

# GALACTOSEMIA, *GALT*-RELATED

## GENETIC TESTING



### What is galactosemia, *GALT*-related?

Galactosemia, *GALT*-related is an inherited disease that in its classic, untreated form is characterized by life-threatening complications in the newborn period, intellectual disabilities, and speech difficulties. The symptoms of galactosemia, *GALT*-related are due to a defect in the production of an enzyme called galactose-1-phosphate uridylyl transferase (*GALT*), which is responsible for breaking down a sugar called galactose. Galactose is found in milk, breast milk, infant formula, and dairy products. Without this enzyme, galactose accumulates in various tissues of the body and acts as a toxin.<sup>1</sup> Galactosemia, *GALT*-related is also known as *GALT* deficiency or classic galactosemia.<sup>2</sup>

### What are the symptoms of galactosemia, *GALT*-related and what treatment is available?

Individuals with galactosemia, *GALT*-related can develop life-threatening complications in the newborn period, shortly after the introduction of galactose into the diet.<sup>1</sup>

Symptoms of untreated galactosemia, *GALT*-related may include<sup>1,2</sup>:

- Feeding problems/diarrhea
- Failure to thrive (not gaining weight or growing well)
- Lethargy (low energy)
- Hypotonia (low muscle tone)
- Jaundice (yellowing of the skin and eyes)
- Hepatomegaly (enlarged liver)
- Infection
- Cataracts
- Bleeding tendencies
- Neonatal death

While there is no cure for galactosemia, placing infants on a galactose-free diet within the first 10 days of life can resolve or prevent neonatal symptoms. Long-term management usually includes avoidance of galactose-containing foods throughout life. Even with this regimen, however, individuals with galactosemia are at increased risk for cognitive and developmental delay, verbal apraxia (speech problems), and (in females) premature ovarian insufficiency.<sup>2</sup>

Galactosemia is included in newborn screening panels in all 50 states.<sup>3</sup> Newborn screening may identify babies with galactosemia, *GALT*-related as well as milder versions that may not require treatment.<sup>2</sup>

### How is galactosemia, *GALT*-related inherited?

Galactosemia, *GALT*-related is an autosomal recessive disease caused by mutations in the *GALT* gene.<sup>2</sup> An individual who inherits one copy of a *GALT* gene mutation is a “carrier” and is not expected to have related health problems. An individual who inherits two classic mutations in this gene, one from each parent, is expected to be affected with galactosemia, *GALT*-related. Inheritance of other mutation combinations may result in galactosemia, *GALT*-related or a milder presentation.

If both members of a couple are carriers, the risk of having an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

### Who is at risk for galactosemia, *GALT*-related?

Galactosemia, *GALT*-related can occur in individuals of all races and ethnicities.

Having a relative who is a carrier or is affected can also increase an individual’s risk of being a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

## Incidence and Carrier Frequency in Select Ethnic Groups

	Incidence	Carrier Frequency
African American	1 in 24,000 <sup>4</sup>	1 in 78
Caucasian	1 in 47,000 <sup>4</sup>	1 in 108
Ashkenazi Jewish	1 in 64,500	1 in 127 <sup>1</sup>

### What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetics health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

### What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

### Where can I get more information?

- Galactosemia Foundation, formally, Parents of Galactosemic Children, Inc. (PGC):  
<http://www.galactosemia.org>

**Note:** This material is provided for general information purposes only. It is not intended as a substitute for medical advice and/or consultation with a physician or technical expert.

#### References

- Goldstein N. et al. The GALT rush: High carrier frequency of an unusual deletion mutation of the GALT gene in the Ashkenazi population. *Mol Genetics and Metabolism*. 2011; 102: 157-160.
- Elsas L. Galactosemia. GeneReviews. Available at: <http://www.ncbi.nlm.nih.gov/books/NBK1518/>. Accessed February 14, 2012.
- National Newborn Screening Status Report: Updated 2/15/12. Available at <http://genes-r-us.uthscsa.edu/nbsdisorders.pdf>.
- Suzuki M et al. Large-scale molecular screening for galactosemia alleles in a pan-ethnic population. *Hum Genet*. 2001 Aug;109: 210-215.



[www.LabCorp.com](http://www.LabCorp.com)

#### Contact Us

For more information about LabCorp, the testing services we provide, and where to find a specimen collection lab near you, visit [www.labcorp.com](http://www.labcorp.com).