What is galactosialidosis?

Galactosialidosis is an inherited lysosomal storage disorder. Individuals with galactosialidosis have a defect in the cathepsin A protein. Cathepsin A normally interacts with other enzymes in lysosomes to break down oligosaccharides (sugars) attached to some proteins and fats. Cathepsin A also has a role in the normal development of connective tissue. When cathepsin A is defective, it cannot interact with other enzymes. Symptoms of galactosialidosis are due to substances accumulating in lysosomes and problems with the development of connective tissue as result of defective cathepsin A.

What are the symptoms of galactosialidosis and what treatment is available?

Galactosialidosis is a disease with variable severity and age at onset. Three forms are recognized.

An early infantile form is usually diagnosed between birth and three months of age. Life expectancy is into late infancy. Symptoms may include:

- Hydrops fetalis (abnormal fluid accumulation in a fetus)
- Hepatosplenomegaly (enlarged liver and spleen)
- Dyostosis multiplex (skeletal abnormalities seen by X-ray)
- Distinctive facial features
- Cherry-red spot in the eye
- Kidney disease

A late infantile form is usually diagnosed within the first year of life. Life expectancy varies and depends on the severity of symptoms. Symptoms may include:

- Hepatosplenomegaly
- Dysostosis multiplex
- Distinctive facial features
- Cherry-red spot in the eye
- Short stature
- Heart valve problems
- Intellectual disability
- Hearing loss

A juvenile/adult form is diagnosed at varying ages, with symptoms appearing on average at the age of 16. Life expectancy is normal. Symptoms may include:

- Ataxia (loss of muscle coordination) and myoclonus (muscle twitches)
- Angiokeratomas (benign skin lesions)
- Distinctive facial features
- Cherry-red spot in the eye
- Seizures
- Vision and hearing loss
- Progressive intellectual disability

There is no cure for galactosialidosis. Treatment is supportive.
How is galactosialidosis inherited?

Galactosialidosis is an autosomal recessive disease caused by mutations in the CTSA gene. An individual who inherits one copy of a CTSA gene mutation is a carrier and is not expected to have related health problems. An individual who inherits two CTSA mutations, one from each parent, is expected to be affected with galactosialidosis.

If both members of a couple are carriers of mutations in the same gene, the risk for 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 galactosialidosis?

Galactosialidosis is a very rare condition and its prevalence is unknown.

Having a relative who is a carrier or who is affected can increase an individual's risk to be 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?


The International Advocate for Glycoprotein Storage Diseases: http://www.ismrd.org/glycoprotein_diseases/galactosialidosis

References