Kansas
Department of Health and Environment

Classical Galactosemia
Information for Parents

Overview

Classical galactosemia is an inherited defect of galactose metabolism. It is caused by an enzyme deficiency that prevents the body from metabolizing galactose, or milk sugar, into glucose. The main dietary source of galactose is lactose, and is found in all forms of milk, except soy.

What is Classical Galactosemia?

Classical galactosemia is a treatable disorder. It affects the way the body processes the sugar galactose, a component of milk and dairy products. Children with classical galactosemia cannot process galactose. As a result, galactose and other by-products build up in the bloodstream and cause physical and developmental damage. Poor mental and physical growth, cataracts, and serious liver and kidney problems are just a few of the possible effects of this disorder. In a child with classical galactosemia, galactose cannot be converted to glucose because the GALT enzyme does not work properly. Classical galactosemia can be life threatening in the first few weeks of life. Milder variants of galactosemia are not associated with serious complications. Galactosemia and lactose intolerance are totally separate medical conditions.

Why is newborn screening done for Classical Galactosemia?

Newborn screening is done for classical galactosemia and other milder forms of galactosemia so babies with this condition can be diagnosed quickly. If babies are diagnosed quickly, treatment can begin immediately, reducing the chances of permanent damage from the accumulation of galactose and its by-products in the body.

Does a positive result from the Kansas Newborn Screening Lab mean that my baby has Classical Galactosemia?

No, not necessarily. Newborn screening tests the baby's level of GALT or Galactose-1-phosphate uridyl transferase enzyme. The level was outside of the normal range in your baby. **Prompt follow up is important to help distinguish classical galactosemia from other benign forms**. Additional tests will need to be done to determine if your baby has classical galactosemia or not.

How common is Classical Galactosemia?

Classical galactosemia occurs in approximately 1 in 50,000 births.

How is Classical Galactosemia inherited?

Classical galactosemia is inherited in an autosomal recessive pattern. The parents of a child diagnosed with galactosemia do not have the condition. Parents are carriers of the condition. Carriers have one normal copy of the gene for the GALT enzyme and one abnormal copy. In order to have classical galactosemia, a child must inherit two abnormal copies of the gene, one from each parent. Each pregnancy between carrier parents has a 25% chance of producing a child affected with galactosemia, a 50% chance of producing an

unaffected carrier child, and a 25% chance of producing a child who is unaffected and is not a carrier.

What are the signs and symptoms of Classical Galactosemia?

Signs and symptoms of classical galactosemia appear in the newborn period. Symptoms may include:

- Problems with feeding
- Vomiting
- Diarrhea
- Yellowing of skin or whites of the eyes (jaundice)
- Over-sleepiness
- Excessive bleeding
- Swollen belly due to liver enlargement
- Cataracts (clouding of the eyes)

How is Classical Galactosemia diagnosed?

Kansas newborn screening measures the activity of the GALT enzyme in your baby's blood. If your baby has diminished GALT activity and has no other symptoms, the newborn screening test should be repeated. **An absence of GALT activity in a newborn screen requires immediate suspension of breastfeeding or cow milk-based formula; soy-based formula feedings should be started.** The child should be evaluated as soon as possible for symptoms of galactosemia and referred to a metabolic specialist for diagnostic testing. The baby's enzyme levels will be re-measured and biochemical or genetic testing may be performed to confirm the diagnosis. Diminished GALT enzyme level means that your child most likely has a benign form of galactosemia that may not need treatment**.**

Is there a cure for Classical Galactosemia?

No, there is no cure for classical galactosemia. Individuals who are diagnosed and treated before damage from galactose accumulation occurs can develop and grow normally. Even with early diagnosis and strict adherence to the diet, some children remain at risk for some problems, including growth retardation or developmental delays (particularly speech delays). Females with classical galactosemia are at risk for ovarian failure. The galactose free diet must be followed for life and requires close medical supervision.

How is Classical Galactosemia treated**?**

Classical galactosemia is treated by removing all lactose and galactose from the diet.

Where can I get additional information?

Genetic Fact Sheets for Parents:
http://www.newbornscreening.info/Parents/otherdisorders/Galactosemia.html

Parents of Galactosemic Children: http://www.galactosemia.org/

Children Living with Inherited Metabolic Disorders (CLIMB): http://www.climb.org.uk