



Genetic Fact Sheets for Parents

Other Disorders

Screening, Technology, and Research in Genetics is a multi-state project to improve information about the financial, ethical, legal, and social issues surrounding expanded newborn screening and genetic testing – [http:// www.newbornscreening.info](http://www.newbornscreening.info)

Disorder name: Galactosemia
Acronym: GALT deficiency

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This fact sheet has general information about galactosemia. Every child is different and some of these facts may not apply to your child specifically. Certain treatments may be advised for some children but not others. All children with galactosemia should be followed by a metabolic doctor in addition to their primary care provider.

If your newborn has had a 'presumptive positive screen for galactosemia' as a result of newborn screening, it does not yet mean that he or she has galactosemia. There are other tests that need to be done to confirm whether your baby actually has galactosemia. Some babies are found not to have galactosemia and do not need treatment. Others are found to have a milder condition called "Duarte galactosemia." Many children with Duarte galactosemia do not need treatment. Blood and urine tests can be done to determine whether your child has classic or Duarte galactosemia.

This fact sheet contains information about classic galactosemia only. Therefore, the details below are relevant only to babies who have been confirmed to have classic galactosemia. It does not contain information on Duarte galactosemia or other variants.

What is galactosemia?

Galactosemia, which means “galactose in the blood,” is a rare inherited condition. People with galactosemia have problems digesting a type of sugar called galactose from the food they eat. Because they cannot break galactose down properly, it builds up in their blood. Galactose is found in milk and all foods that contain milk.

What causes galactosemia?

In order for the body to use different types of carbohydrates and sugars from the foods we eat, special enzymes break them down into smaller sugar molecules called glucose, which the body uses for fuel.

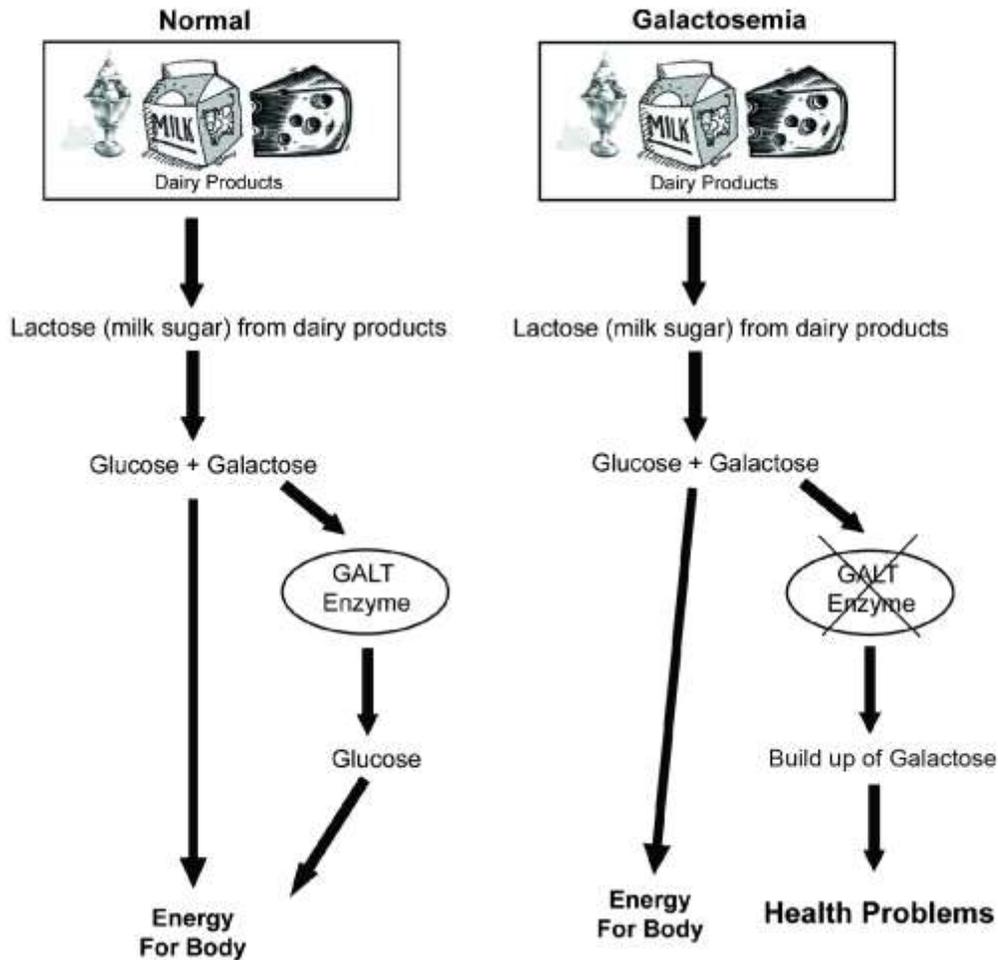
Lactose, also called ‘milk sugar,’ is the main type of sugar found in milk and milk products. It is made of one molecule of galactose and one molecule of glucose. Thus, all lactose, and all milk and milk products, contain galactose. During digestion, lactose is broken down to galactose and glucose. Then galactose is further changed by the body into glucose so it can be used as energy.

Galactosemia is caused by problems with the enzyme ‘galactose-1-phosphate uridyl transferase’ (GALT). In people with galactosemia, the GALT enzyme is either missing or not working properly. This enzyme’s job is to change galactose into glucose. When the GALT enzyme is missing or not working properly, galactose cannot be changed to glucose so it builds up in the blood in large amounts. Unless treated, the excess galactose will affect many parts of the body and, over time, may be life-threatening.

Galactosemia

Galactosemia occurs when babies do not have enough of the GALT enzyme. Babies start showing health effects within days of feeding on breast milk or milk-containing formulas. Virtually all cases of classic galactosemia can be detected by newborn screening.

GALACTOSEMIA



If galactosemia is not treated, what problems occur?

Excess galactose in the blood affects many parts of the body. Some of the organs that may be affected include the brain, eyes, liver, and kidneys.

Infants with galactosemia usually have diarrhea and vomiting within a few days of drinking milk or formula containing lactose.

Some of the other early effects of untreated galactosemia include:

- Failure to gain weight or grow in length
- Poor feeding and poor suck
- Lethargy
- Irritability

If treatment is not started, other symptoms are likely to follow:

- Low blood sugar, called hypoglycemia
- Seizures
- Enlarged liver that does not work properly
- Jaundice (yellow color to the skin or whites of the eyes)
- Bleeding
- Serious blood infections that could lead to shock and death
- Early cataracts, which occur in about 10% of children

Some untreated babies have high levels of ammonia, a toxic substance, in their blood. High ammonia levels and hypoglycemia can both lead to coma and, if not treated, can cause death.

Most untreated children eventually die of liver failure. Surviving babies who remain untreated may have intellectual disabilities and other damage to the brain and nervous system.

Even with adequate treatment, individuals with galactosemia may develop one or more of the following:

- Early cataracts
- Mild intellectual disabilities or learning delays
- Ataxia (unsteady gait)
- Delays in growth
- Speech problems and delays

Most girls with galactosemia will have delayed periods or do not get their periods at all. Some women with galactosemia start menopause early or have 'premature ovarian failure' in which the ovaries stop releasing eggs earlier than normal menopause.

What is the treatment for galactosemia?

Your baby's primary doctor will work with a metabolic doctor and a dietitian familiar with galactosemia to care for your child.

Prompt treatment is needed to prevent serious health problems and intellectual disabilities. Babies with galactosemia who do not start treatment shortly after birth may have permanent effects.

Infants and children who are either missing the GALT enzyme completely or who have less than 10% of the normal amount of enzyme must follow a special food plan. They need to avoid all foods with lactose and galactose. All milk and milk products must be replaced with formula that contains no lactose.

The following are treatments often advised for children with classic galactosemia:

1. Lactose and galactose-free diet

People with classic galactosemia are encouraged to follow a lactose and galactose-free food plan throughout life. Lactose or galactose are found in the following foods, all of which must be avoided:

- Milk and all dairy products
- Processed and pre-packaged foods often contain lactose
- Tomato sauces
- Some candies
- Certain medications – tablets, capsules, sweetened liquid drops that contain lactose as a filler
- Some fruits and vegetables also contain galactose
- Any foods or drugs which contain the ingredients lactulose, casein, caseinate, lactalbumin, curds, whey, or whey solids

Your dietitian will help you develop a food plan that allows your child to avoid lactose and galactose while still eating the right amount of protein, nutrients, and energy to keep him or her healthy.

Your child's food plan will depend on many things such as his or her age, weight, general health, and blood test results. Your dietician will fine-tune your child's diet over time. The special food plan should be continued throughout life.

2. Special lactose-free formula

Newborns with galactosemia are given a special formula free of lactose. The most common formulas used for babies with galactosemia are those made with soy protein isolate. Soy milk itself contains galactose and should not be used.

Your metabolic doctor and dietitian will tell you what type of formula is best and how much to use. Some states offer help with payment for this formula and others require private insurance coverage for the formula and other special medical foods.

3. Calcium supplements

Since children with galactosemia are not eating milk products, calcium intake may be too low. This may cause their bones to be weaker than average (osteopenia). Therefore, children with galactosemia are often advised to take calcium supplements to ensure they receive enough calcium each day.

Some doctors also advise Vitamin D and Vitamin K supplements in addition to calcium.

Your doctor will tell you what supplements to give your child and how much. Do not use any medication or supplement without checking with your doctor.

4. Monitoring health

Babies and young children with galactosemia usually need regular blood and urine tests. These tests are used to detect toxic substances made when galactosemia is not in good control. The test results will help your doctors and dietitian fine-tune the treatment to meet your child's needs.

Your doctor may also suggest a formal evaluation of your child's mental development and his or her speech and language skills. If your child shows delays in certain areas of learning or speech, extra help can be arranged. Early intervention programs are available in most states to provide services before children reach school age.

Monitoring for other health problems associated with galactosemia such as cataracts, bone problems, and premature ovarian failure may also be done.

5. Informing friends, relatives, teachers, and child-care providers

It is important for you to tell everyone who helps care for your child that he or she cannot eat or drink milk-containing foods. A Medic-Alert bracelet that states your child's food restrictions can be helpful. In addition, your doctor may advise you to carry an emergency treatment letter with steps for your child's care.

What happens when galactosemia is treated?

Because the body also makes some galactose, symptoms cannot be completely avoided by removing all lactose and galactose from the diet. Researchers are working on finding a treatment to lower the amount of galactose made by the body, but there is no effective method to do so at this time.

When treatment starts before a baby is 10 days old, there is a much better chance for normal growth, development, and intelligence. Some children who receive early treatment may have delays in growth, but most attain normal adult heights.

Even with careful treatment from an early age, some children with classic galactosemia show delays in learning and development and may need extra help in school. Some children develop speech and language delays. Some have delays in motor skills such as walking and coordination and balance problems.

Even when carefully treated, girls with galactosemia have a higher chance of having delayed periods and having premature ovarian failure.

If treatment is started after 10 days of life, delays or learning problems are more likely. The level of delay varies from child to child. Treatment is still important, even if started late, because it can help prevent further delays and symptoms.

What causes the GALT enzyme to be absent or not working correctly?

Genes tell the body how to make enzymes. The GALT gene instructs the body to make the GALT enzyme. Everyone has two copies of the GALT gene. People with galactosemia have changes, also called variants, in both copies of their GALT genes. Because of the variants in the GALT genes, the GALT enzyme either does not work properly or is not made at all.

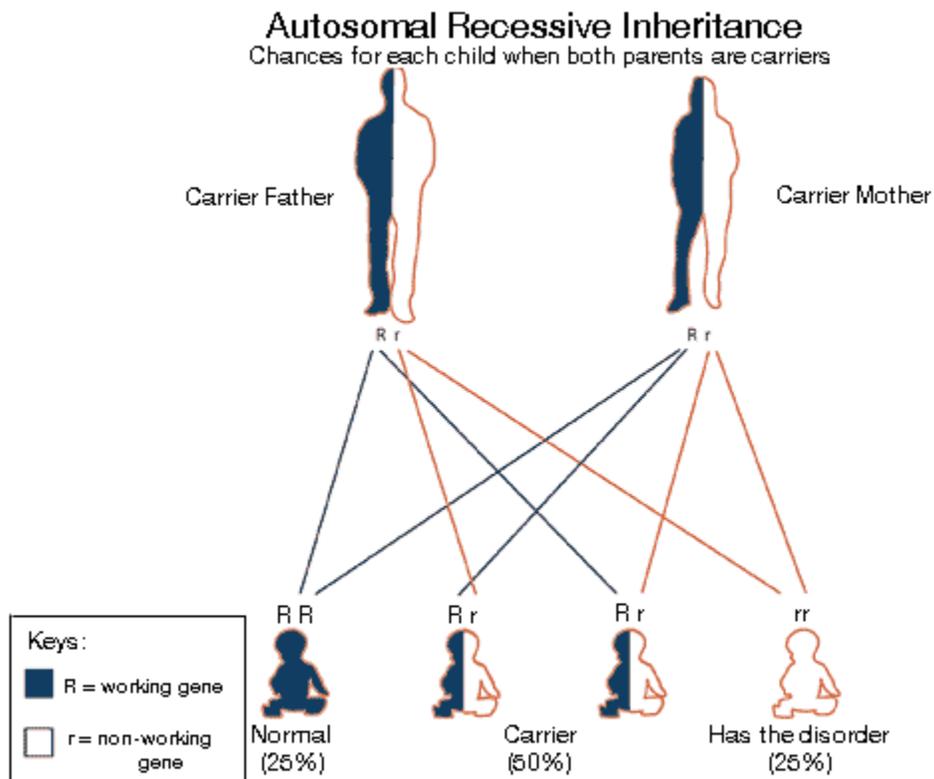
How is galactosemia inherited?

Galactosemia is inherited in an autosomal recessive manner. It affects both boys and girls equally.

Everyone has two copies of the GALT gene that make the GALT enzyme. In children with galactosemia, neither of their GALT genes works correctly. These children inherit one non-working GALT gene from each parent.

Parents of children with galactosemia rarely have the condition themselves. Instead, each parent has a single non-working GALT gene for galactosemia. They are called carriers. Carriers do not have galactosemia because the other GALT gene is working correctly.

When both parents are carriers, there is a 25% chance in each pregnancy for the child to have galactosemia. There is a 50% chance for the child to be a carrier, just like the parents. And, there is a 25% chance for the child to have two working genes.



Genetic counseling is available to families who have children with galactosemia. Genetic counselors can answer your questions about how galactosemia is inherited, choices during future pregnancies, and how to test other family members. Ask your doctor about a referral to a genetic counselor.

Is genetic testing available?

Genetic testing for galactosemia can be done on a blood sample. Genetic testing, also called DNA testing, looks for changes in the GALT genes that causes galactosemia. Over 99% of the time, DNA testing can identify the variants in the GALT genes in a child with this condition.

DNA testing is not necessary to diagnose your child. However, it may be helpful in determining what type of galactosemia your child has.

What other testing is available?

If your child has had a positive screen for galactosemia through a newborn screening program, other tests still need to be done in order to confirm the diagnosis. One of these special tests detects the amount of GALT enzyme present in red blood cells and is often used to confirm galactosemia.

Other blood or urine tests may be helpful to determine whether your child needs treatment or whether treatment is working properly. Ask your doctor if you have any questions about testing for galactosemia.

Can you test during a future pregnancy?

If both gene changes (variants) have been found in your child with galactosemia, DNA testing can be done during any future pregnancies. If DNA testing is not helpful, an enzyme test using cells from the fetus can be done during pregnancy. The sample needed for this test is obtained by either CVS or amniocentesis.

Parents may choose to have testing during pregnancy or wait until birth to have the baby tested. Parents may also choose to use assisted reproductive techniques to decrease the chance that their future children would have galactosemia. A genetic counselor can talk to you about your choices and answer questions about prenatal testing or testing your baby after birth.

If you do not have prenatal diagnosis during your next pregnancies, you should feed your newborn with the recommended formulas instead of breast milk or regular formula until the results of newborn screening or additional diagnostic tests have been completed.

Can other members of the family have galactosemia or be carriers?

Having galactosemia

If they are healthy and show typical development, older brothers and sisters of a baby with galactosemia are unlikely to have the condition. Talk to your doctor or genetic counselor if you have questions about your other children.

Galactosemia carriers

Brothers and sisters who do not have galactosemia still have a 2/3rds chance to be carriers like their parents. Except in special cases, carrier testing should only be done in people over 18 years of age.

If you are a parent of a child with galactosemia, your brothers and sisters have a 50% chance to be a carrier. It is important for other family members to be told that they could be carriers. There is a small chance they are also at risk to have children with galactosemia.

All 50 US states offer newborn screening for galactosemia. Newborn screening can detect virtually all babies with classic galactosemia. However, in families in which a child has mild galactosemia, newborn screening results are not sufficient to rule out this condition in future siblings. In newborns who have a sibling with

mild galactosemia, special diagnostic testing is recommended in addition to newborn screening.

Can other family members be tested?

Diagnostic testing

If there is concern about whether they have galactosemia, your other children can be tested. Talk to your doctor or genetic counselor if you have questions about testing for galactosemia.

Carrier testing

If both gene changes (variants) have been found in your child, other adult family members can have DNA testing to see if they are carriers.

If DNA testing is not helpful, other methods of carrier testing may be available. If you have questions about carrier testing, ask your genetic counselor or metabolic doctor.

How many people have galactosemia?

About one in every 30,000 to 60,000 babies in the United States is born with galactosemia.

Does galactosemia happen more often in a certain ethnic group?

Galactosemia occurs in people of all ethnic groups around the world. It is more common in people from Ireland. One in 24,000 Irish babies is born with this condition.

Does galactosemia go by any other names?

Galactosemia is sometimes also called:

- Galactose-1-phosphate uridyl transferase deficiency
- GALT deficiency
- GALT

Variants of classic GALT that are not discussed in this fact sheet include:

- Galactokinase deficiency
- UDP-galactose 4-epimerase deficiency (GALE)

Where can I find more information?

Metabolic Support UK

<https://www.metabolicsupportuk.org/>

Genetic Home Reference

<https://ghr.nlm.nih.gov/condition/galactosemia>

The Galactosemia Foundation

<http://www.galactosemia.org>

Baby's First Test

<http://www.babysfirsttest.org/newborn-screening/conditions/classic-galactosemia>

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