

# Galactosemia

## What is Galactosemia?

Galactosemia is an inherited condition that affects the way a person's body breaks down galactose. When a person eats or drinks a product that contains lactose or milk sugar (e.g., dairy products such as milk, cheese, butter, breast milk, and baby formula), the body breaks the lactose down into galactose and glucose. Glucose is the sugar used by the body for energy. Galactose is also a sugar used by the body for energy, but first enzymes must break it down. In the case of most galactosemia, one enzyme is absent or insufficient. Too much galactose and its metabolites then build up in the blood and can cause brain damage and serious health problems. If galactosemia is left untreated, it can lead to death.

About one baby in 53,000 is born with galactosemia in the United States. The condition can be found in all ethnic groups.

## How Does Galactosemia Affect a Child?

The symptoms of this condition vary depending on the form of galactosemia. The most severe form of this condition is known as classic galactosemia. Infants with untreated classic galactosemia appear normal at birth, but symptoms may start soon after a baby's first lactose based feeding. Generally, the symptoms include vomiting, rapid weight loss, jaundice, and an enlarged liver (hepatomegaly). Without treatment, these babies can develop permanent brain damage, cataracts, an enlarged liver, kidney failure, and death. Treatment of classic galactosemia is continued for life.

A different or variant form of galactosemia is called Duarte Galactosemia (D/g). This is a mild form of galactosemia. Most babies do not have symptoms and appear to be healthy. If untreated, there can be some health problems associated with the liver in the first few years of life. Treatment is usually terminated by two years of age.

## What Causes Galactosemia?

Galactosemia is a genetic condition caused by a change in the GALT (Galactose-1-Phosphate Uridyltransferase) gene. The GALT gene is responsible for making an enzyme called galactose-1-phosphate uridyltransferase. The GALT enzyme is responsible for changing galactose to glucose. When there is a change in the GALT gene (G), the GALT enzyme is absent or insufficient and galactose builds up in the blood.

Two other intermediate metabolites, called Gal-1-P (galactose 1 phosphate) and galactitol, can also build up in the blood and have a toxic effect. These rare forms of galactosemia are caused by a deficiency in other enzymes, respectively called Epimerase and Galactokinase.

Classical galactosemia is inherited in an autosomal recessive pattern, which means two copies of the GALT gene must be changed for a person to have classic galactosemia. Most often, the parents of a child with an autosomal recessive condition are not affected because they are "carriers," with one copy of the changed gene (G) and one copy of the normal gene (N).

When both parents are carriers, there is a one-in-four (or 25%) chance that both will pass the changed GALT gene on to a child, causing the child to be born with the classic galactosemia (GG). There also is a one-in-four (or 25%) chance that they will each pass on a normal GALT gene, and the child will be free of the condition (NN). There is a two-in-four (or 50%) chance that a child will inherit a changed GALT gene from one parent and a normal GALT gene from the other, making that child a carrier like the parents (GN). These chances are the same in each pregnancy with the same parents.

In the case of Duarte galactosemia, a child with the condition has inherited a variant GALT gene (D) and a changed GALT gene (G). This variant is at least ten times more common than classic galactosemia.

### **Is There a Test for Galactosemia?**

Yes. Babies are tested through newborn screening for galactosemia before they leave the hospital. The baby's heel is pricked and a few drops of blood are taken. The blood is sent to the state laboratory to find out if it has more than a normal amount of galactose in the blood. Most states are currently screening for galactosemia, and Tennessee is one of them.

### **Can Galactosemia Symptoms Be Prevented?**

Yes. In most cases, taking galactose and lactose out of the diet can prevent most of the symptoms of classic galactosemia. This special diet should begin as soon as possible following diagnosis. Children and adults with classic galactosemia require follow-up care at a medical center or a clinic that specializes in this condition. Galactose and lactose are in many different foods, so an experienced metabolic dietician or nutritionist will provide a special diet that has reduced amounts of galactose and lactose. In addition, regular blood tests are used to monitor the metabolite levels.

Treatment for Duarte galactosemia can vary. An individual with Duarte galactosemia may be monitored on a galactose and lactose reduced diet for six to twelve months. Following this period of time, a "challenge" will be performed with lactose formula and a blood test will be performed to check the levels of metabolites. Consult with your Regional Genetic Center concerning treatment recommendations for Duarte galactosemia.

**DISCLAIMER:** The information contained on this page is not intended to replace the advice of a genetic metabolic medical professional.

### **Resources:**



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1500 Highland Avenue  
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### **References:**

- Gene Tests (Galactosemia) <http://www.genetests.org>

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- Online Mendelian Inheritance in Man (OMIM topic 230400)  
<http://www.ncbi.nlm.nih.gov/Omim>