problem is a red, scaly, or blistering rash after sun exposure. It may resemble a severe sunburn. It most often affects the face, neck, hands, arms, or legs. The skin may later become darker or rougher.

Nervous system symptoms can include:

Digestive symptoms, including diarrhoea, may occur but are less central to the condition. Poor nutrition or reduced appetite can make symptoms more likely because the body has fewer alternative sources of amino acids and niacin.

An episode may last for days or weeks. Symptoms can be triggered or worsened by strong sunlight, fever, infection, diarrhoea, emotional or physical stress, or a diet that does not provide enough protein and other nutrients.

These symptoms are not specific to Hartnup disorder. A new rash, loss of coordination, or confusion can have many other causes and needs appropriate medical assessment.

- Unsteady walking or poor coordination, known as ataxia
- Shaking or tremor
- Muscle stiffness or unusual movements
- Headache, dizziness, or weakness
- Confusion or difficulty concentrating
- Changes in mood, behaviour, or perception

Causes and risk factors

Hartnup disorder is caused by changes in a gene called SLC6A19 . This gene provides instructions for a transport protein that helps move neutral amino acids through cells in the

small intestine and kidneys.

When the transporter does not work normally, the intestines may absorb less of these amino acids from food. The kidneys also allow unusually large amounts to pass into the urine instead of returning them to the blood. This pattern is called neutral aminoaciduria.

Hartnup disorder has an autosomal recessive inheritance pattern. This means a person usually needs to inherit an altered copy of the gene from both biological parents. Parents who carry one altered copy generally do not have the condition.

When both parents are carriers, each pregnancy has:

These chances are the same for each pregnancy. Having the genetic changes does not predict how severe symptoms will be. Even people from the same family may have different experiences.

- A 1 in 4 chance of producing a child with Hartnup disorder
- A 1 in 2 chance of producing a child who is a carrier
- A 1 in 4 chance of producing a child with neither altered copy

Diagnosis

Hartnup disorder may be considered when someone has a sun-sensitive rash together with episodes affecting balance, movement, or thinking. It may also be found when a urine test shows an unusual amino acid pattern.

Urine amino acid testing is central to diagnosis. It can show high levels of several neutral amino acids, including tryptophan. The pattern helps distinguish Hartnup disorder from conditions that cause a broader loss of amino acids through the kidneys.

Genetic testing can identify disease-causing changes in both copies of the SLC6A19 gene and confirm the diagnosis. Testing may also help relatives understand whether they have the condition or carry one altered gene copy.

Other tests may be used to check nutritional health or rule out other explanations. These can include blood tests, a skin assessment, and a neurological examination of strength, reflexes, coordination, and walking. Brain imaging is not usually what confirms Hartnup disorder, but it may be needed when symptoms are sudden or do not fit the expected pattern.

Treatment and management

People who have no symptoms may not need active medical treatment. Care is based on nutrition, prevention of known triggers, and treatment of symptoms if they occur.

A balanced diet with adequate protein can provide enough amino acids through alternative absorption pathways. A dietitian can help if eating is restricted, growth is a concern, or another health condition affects nutritional needs. Large dietary changes or protein supplements should not be started without advice, especially when kidney or liver problems are also present.

Nicotinamide, a form of vitamin B3, may be used for people with symptoms related to niacin deficiency. It can help treat or prevent the rash and neurological episodes. The appropriate form and amount should be decided by the treating healthcare team rather than taken as a high-dose over-the-counter supplement.

Protecting the skin from ultraviolet light is also important. Useful measures include broad-spectrum sunscreen, protective clothing, shade, and avoiding prolonged exposure when sunlight is strongest. These steps are particularly relevant for anyone who has previously developed the typical rash.

During an episode, care may include treating an infection or diarrhoea, restoring adequate food and fluids, and reducing sun exposure. Physical therapy or other rehabilitation may be useful if balance or movement problems take time to settle.

Complications

Most people do not develop permanent complications. Episodes commonly improve when the nutritional shortage or other trigger is corrected.

A severe rash can be painful and may blister. Scratching or broken skin can lead to a secondary skin infection. Repeated inflammation may leave temporary changes in skin colour.

Poor coordination can increase the risk of falls and injuries. Confusion or marked changes in behaviour can also affect safety during an episode.

Severe or prolonged nutritional deficiency may cause more persistent neurological problems, although this is uncommon when the condition is recognised and managed. Hartnup disorder itself does not usually cause progressive intellectual decline, chronic kidney failure, or a shortened lifespan.

Not every new neurological symptom should be assumed to be part of Hartnup disorder. Stroke, infection, medication effects, and other conditions can cause similar symptoms and may require urgent treatment.

Prevention

The inherited gene changes that cause Hartnup disorder cannot be prevented or removed through lifestyle changes.

Symptoms and episodes can often be prevented or reduced. The main measures are maintaining adequate nutrition, using reliable sun protection, and getting early care for illnesses that interfere with eating or cause prolonged vomiting or diarrhoea.

Genetic counselling can explain inheritance, carrier testing, and reproductive testing options. It may be useful for adults with the condition, parents of an affected child, or relatives who want to understand their chance of carrying an altered gene.

Living with it and what to expect

Hartnup disorder is lifelong, but the amino acid changes found in urine do not necessarily mean that symptoms will continue. Many people remain symptom-free. In those who have episodes during childhood, attacks often become less frequent with age, although they can still occur later.

Most people can attend school, work, exercise, travel, and have families. Everyday management usually centres on eating regularly, getting adequate protein and other nutrients, and protecting sun-exposed skin. A medical identification card or a brief record of the diagnosis may help if a sudden episode leads to emergency assessment.

Routine follow-up may include reviewing nutrition, growth in children, skin symptoms, and any episodes involving movement or thinking. Follow-up is also a chance to review whether vitamin replacement is still needed. Testing schedules differ according to symptoms and overall health.

A return of the rash or mild coordination problems should not be ignored, but it does not always mean that the condition is worsening. It may signal an infection, nutritional problem, or increased sun exposure that can be addressed.

The outlook is generally favourable with appropriate management. No one course can be guaranteed, so persistent, changing, or unusual symptoms should be reassessed rather than automatically linked to Hartnup disorder.

When to get medical help

Arrange a routine medical appointment if you or your child has:

Seek urgent medical advice for a rapidly worsening rash, blistered or infected skin, repeated vomiting or diarrhoea that prevents normal eating, or a clear increase in unsteadiness or confusion.

Call emergency services for sudden severe confusion, collapse, a seizure, inability to stand or walk, one-sided weakness, difficulty speaking, or reduced consciousness. These symptoms may have causes other than Hartnup disorder and need immediate assessment.

- Repeated sun-sensitive rashes
- Ongoing concerns about nutrition, appetite, or growth
- Mild recurring tremor or balance problems
- Questions about family testing or genetic counselling
- Side effects or concerns related to vitamin supplements

Common questions

Is Hartnup disorder the same as pellagra?

No. Pellagra is a syndrome caused by severe niacin deficiency. Hartnup disorder can produce pellagra-like skin and neurological symptoms because reduced tryptophan absorption may limit the body's niacin supply. Many people with Hartnup disorder never develop pellagra-like symptoms.

Can Hartnup disorder be cured?

The inherited gene changes cannot currently be cured. However, symptoms can often be prevented or controlled with adequate nutrition, vitamin replacement when needed, and protection from strong sunlight.

Will I always have amino acids in my urine?

The characteristic urine findings are usually lifelong because they result from how the kidneys transport amino acids. The abnormal test does not necessarily mean that symptoms or kidney damage are present.

Does Hartnup disorder cause intellectual disability?

Most people have typical intellectual development. Confusion, mood changes, or difficulty concentrating can occur during an episode, but these are often temporary. Persistent developmental or learning concerns need a separate assessment.

Can someone with Hartnup disorder eat a normal diet?

Many people can. A balanced diet that provides adequate protein is usually the main nutritional approach. A dietitian can help when there are allergies, restricted eating, poor growth, or other medical needs.

Can I pass Hartnup disorder to my children?

A person with Hartnup disorder passes one altered SLC6A19 gene copy to each child. Whether a child develops the condition also depends on whether the other biological parent carries an altered copy. Genetic counselling and carrier testing can clarify the chance for a particular family.

Sources

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