CONDITION

Galactosemia

Galactosemia is an inherited condition that prevents the body from properly processing the sugar galactose.

WHAT IT IS
Inherited inability to process the sugar galactose

BODY SYSTEM
Liver, brain, eyes, ovaries, and metabolism

MAIN SYMPTOMS
Poor feeding, vomiting, jaundice, poor weight gain

WHAT CAUSES IT
Inherited changes affecting galactose-processing enzymes

HOW IT IS DIAGNOSED
Newborn screening, enzyme testing, and genetic testing

MAIN TREATMENT
Dietary galactose restriction and specialist monitoring

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

IS IT AN EMERGENCY?
Sometimes - severe illness or jaundice in a newborn

LONG-TERM OUTLOOK
Often good survival, but lifelong complications may occur

Overview

Galactosemia is a rare inherited condition affecting how the body processes galactose. Galactose is a simple sugar and part of lactose, the natural sugar in breast milk, standard infant formula, and dairy products.

Normally, enzymes change galactose into substances the body can use. In galactosemia, a required enzyme is missing or does not work properly.

Several related conditions are called galactosemia. Classic galactosemia, caused by low activity of the GALT enzyme, is usually the most serious. Other forms involve the GALK1 or GALE enzymes. Milder GALT variants, such as Duarte galactosemia, are managed differently from classic galactosemia.

Without early treatment, classic galactosemia can make a newborn seriously ill after milk feeding begins. It can damage the liver, brain, kidneys, and eyes and increase the risk of bloodstream infection. Newborn screening can identify many babies before major problems develop.

Removing major dietary sources of galactose prevents many immediate complications. However, the body also produces galactose, so learning, speech, movement, bone, or reproductive problems may occur despite careful dietary management.

Symptoms

A baby with classic galactosemia often appears healthy at birth. Symptoms may begin within the first days of breast milk or standard formula feeding.

Early symptoms include:

Untreated babies can develop liver failure or sepsis, a dangerous whole-body reaction to infection. Infection with Escherichia coli (E. coli) is a particular concern.

Other forms may be less severe. Galactokinase deficiency mainly causes cataracts. GALE deficiency ranges from a form affecting only red blood cells to a severe condition involving several organs.

After early treatment, liver and feeding problems often improve. Some people with classic galactosemia later develop:

Not everyone develops these concerns, and diet alone does not predict their severity.

- Poor feeding or refusing feeds
- Vomiting
- Sleepiness or reduced alertness
- Poor weight gain
- Yellow skin or eyes, called jaundice
- A swollen abdomen or enlarged liver
- Easy bleeding or bruising
- Clouding of the eye lens, called a cataract
- Delayed or unclear speech
- Learning or information-processing difficulties

- Problems with balance, coordination, or tremor
- Anxiety, low mood, or social difficulties
- Reduced bone strength
- Delayed puberty or reduced ovarian function

Causes and risk factors

Galactosemia results from a gene change affecting galactose breakdown. Classic galactosemia is caused by changes in the GALT gene. Other types involve GALK1 or GALE .

These conditions are usually inherited in an autosomal recessive pattern. A child inherits one altered gene copy from each biological parent. The parents are usually healthy carriers.

When both parents carry an altered copy of the same gene, each pregnancy has:

These chances are the same for every pregnancy. Having an affected biological sibling or known carrier parents increases the likelihood of galactosemia. The condition is not caused by anything done during pregnancy and is not contagious.

- A 1 in 4 chance that the child will have the condition
- A 1 in 2 chance that the child will be a healthy carrier
- A 1 in 4 chance that the child will inherit neither altered copy

Diagnosis

Where galactosemia is included in newborn screening, a heel-prick blood sample may identify a baby at risk. Screening may measure GALT enzyme activity, galactose-related substances, or both. Practices vary by location.

Screening does not confirm the condition. Prompt follow-up usually includes measuring GALT enzyme activity in red blood cells and testing the GALT gene. Other biochemical or genetic tests may be used if another type is suspected.

A recent blood transfusion can interfere with red blood cell enzyme testing because the sample may contain donor cells. Tell the laboratory and metabolic team about any transfusions.

If symptoms suggest classic galactosemia, dietary treatment may begin before results are confirmed to reduce immediate risk.

After diagnosis, tests may assess liver and kidney function, blood clotting, infection, and eye

health. Ongoing reviews monitor growth, nutrition, speech, development, movement, bones, and reproductive development.

Treatment and management

Treatment depends on the affected enzyme and its remaining activity. Classic galactosemia requires care from a metabolic team and dietitian.

Early treatment removes breast milk, standard milk-based formula, and other major sources of lactose and galactose. Babies receive an appropriate alternative formula chosen with their healthcare team. Do not change feeds after a positive newborn screen without prompt medical guidance, because confirmatory testing and clinical assessment are essential.

As the child grows, dietary management avoids milk and dairy ingredients containing substantial lactose or galactose. Many non-dairy foods contain only small amounts and may be allowed. Modern care generally does not remove every trace from fruit, vegetables, and legumes, but the plan should come from a metabolic dietitian.

Dairy restriction can reduce calcium and vitamin D intake. Nutrition reviews can identify whether fortified foods or supplements are needed. Check supplements, medicines, and nutritional products with the metabolic team because some contain lactose.

Ongoing care may include:

Severe infection, liver failure, or other acute complications require hospital treatment. Dietary restriction limits harmful substances but does not correct the altered gene.

Duarte galactosemia is not managed in the same way as classic galactosemia. Any restriction should reflect the confirmed diagnosis and specialist guidance.

- Growth and nutrition reviews
- Developmental, learning, and speech assessments
- Speech and language therapy
- Physical or occupational therapy
- Eye examinations
- Bone health monitoring
- Assessment of puberty, menstrual health, and ovarian function
- Psychological or educational support

Complications

Early treatment greatly reduces the risk of liver failure, cataracts, and severe infection during infancy. Delayed treatment can be life-threatening.

Long-term complications may occur despite consistent dietary care, including speech disorders, learning difficulties, tremor, coordination problems, reduced bone density, and emotional or social difficulties.

Many girls and women with classic galactosemia develop primary ovarian insufficiency, meaning the ovaries stop working normally earlier than expected. This can affect puberty, periods, hormone levels, and fertility. Pregnancy is still possible for some people.

Boys and men do not usually have the same degree of reproductive impairment, although individual assessment may be appropriate.

Dairy restriction may cause low calcium or vitamin D intake unless suitable alternatives are included, so regular nutrition and bone reviews are important.

Prevention

Galactosemia cannot be prevented because it is inherited. Nothing a parent eats, takes, or does during pregnancy causes it.

Newborn screening cannot prevent the condition, but early identification can prevent or limit many acute complications.

People with a family history can consider genetic counselling and carrier testing. When the family's gene changes are known, prenatal testing or testing embryos created through in vitro fertilisation may be available. These are personal choices, not required steps.

Living with it and what to expect

During infancy, care focuses on safe feeding, growth, and complications. After the correct feeding plan begins, jaundice, liver problems, and cataracts caused by recent exposure may improve. Recovery may take time if the baby was seriously ill before diagnosis.

During childhood, follow-up focuses on development, speech, learning, movement, and balanced nutrition. Some children need school support or speech and language therapy, while others reach milestones without major help.

Teenagers can gradually learn to read food labels, explain the condition, and manage meals away from home. Planning for school, travel, social events, and emergencies can help. A

written care plan allows other adults to understand dietary and medical needs.

Adults still need metabolic follow-up covering nutrition, bones, movement symptoms, emotional health, and reproductive concerns. Dietary treatment remains important, but stricter restriction does not necessarily prevent long-term complications because the body produces galactose.

Many people with galactosemia attend school, work, form relationships, travel, and live independently. Support needs vary. Regular follow-up helps identify treatable concerns early.

When to get medical help

Seek emergency medical care for a newborn who is difficult to wake, has trouble breathing, appears pale or mottled, has a seizure, or is not responding normally.

A newborn needs urgent same-day assessment for jaundice, repeated vomiting, poor feeding, unusual sleepiness, fever, or signs of dehydration. This is especially important after a positive newborn screening result or when galactosemia runs in the family.

If a baby with suspected or confirmed galactosemia becomes unwell, contact the metabolic team or emergency service using the baby's emergency care plan. Do not wait for a routine appointment.

Contact the metabolic team promptly after a significant feeding or dietary error, especially in an infant, or if prescribed formula is unavailable.

Arrange routine assessment for slow growth, speech or school difficulties, tremor, balance problems, bone pain, delayed puberty, irregular or absent periods, or difficulty following the dietary plan.

Common questions

Can a baby with galactosemia have breast milk?

Breast milk contains lactose and is not suitable for a baby being treated for classic galactosemia. The metabolic team will recommend an alternative formula. Advice may differ for milder variants.

Is galactosemia the same as lactose intolerance?

No. Lactose intolerance affects digestion in the intestine and usually causes bloating, gas, or diarrhoea. Galactosemia affects galactose processing after absorption. Classic galactosemia can cause serious liver, brain, eye, and infection-related complications.

Can galactosemia go away with age?

No. The inherited enzyme problem is lifelong, although dietary and monitoring needs may change.

Can someone with galactosemia eat fruit and vegetables?

Many fruits, vegetables, and legumes contain small amounts of galactose but are often allowed in modern plans. Avoiding every trace is generally neither practical nor nutritionally helpful. Follow advice from a metabolic dietitian.

Does following the diet prevent every complication?

No. Early treatment prevents many serious newborn complications, but speech, learning, movement, bone, or ovarian problems may still occur, partly because the body produces galactose.

Can people with galactosemia have children?

Yes, some can. Reduced ovarian function may make pregnancy more difficult, but spontaneous pregnancy can occur. Fertility in men is usually less affected. Reproductive assessment can explain individual options.

Sources

- Berry GT. "Classic Galactosemia and Clinical Variant Galactosemia." GeneReviews , University of Washington, Seattle; updated March 11, 2021.
- Welling L, Bernstein LE, Berry GT, et al. "International clinical guideline for the management of classical galactosemia: diagnosis, treatment, and follow-up." Journal of Inherited Metabolic Disease . 2017;40(2):171-176.
- American College of Medical Genetics and Genomics. "Classic Galactosemia: Newborn Screening ACT Sheet." ACMG ACT Sheets and Algorithms.
- MedlinePlus Genetics. "Galactosemia." U.S. National Library of Medicine.
- National Organization for Rare Disorders. "Galactosemia." Rare Disease Database.