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. Galactosemia, GALT-related, is also known as GALT deficiency or classic galactosemia.

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. Symptoms of untreated galactosemia, GALT-related, may include:

- 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.

Galactosemia is included in newborn screening panels in all 50 states.

How is galactosemia, GALT-related inherited?

Galactosemia, GALT-related, is an autosomal recessive disease caused by mutations in the GALT gene. 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

<table>
<thead>
<tr>
<th>Population</th>
<th>Incidence</th>
<th>Carrier frequency</th>
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</thead>
<tbody>
<tr>
<td>African American</td>
<td>1 in 24,000*</td>
<td>1 in 78</td>
</tr>
<tr>
<td>Caucasian</td>
<td>1 in 47,000*</td>
<td>1 in 108</td>
</tr>
<tr>
<td>Ashkenazi Jewish</td>
<td>1 in 64,500</td>
<td>&gt;1 in 127</td>
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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?


References