CONDITION

Hereditary fructose intolerance

Hereditary fructose intolerance is an inherited condition that prevents the body from safely breaking down fructose.

WHAT IT IS
Inherited inability to safely break down fructose

BODY SYSTEM
Digestive system, liver, kidneys, and blood sugar control

MAIN SYMPTOMS
Vomiting, abdominal pain, low blood sugar, poor growth

WHAT CAUSES IT
Inherited ALDOB gene changes causing aldolase B deficiency

HOW IT IS DIAGNOSED
ALDOB genetic testing, rarely liver enzyme testing

MAIN TREATMENT
Lifelong avoidance of fructose, sucrose, and sorbitol

CAN IT BE CURED?
No - inherited enzyme deficiency remains lifelong

IS IT AN EMERGENCY?
Sometimes - severe low blood sugar or collapse

LONG-TERM OUTLOOK
Usually healthy with early diagnosis and careful dietary management

Overview

Hereditary fructose intolerance, or HFI, is a rare inherited condition that affects how the body processes fructose. Fructose is a natural sugar found in fruit, honey, some vegetables, and many sweetened foods.

People with HFI do not have enough working aldolase B, an enzyme needed to break down fructose. An enzyme is a protein that helps a chemical process happen in the body. Without enough aldolase B, a substance called fructose-1-phosphate builds up, mainly in the liver and kidneys.

This buildup interferes with the body's ability to release glucose into the blood. Glucose is the main sugar used for energy. Fructose exposure can therefore cause low blood sugar and damage the liver or kidneys.

Symptoms usually begin when a baby first receives foods, drinks, formulas, or medicines containing fructose. Breast milk itself does not contain fructose. Timing differs because the age of first exposure differs.

HFI is not the same as fructose malabsorption. Fructose malabsorption is a digestive problem that can cause bloating, gas, or diarrhoea. HFI is a genetic metabolic condition that can affect blood sugar and internal organs.

Symptoms

Symptoms usually appear after eating or drinking fructose, sucrose, or sorbitol. Sucrose is ordinary table sugar and is made from glucose and fructose. Sorbitol is a sweetener that the body can convert into fructose.

Symptoms following exposure may include:

Babies may develop symptoms when fruit, juice, sweetened foods, or sucrose-containing formula is introduced. Repeated exposure may lead to an enlarged liver, jaundice, or problems with blood clotting. Jaundice causes yellow skin or yellowing in the whites of the eyes.

Some children develop a strong dislike of sweet foods. This learned avoidance can reduce exposure and may make the condition less obvious. An older child or adult may therefore appear well while naturally following a low-fructose diet.

Symptoms are not enough to confirm HFI. Several other conditions can cause vomiting, low blood sugar, liver problems, or poor growth.

- Nausea, vomiting, or abdominal pain
- Sweating, shaking, paleness, or unusual sleepiness
- Irritability, weakness, or confusion from low blood sugar
- Refusal of sweet foods or drinks
- Poor weight gain or slowed growth

Causes and risk factors

HFI is caused by harmful changes in the ALDOB gene. This gene provides instructions for making aldolase B.

The condition follows an autosomal recessive inheritance pattern. This means a person usually has a harmful gene change in both copies of the ALDOB gene, one inherited from each

biological parent.

Parents who carry one altered copy generally do not have HFI. If both parents are carriers, each pregnancy has:

These chances apply separately to each pregnancy. They do not change based on the outcomes of earlier pregnancies.

The main risk factor is having biological parents who both carry an ALDOB gene change. A known family history increases the chance, but HFI may be diagnosed in a family with no previous known cases.

- A 1 in 4 chance of a child having HFI
- A 1 in 2 chance of a child being an unaffected carrier
- A 1 in 4 chance of a child inheriting neither altered copy

Diagnosis

Diagnosis begins with the pattern of symptoms and their relationship to fructose exposure. A clinician may ask about reactions to fruit, juice, sweets, table sugar, or certain medicines. A strong dislike of sweet foods can also be relevant.

Blood and urine tests may show the effects of an episode, such as low blood sugar, liver inflammation, abnormal clotting, or kidney dysfunction. These findings can support concern about HFI, but they do not confirm it.

The usual confirmatory test is genetic testing for harmful changes in both copies of the ALDOB gene. Testing may begin with known family variants when a relative has already been diagnosed.

Testing aldolase B activity in a liver sample can also confirm the condition. However, this requires a liver biopsy, in which a small piece of liver tissue is removed. It is now rarely needed when genetic testing provides a clear answer.

A supervised fructose challenge was used historically. It involves giving fructose and watching for a reaction. Because it can cause severe low blood sugar and liver injury, it should not be used when HFI is suspected.

After confirmation, genetic counselling can help family members understand inheritance and whether carrier or diagnostic testing may be appropriate.

Treatment and management

The main treatment is lifelong avoidance of fructose, sucrose, and sorbitol. Removing these sugars usually prevents acute reactions and allows liver, kidney, growth, and blood sugar problems to improve.

This diet is more restrictive than simply avoiding fruit. Fructose-related sugars may be found in sweetened drinks, desserts, sauces, processed foods, nutritional products, and some medicines. Ingredient names and food formulations can change, so labels need regular review.

A metabolic specialist and a dietitian familiar with inherited metabolic conditions can help identify safe foods and maintain balanced nutrition. Advice should account for age, growth, culture, food access, and other medical needs.

Management may include:

Some liquid medicines, chewable tablets, syrups, and intravenous products may contain fructose, sucrose, or sorbitol. A pharmacist can check inactive ingredients and help identify suitable alternatives. Prescribed medicine should not be stopped without first arranging an appropriate replacement.

An acute reaction may require hospital care. Treatment focuses on correcting low blood sugar, replacing fluids, and monitoring the liver, kidneys, salts, and acid balance. The exact treatment is based on the person's symptoms and test results.

- Reviewing foods, drinks, supplements, and medicine ingredients
- Planning safe meals for childcare, school, travel, and hospital visits
- Monitoring growth, nutrition, liver health, and kidney health
- Considering supplements when testing or diet review identifies a need
- Providing an emergency plan for accidental exposure or illness

Complications

Repeated or substantial fructose exposure can cause severe low blood sugar. The brain relies heavily on glucose, so very low levels may lead to confusion, seizures, loss of consciousness, or coma.

Fructose-1-phosphate buildup can also injure the liver. Possible complications include liver enlargement, jaundice, inflammation, impaired clotting, and, with ongoing exposure, liver failure.

Kidney injury may affect the tubules, the small structures that control which substances are

kept or removed in urine. This can disturb body salts and acid balance. Dehydration may make these problems worse.

Babies and children with unrecognised HFI may have feeding difficulties, poor weight gain, and delayed growth. Most metabolic disturbances improve after the harmful sugars are removed, especially when the condition is recognised early.

A restricted diet can create nutritional gaps if it is not planned carefully. Regular diet review helps ensure adequate energy, vitamins, minerals, and fibre without using unsafe ingredients.

Prevention

HFI itself cannot be prevented because it is inherited. Nothing a parent did during pregnancy or infancy causes the gene changes.

Symptoms and organ injury can usually be prevented by avoiding fructose, sucrose, and sorbitol after diagnosis. Clear food plans, medicine checks, and communication with schools and healthcare services reduce the chance of accidental exposure.

People with HFI or a family history of the condition may choose genetic counselling before or during pregnancy. Counselling explains carrier testing and reproductive testing options without directing personal decisions.

When HFI is known in a family, testing at-risk siblings early can allow dietary protection before fructose is introduced.

Living with it and what to expect

HFI remains lifelong because the underlying gene changes do not go away. The amount of fructose that causes a reaction is not a safe basis for experimenting with the diet. Ongoing avoidance is needed even when a person feels well.

After diagnosis and dietary treatment, low blood sugar episodes and digestive symptoms usually stop. Liver and kidney abnormalities often improve. Growth may recover, although the amount and timing of improvement depend on age at diagnosis and any previous organ injury.

Children can usually attend school, exercise, travel, and take part in ordinary activities. Planning is important around shared food, celebrations, school meals, camps, and medicines. Written instructions can help other adults understand that this is not simply a food preference.

Adults need to continue checking labels and telling healthcare professionals about HFI. Hospital admissions, procedures, pregnancy, and illnesses may involve medicines or nutrition

products with unsafe sugars.

Routine follow-up may include review of growth or weight, dietary intake, liver tests, kidney tests, and any symptoms after possible exposure. Follow-up needs may change over time. Most people who are diagnosed and maintain the diet can remain well, but long-term monitoring is still useful.

When to get medical help

Arrange routine medical review if you or your child has repeated vomiting, sweating, weakness, or unusual sleepiness after sweet foods. Poor growth, persistent food refusal, or a strong pattern of avoiding sweets also deserves assessment, especially when HFI is present in the family.

If HFI has been diagnosed, seek prompt advice after a meaningful accidental exposure, particularly if vomiting, abdominal pain, weakness, or unusual behaviour develops. Follow the person's established emergency plan if one is available.

Get urgent or emergency help for:

Do not deliberately give fructose to test whether symptoms occur. A suspected exposure should be managed based on symptoms and the person's medical plan.

- Severe sleepiness, confusion, or difficulty waking
- A seizure, collapse, or loss of consciousness
- Symptoms of severe low blood sugar
- Repeated vomiting with inability to keep fluids down
- Yellow skin or eyes, unusual bleeding, or a swollen abdomen
- Signs of serious dehydration, such as very little urine

Common questions

Can I eat fruit if I have hereditary fructose intolerance?

Fruit contains fructose and is generally excluded from an HFI diet. The exact food plan should be developed with a metabolic dietitian because fructose, sucrose, and sorbitol occur in more foods than fruit alone. Avoid testing personal tolerance by eating small amounts.

Is hereditary fructose intolerance the same as fructose malabsorption?

No. Fructose malabsorption happens when the intestine does not absorb fructose well. It

mainly causes gas, bloating, pain, or diarrhoea.

HFI is caused by aldolase B deficiency. It can cause low blood sugar and liver or kidney injury, so it requires a stricter lifelong diet.

Do children grow out of hereditary fructose intolerance?

No. HFI is caused by inherited gene changes and remains throughout life. Symptoms can disappear when unsafe sugars are avoided, but this does not mean the condition has gone away.

What happens if I accidentally eat fructose?

The response may be influenced by the amount eaten and how quickly it is recognised. Possible symptoms include nausea, vomiting, abdominal pain, sweating, weakness, or sleepiness.

Follow the established emergency plan and seek prompt medical advice if symptoms develop. Severe confusion, seizures, collapse, or difficulty waking require emergency help.

Can hereditary fructose intolerance be cured?

There is currently no cure that restores aldolase B function. Lifelong dietary management is effective at preventing most symptoms and complications. Research into inherited metabolic conditions continues, but avoidance remains the standard treatment.

Will my children inherit hereditary fructose intolerance?

A person with HFI passes an altered ALDOB gene copy to each biological child. Whether a child develops HFI also depends on the other biological parent's ALDOB genes. Genetic counselling and carrier testing can clarify the possibilities for a particular family.

Sources

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