CONDITION

Galactose-1-phosphate uridyl transferase deficiency

An inherited condition in which the body cannot properly process the milk sugar galactose.

WHAT IT IS
Inherited difficulty breaking down galactose

BODY SYSTEM
Metabolism, liver, brain, eyes, and ovaries

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

WHAT CAUSES IT
Inherited GALT gene changes

HOW IT IS DIAGNOSED
Blood enzyme, galactose-1-phosphate, and genetic tests

MAIN TREATMENT
Lifelong dietary galactose restriction and specialist monitoring

CAN IT BE CURED?
No - the enzyme deficiency is lifelong

IS IT AN EMERGENCY?
Sometimes - untreated illness in a newborn

LONG-TERM OUTLOOK
Survival is good, but lasting complications remain possible

Overview

Galactose-1-phosphate uridyl transferase deficiency, usually called GALT deficiency , is an inherited condition affecting how the body processes galactose. Galactose is a simple sugar found mainly as part of lactose, the natural sugar in breast milk, cow's milk, and many dairy products.

GALT is an enzyme that helps process galactose. When its activity is very low or absent, galactose-1-phosphate and related substances build up and can damage the liver, brain, kidneys, and eyes.

Severe GALT deficiency is commonly known as classic galactosemia . Symptoms usually begin in the first days of life after a baby starts breast milk or standard milk-based formula. Without

prompt treatment, a newborn can become seriously ill.

Some people have partial enzyme activity, sometimes called clinical variant galactosemia. The Duarte variant is a common partial-deficiency form that usually does not cause the severe newborn illness associated with classic galactosemia. Its management may differ.

Removing major galactose sources usually reverses immediate feeding, liver, and infection-related problems. However, long-term speech, learning, movement, and reproductive complications may occur despite early treatment.

Symptoms

A baby with classic GALT deficiency often appears healthy at birth. Symptoms develop after milk feeding begins and may include:

The liver may stop working properly, causing low blood sugar and clotting problems. Some babies develop a serious bloodstream infection, particularly from Escherichia coli . This is known as sepsis.

People with partial GALT activity may have mild or no symptoms and may be identified only through newborn screening or family testing.

Possible long-term signs include unclear speech, tremor, poor coordination, learning difficulties, and problems with attention or planning. Girls and women may have delayed puberty, irregular or absent periods, or reduced fertility because of impaired ovarian function.

Not everyone develops these complications, 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
- An enlarged liver
- Easy bruising or bleeding
- Clouding of the eye's lens, called a cataract

Causes and risk factors

GALT deficiency is caused by harmful changes in the GALT gene , which provides instructions for making the GALT enzyme.

The condition has an autosomal recessive inheritance pattern. A child must inherit one altered gene copy from each biological parent. Carriers with one altered copy usually have no symptoms.

When both biological parents are carriers, each pregnancy has:

These chances apply independently to every pregnancy. Having an affected close relative increases the likelihood of carrying a GALT gene change.

The condition is present from conception. It is not caused by anything a parent ate, did, or encountered during pregnancy.

- A 1 in 4 chance of a child having GALT deficiency
- 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

In many countries, classic galactosemia is included in newborn blood spot screening. Blood collected from the baby's heel may be tested for GALT enzyme activity, galactose-related substances, or both.

A positive screening result is not a final diagnosis. False-positive results and partial forms can occur. Follow-up testing should happen promptly, with feeding advice from the newborn or metabolic team.

Confirmatory tests include:

A recent blood transfusion can affect enzyme testing because the sample may contain donor red blood cells. Genetic testing is particularly helpful in this situation.

Other tests may assess liver function, blood clotting, blood sugar, kidney function, or infection. An eye examination may check for cataracts.

If a relative has confirmed GALT deficiency, carrier testing and testing during a future pregnancy may be possible once the family's gene changes are known. A genetics professional can explain the options and limitations.

- GALT enzyme testing , usually in red blood cells
- Galactose-1-phosphate measurement in red blood cells

- Genetic testing for harmful changes in both GALT gene copies

Treatment and management

A newborn with suspected classic galactosemia is usually taken off breast milk and standard lactose-containing formula immediately while confirmatory tests are arranged. The metabolic and paediatric team will select a suitable low-galactose or galactose-free formula. Treatment should not be delayed when screening results and symptoms strongly suggest the condition.

Confirmed classic GALT deficiency requires lifelong avoidance of major dietary galactose sources, generally including milk and most ordinary dairy products. Food labels should be checked for milk and lactose ingredients.

The diet does not usually require avoiding every trace of galactose. Fruits, vegetables, legumes, and other non-milk foods containing small amounts are generally allowed. Some mature cheeses or specific low-galactose products may be acceptable under local guidance. An inherited metabolic dietitian should define a suitable diet because recommendations can change.

Calcium and vitamin D intake require attention because dairy products are major sources. Supplements may be recommended after reviewing diet and blood results, especially to support bone health during childhood, puberty, and adulthood.

Routine follow-up may include:

Speech therapy, educational support, occupational therapy, or treatment for movement symptoms may help. Some girls and women with reduced ovarian function may be offered hormone or fertility care.

The body naturally produces some galactose, which may help explain why long-term complications can occur despite careful dietary management. These complications do not necessarily mean the diet was managed incorrectly.

- Growth, nutrition, calcium, and vitamin D assessment
- Developmental, learning, and school-progress reviews
- Speech and language assessment
- Checks for tremor, coordination, or movement problems
- Bone health monitoring
- Puberty, menstrual, ovarian, and fertility assessment when relevant
- Emotional and mental health support

Complications

Before diagnosis, classic GALT deficiency can cause liver failure, abnormal bleeding, low blood sugar, cataracts, kidney problems, and sepsis. These problems often improve after dietary galactose is removed and infection or organ dysfunction is treated.

Possible long-term complications include:

Reduced ovarian function may cause delayed puberty, absent periods, or difficulty becoming pregnant. Spontaneous pregnancy remains possible, so reduced fertility does not mean complete infertility.

Boys and men generally do not have the same degree of reproductive impairment. Gene changes and remaining enzyme activity may influence the condition but cannot predict every outcome. Even affected siblings may have different experiences.

- Speech and language difficulties
- Learning or intellectual difficulties
- Problems with planning, attention, or processing speed
- Poor coordination, tremor, or other movement disorders
- Low bone density
- Anxiety, low mood, or social difficulties
- Reduced ovarian function in many affected girls and women

Prevention

GALT deficiency cannot be prevented because it is inherited.

Newborn screening can identify many affected babies before severe symptoms develop, allowing treatment to begin quickly.

If the condition runs in a family, genetic counselling can explain carrier testing and reproductive options. These may include testing during pregnancy or testing embryos created through in vitro fertilisation. Whether to use these options is a personal decision.

Diet during pregnancy does not cause or prevent GALT deficiency in the baby.

Living with it and what to expect

After newborn illness is treated, most children with classic GALT deficiency grow up at home, attend school, and participate in everyday activities. Families may need to plan for childcare, school meals, travel, restaurants, and social events.

Sensitivity to major galactose sources is lifelong, so dietary limits usually continue through adulthood. The diet should be reviewed periodically as evidence and specialist recommendations change.

Long-term progress varies. Some children develop typically, while others need help with speech, coordination, learning, or emotional wellbeing. Difficulties may become clearer over months or years.

Regular developmental and school reviews can identify support needs. Adults may continue to need metabolic follow-up, nutritional and bone health checks, and reproductive care. Transition from children's to adult services should include a clear written plan.

A medical identification card or emergency information letter can explain the diagnosis. Schools and other caregivers should also receive written dietary guidance.

When to get medical help

A newborn needs urgent same-day medical assessment if newborn screening suggests galactosemia. Follow the screening team's feeding instructions while confirmatory tests are arranged.

Seek emergency care for a baby with possible or confirmed GALT deficiency who has:

These signs may indicate sepsis, liver dysfunction, low blood sugar, or another serious newborn illness.

For someone already diagnosed, contact the metabolic team promptly after a major accidental milk exposure, especially if vomiting or illness develops. A small accidental exposure is not always an emergency, but the established care plan should be followed.

Discuss concerns about growth, diet, speech, school progress, coordination, tremor, puberty, periods, bone health, fertility, or emotional wellbeing at routine appointments.

- Difficulty waking, unusual limpness, or reduced responsiveness
- Poor feeding with repeated vomiting
- Yellow skin or eyes with worsening illness
- Fever, abnormal temperature, or concern for infection
- Breathing difficulty

- Seizures
- Unusual bruising, bleeding, or a swollen abdomen

Common questions

Is GALT deficiency the same as galactosemia?

GALT deficiency is the most common cause of classic galactosemia. Galactosemia can also refer to other rare enzyme deficiencies affecting galactose processing. Exact test results matter because treatment and expected outcomes differ.

Can a baby with classic GALT deficiency be breastfed?

Breast milk contains lactose and is a major galactose source. A baby with suspected or confirmed classic GALT deficiency is usually changed promptly to a suitable alternative formula under specialist guidance. Partial forms, including the Duarte variant, may be managed differently.

Will avoiding dairy prevent every complication?

No. Restricting major galactose sources is essential for preventing or reversing dangerous newborn effects. However, speech, movement, learning, bone, or ovarian complications may still occur, partly because the body produces galactose internally.

Is the Duarte variant dangerous?

The Duarte variant causes partial rather than severe GALT deficiency. Most affected infants do not develop the acute illness or long-term pattern associated with classic galactosemia. Dietary recommendations vary, so management should be based on exact enzyme and genetic results.

Can someone with GALT deficiency have children?

Yes. Men are generally fertile. Many women have reduced ovarian function and may find pregnancy difficult, but spontaneous pregnancy can occur. Hormone, fertility, and pregnancy care can be tailored to the individual.

Can GALT deficiency go away with age?

No. The inherited enzyme deficiency is lifelong. Severe newborn symptoms can improve quickly with treatment, but dietary management and follow-up usually continue into adulthood.

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. "Newborn Screening ACT Sheet: Absent or Reduced GALT - Classic Galactosemia." ACMG ACT Sheets and Algorithms.
- National Library of Medicine. "Galactosemia." MedlinePlus Genetics . Reviewed May 1, 2015.
- Demirbas D, Coelho AI, Rubio-Gozalbo ME, Berry GT. "Hereditary galactosemia." Metabolism . 2019;93:39-51.