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CARRIER SCREEN

Galactosemia, *GALT*-related

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 uridyl 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 the GALT enzyme, galactose accumulates in various tissues of the body and acts as a toxin.[1] Galactosemia, *GALT*-related, is also known as GALT deficiency or classic galactosemia.[2]

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

Individuals with galactosemia can develop life threatening complications in the newborn period, shortly after the introduction of galactose into the diet.[1]

Symptoms of untreated galactosemia, *GALT*-related, may include:[1,2]

- 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.[2]

Galactosemia is included in newborn screening panels in all 50 states.[3]

How is galactosemia, *GALT*-related inherited?

Galactosemia, *GALT*-related, is an autosomal recessive disease caused by mutations in the *GALT* gene.[2] 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 mutations in the *GALT* gene, one from each parent, is expected to be affected with galactosemia.

If both members of a couple are carriers of a mutation in the same gene, 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.

Incidence and Carrier Frequency in Select Ethnic Groups

Population	Incidence	Carrier frequency
African American	1 in 24,000[4]	1 in 78
Caucasian	1 in 47,000[4]	1 in 108
Ashkenazi Jewish	1 in 64,500	>1 in 127[1]

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.

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/

Genetics Home Reference: http://ghr.nlm.nih.gov/condition/galactosemia

References

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3. National Newborn Screening Status Report. November 2, 2014. Available at: http://genes-r-us.uthscsa.edu/ sites/genes-r-us/files/nbsdisorders.pdf . Accessed January 6, 2016.
4. Suzuki M *et al*. Large-scale molecular screening for galactosemia alleles in a pan-ethnic population. *Hum Genet*. 2001 Aug; 109:210-215.