Galactosemia

What is Galactosemia?

Lactose is the main carbohydrate in breast milk and most non-soy infant formulas and is broken down into glucose and galactose in the intestine. Individuals with galactosemia are not able to utilize galactose because an enzyme, called GALT (galactose-1-phosphate uridyl transferase), is defective or deficient. This leads to an accumulation of galactose and other harmful substances in the blood and urine, which can cause serious health problems. Some individuals have a milder form of the condition in which there is some GALT activity.

What is its incidence?

The incidence of classic galactosemia has been estimated to be approximately 1 in 40,000, although the numbers will vary according to different sources.

What causes the disease?

Mutations in the GALT gene produce an enzyme with deficient activity.

What are the clinical features of the disease?

Although babies with galactosemia are normal at birth, they may have serious problems without treatment. The inability to metabolize galactose can result in life-threatening complications including hypoglycemia, feeding problems, failure to thrive, liver damage, lethargy, bleeding, and sepsis. Even with early treatment, however, children with galactosemia are at increased risk for developmental delays, speech problems, abnormalities of motor function. cataracts, and, in females, premature ovarian failure.

How is the diagnosis confirmed?

Newborn screening for galactosemia involves the measurement of GALT enzyme activity in a dried blood spot. The diagnosis of galactosemia can be confirmed by measuring the amount of galactose and galactose-1- phosphate in the blood. Genetic testing to look for mutations in the GALT gene may also assist in confirming the diagnosis. Diagnostic testing is arranged by specialists at BC Children's Hospital.

What is the treatment of the disease?

A galactose-restricted diet is effective in preventing many of the complications of galactosemia, including the liver and kidney problems. It may also reduce the risk for developmental delays.

Treatment is coordinated by specialists at your BC Children's Hospital.

What is the outcome of treatment?

Although early identification and treatment will ensure the best outcome, some individuals with galactosemia are still at increased risk to develop complications, as discussed in Clinical Features.

Can a family have more than one child with Galactosemia?

Galactosemia is inherited as an autosomal recessive disease. Parents of a child with galactosemia are assumed to be carriers for the disease and have a 1 in 4 (25%)

The BC Newborn Screening Program is a collaboration of BC Women's Hospital & Health Centre, BC Children's Hospital and the BC Perinatal Health Program, all part of the Provincial Health Services Authority.

Provincial Health Services Authority
Province-wide solutions. Better health.

chance, in each pregnancy, of having another child with this condition. Prenatal testing for galactosemia can be done as early as 15-16 weeks of pregnancy. Genetic counselling to discuss the benefits of prenatal testing options in more detail is recommended.

Unaffected siblings of a child with galactosemia have a 2/3 chance of being carriers. Carriers are healthy and do not have symptoms of galactosemia.

Resources

http://www.galactosemia.org/
http://www.galactosemia.com/
http://www.dshs.state.tx.us/newborn/galac_1.shtm
http://www.geneclinics.org

Revised November 2009

The BC Newborn Screening Program is a collaboration of BC Women's Hospital & Health Centre, BC Children's Hospital and the BC Perinatal Health Program, all part of the Provincial Health Services Authority.

Provincial Health Services Authority
Province-wide solutions. Better health.