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

Disorder name: **Duarte galactosemia**
Acronym: **DG deficiency**

This fact sheet has general information about Duarte galactosemia (DG). New research has shown that most children with Duarte galactosemia never have any symptoms. There is no standard recommended treatment at this time. All children with Duarte galactosemia should be followed by a healthcare provider. Most children with Duarte galactosemia are discovered to have this condition by newborn screening for other more serious forms of galactosemia.

Classic galactosemia is a separate disorder from Duarte galactosemia. This factsheet is not intended to provide information about classic galactosemia. A factsheet on classic galactosemia can be found here.

What is Duarte galactosemia?

Duarte galactosemia is a specific type of galactosemia. Galactosemias are a group of rare inherited conditions. Galactosemia means “galactose in the blood.” People with Duarte galactosemia digest a type of sugar called galactose more slowly. Galactose is found in milk and all foods that contain milk. Because they may not be able to break galactose down efficiently, it can build up in their blood. However, most babies with newborn screening results showing Duarte galactosemia never have symptoms.

What causes Duarte galactosemia?

In order for the body to use different types of carbohydrates and sugars from the food we eat, special enzymes break them down into a smaller sugar 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.

Duarte galactosemia occurs when an enzyme, called ‘galactose-1-phosphate uridyl transferase’ (GALT), is not working properly. This enzyme’s job is to change galactose into glucose. When the GALT enzyme is not working properly, galactose cannot be changed to glucose efficiently and galactose may build up in the blood. The decision of whether or not to treat someone with Duarte galactosemia depends, in part, on the level of galactose in the blood.

Types of Galactosemia

There are different types of galactosemia.

- Classic galactosemia is a rare, serious, life-threatening disorder.
- Duarte galactosemia is more common and usually causes no symptoms.

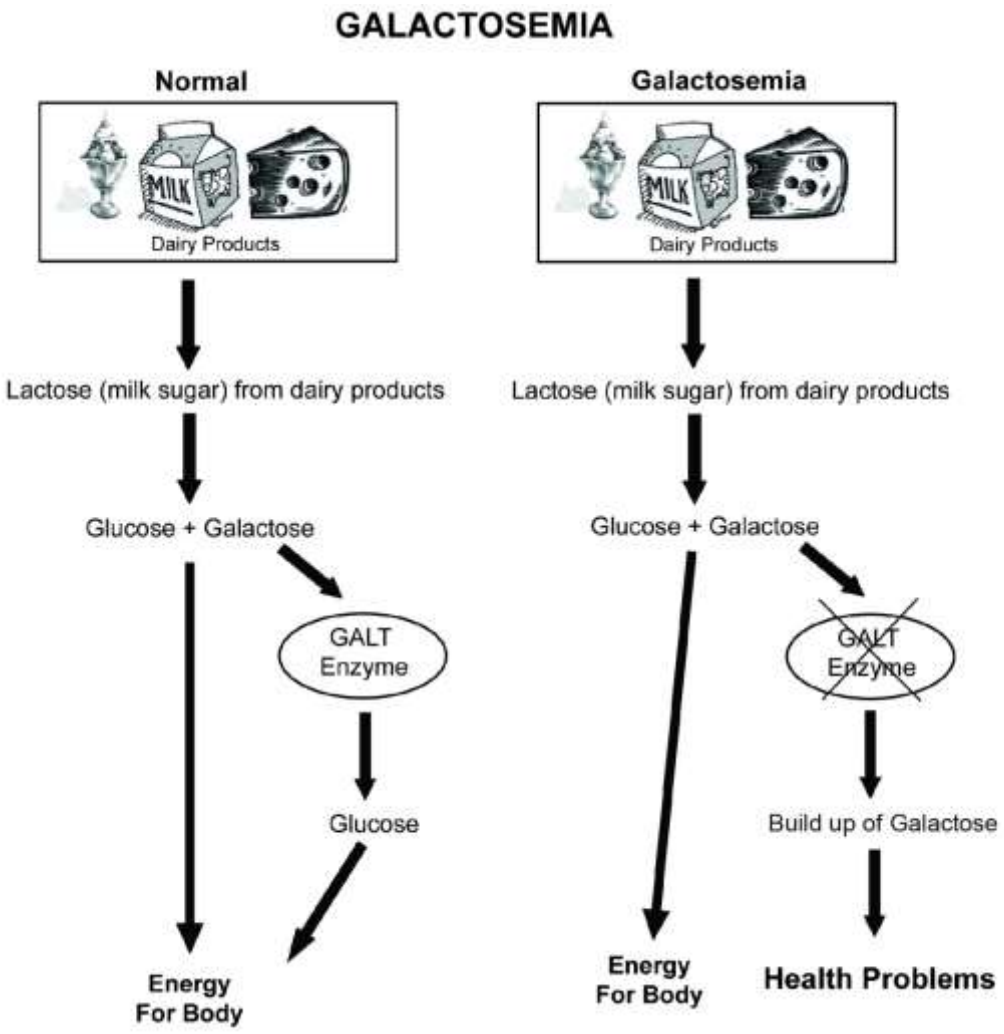
New research has shown that people with Duarte galactosemia do not develop symptoms related to this condition and may not need treatment. Your healthcare provider will help decide how to manage your child’s Duarte galactosemia.

Classic galactosemia

Classic galactosemia occurs when babies have no or very small amounts 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. This factsheet provides more information on classic galactosemia.

Duarte galactosemia

Duarte galactosemia is a more common form of galactosemia that is often, but not always, detected during newborn screening. Duarte galactosemia occurs when babies have smaller amounts of and less efficient forms of the GALT enzyme. Because they have some functioning GALT enzyme, babies with Duarte galactosemia break down galactose slower than other babies. Babies with Duarte galactosemia may need less treatment or no treatment at all. DNA testing, described below, and other blood tests can help determine if your baby has classic or Duarte galactosemia.



If Duarte galactosemia is not treated, what problems occur?

Most children with Duarte galactosemia have no symptoms at all and most do not need treatment. New research studies have shown that children with Duarte galactosemia are not at increased risk to have developmental problems related

to their intake of galactose. Based on the findings of these studies, healthcare providers are re-evaluating if, and how, they should treat infants with Duarte galactosemia. Your metabolic specialist will help you determine if your child needs treatment. Children with Duarte galactosemia may develop jaundice in the newborn period. Jaundice is a common problem in newborn babies and usually resolves over time. Girls with Duarte galactosemia are not known to have premature ovarian failure which can be seen in girls with classic galactosemia.

What is the treatment for Duarte galactosemia?

Your baby's primary doctor will work with a metabolic doctor and a dietitian familiar with Duarte galactosemia to care for your child. Children with mild Duarte galactosemia will probably not need treatment. Babies and young children with Duarte galactosemia may need regular blood and urine tests. The test results will help your doctors and dietitian determine if treatment is necessary. Your metabolic doctor and dietitian will let you know if your child needs to avoid lactose and galactose. If so, they will give you a special food plan designed to keep your child healthy.

Treatment, if needed, consists of the use of a special lactose-free formula and/or a lactose and galactose free diet. The special food plan, if used, is only during the first year of life because children with Duarte galactosemia develop an increased tolerance for dietary galactose as they grow.

What happens when Duarte galactosemia is treated?

Because most individuals with Duarte galactosemia never develop symptoms because of this condition, it is unclear if there is a benefit to early treatment. Children with Duarte galactosemia, regardless of whether they are treated or not, are able to live healthy lives with typical growth and development.

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 Duarte galactosemia have changes, also called variants, in both copies of their GALT genes. Because of the variants in the GALT genes, the GALT enzyme works more slowly than normal.

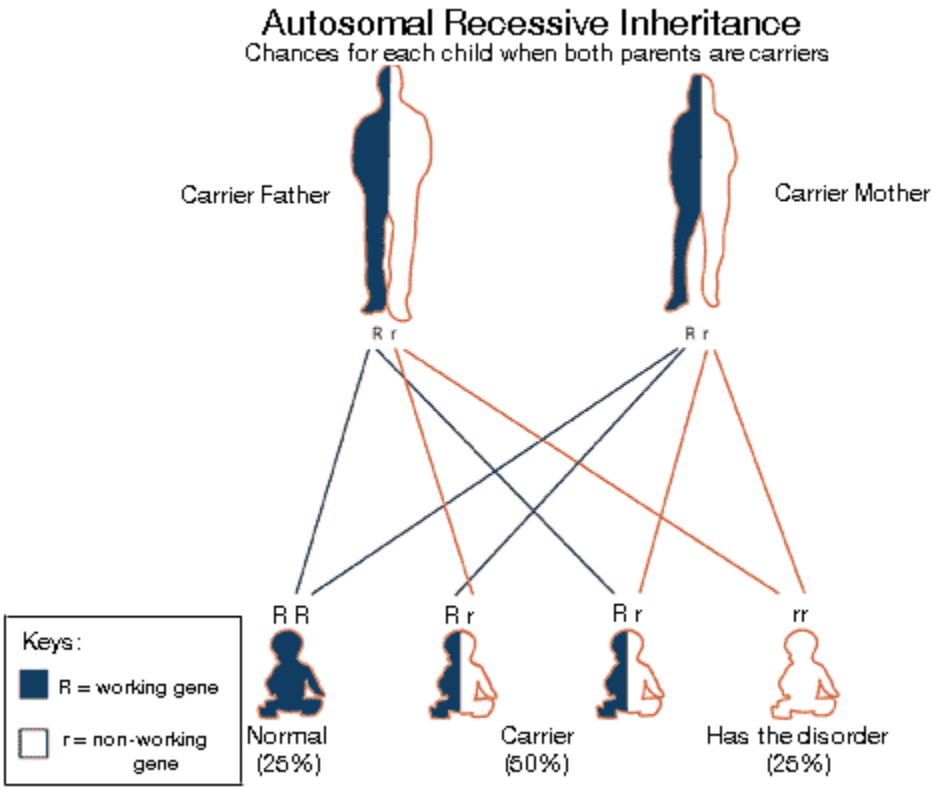
How is Duarte galactosemia inherited?

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

Everyone has two copies of the GALT gene, one inherited from each parent, that make the GALT enzyme. In children with Duarte galactosemia, neither of their GALT genes works correctly. There are two types of non-working genes in Duarte galactosemia. One is very mild, called “D” for “Duarte variant.” The other is called “G” and is a more serious variant. A child with Duarte galactosemia has either two copies of the mild “D” variant, or one copy of the mild “D” variant and one copy of the “G” variant. Because they have the mild “D” variant, children with Duarte galactosemia have GALT enzymes that works slower than usual.

Parents of children with Duarte galactosemia rarely have the condition themselves. Instead, each parent has a single non-working GALT gene for Duarte galactosemia. They are called carriers. Carriers do not have Duarte galactosemia because their other gene is working correctly. Without testing the parents, it is not possible to know which variant, D or G, each parent carries.

When both parents are carriers, there is a 25% chance in each pregnancy for the child to have Duarte 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 Duarte galactosemia. Genetic counselors can answer your questions about how Duarte

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 Duarte galactosemia can be done on a blood sample. Genetic testing, also called DNA testing, looks for changes (variants) in the pair of genes that causes Duarte galactosemia. Over 99% of the time, DNA testing can identify both gene variants 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 Duarte galactosemia your child has.

DNA testing can also be helpful for carrier or prenatal testing, discussed below.

What other testing is available?

If your child has had a positive newborn screen for Duarte galactosemia, 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 Duarte galactosemia.

Ask your doctor if you have any questions about testing for Duarte galactosemia.

Can you test during a future pregnancy?

If both gene changes have been found in your child with Duarte galactosemia, DNA testing can be done during any future pregnancies. However, prenatal testing is rarely done for Duarte galactosemia because symptoms, if any, are mild. If you have questions about prenatal testing, ask your genetic counselor or physician. Parents may choose to have testing during pregnancy or wait until birth to have the baby tested. A genetic counselor can talk to you about your choices and answer questions about prenatal testing or testing your baby after birth.

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

Having Duarte galactosemia

The brothers and sisters of a baby with Duarte galactosemia have a chance of also having Duarte galactosemia. Ask your metabolic doctor whether your other children should be tested.

Duarte galactosemia carriers

Brothers and sisters who do not have Duarte 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 Duarte galactosemia, your brothers and sisters have a 50% chance to be a carrier. Because some of the GALT gene variants that cause Duarte galactosemia can also cause classic galactosemia (more serious disease), 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 classic galactosemia or Duarte galactosemia depending on which gene change they carry.

All 50 US states offer newborn screening for classic galactosemia. Newborns with Duarte variant galactosemia may or may not be detected by the same newborn screening test that detects classic galactosemia. Specifically, some newborn screening protocols are designed to detect Duarte variant galactosemia, while others do not. In families in which a child has Duarte galactosemia, newborn screening results are not sufficient to rule out this condition in future siblings.

Can other family members be tested?

Diagnostic testing

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

About one in every 3,000 to 6,000 babies in the United States is born with Duarte galactosemia.

Does Duarte galactosemia happen more often in a certain ethnic group?

Duarte galactosemia occurs in people of all ethnic groups around the world. It is more common among infants of European ancestry and less common among infants of African, African American, or Asian ancestry.

Does Duarte galactosemia go by any other names?

Duarte galactosemia is sometimes also called:

- Duarte variant galactosemia
- DG

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

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

Where can I find more information?

Duarte Galactosemia
http://www.Duartegalactosemia.org

Baby's First Test
http://www.babysfirsttest.org

Document Info	**Created by:**	www.newbornscreening.info
	Reviewed by:	HI, CA, OR, and WA metabolic specialists
	Review date:	6/17/2020 2/28/2017
	Update on:	6/17/2020

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This project is supported by a grant from the Maternal and Child Health Bureau, Health Resources and Service Administration, Genetic Services Branch, MCH Project #: UH7MC30774-01-00 http://mchb.hrsa.gov