

GALACTOSEMIA

(Guh-LAK-toe-SEE-me-ah)

What is it?

Galactosemia is an inherited defect of galactose metabolism caused by an enzyme deficiency in the liver that prevents proper metabolism and utilization of galactose or milk sugar. Galactose is a breakdown product of lactose, which is most commonly found in milk products. When galactose cannot be broken down, it builds up in the cells and becomes toxic.

How do you get it?

Galactosemia is inherited in an autosomal recessive pattern. As an autosomal recessive disorder, the parents of a child with galactosemia are unaffected, healthy carriers of the condition have one normal gene and one abnormal gene. With each pregnancy, carrier parents have a 25 percent chance of having a child with two copies of the abnormal gene, resulting in classical galactosemia. Carrier parents have a 50 percent chance of having a child who is an unaffected carrier and a 25 percent chance of having an unaffected, non-carrier child. These risks would hold true for each pregnancy. All siblings of infants confirmed to have galactosemia also should be tested; genetic counseling services should be offered to the family.

Although infants with galactosemia may appear normal at birth, within a few days to two weeks after initiating breast milk or formula feedings, the symptoms of untreated galactosemia can become very severe. Early signs of the disease include feeding problems, poor sucking reflex, jaundice and hepatomegaly. Other symptoms may include failure to thrive, lethargy, cataracts, hypoglycemia, coagulation problems, developmental retardation and decreased immunity. Sepsis due to *Escherichia coli* (*E. coli*) seems to be particularly frequent among galactosemic neonates and is usually the cause of death.

How common is it

The incidence of classical galactosemia is 1 in 40,000 births in Missouri.

How is it treated

When a lactose-restricted diet is provided within the first 10 days of life, presenting symptoms may be reversed. Infants with galactosemia are started on milk substitute formula, most likely a lactose-free soybean protein formula. Galactose is a non-essential nutrient, and individuals diagnosed

with classical galactosemia require lactose-restricted diets for life. Endogenous production of galactose can complicate dietary treatment of galactosemia and may result in some developmental delays. Close dietary supervision, monitoring and the assistance of a trained dietician are required for infants and children diagnosed with classical galactosemia. Caution concerning administration of anesthesia and certain drugs that may contain lactose is also necessary.

If your child needs additional testing or diagnostic evaluation, it is important that you follow through with the pediatrician's and/or specialist's recommendations for additional testing and referrals.

These guidelines should be followed after a diagnosis of galactosemia has been confirmed:

Infants and children with galactosemia should have regular follow-up appointments with a metabolic disease specialist.

Treatment is not curative and all morbidity cannot necessarily be prevented. Long-term management, monitoring and compliance with treatment recommendations are essential to the child's well being. A multidisciplinary approach is recommended and should include the following specialties: pediatrics, genetics and nutrition.

With the lactose-restricted diet and the child's inability to consume dairy products, the specialist will probably recommend calcium supplements.

Regular appointments with an ophthalmologist to check for the possible development of cataracts are recommended; developmental assessment and speech therapy also may be indicated.

Where can I get services?

Provision of the names below does not necessarily include all hospitals or private practice physicians who may treat children with galactosemia.

Cardinal Glennon Memorial Hospital for Children
St. Louis, MO
314-577-5639

Children's Mercy Hospital
Kansas City, MO
816-234-3804

St. Louis Children's Hospital
St. Louis, MO
314-454-6051

University of Missouri Children's Hospital
Columbia, MO
573-882-6991

Related Links

Medline Plus (National Library of Medicine and the National Institute of Health) www.medlineplus.gov

National Institutes of Health www.nih.gov

National Newborn Screening and Genetics Resource Center
<http://genes-r-us.uthscsa.edu>

GeneTests <http://www.genetests.org>

Parents of Galactosemia Children www.galactosemia.org

Save Babies Through Screening Foundation
4 Manor View Circle Malvern, PA 19355-1622 Toll Free Phone: 1-888-454-3383
Fax: (610) 993-0545 Email: email@savebabies.org
Website: <http://www.savebabies.org/diseasedescriptions.php/>

American College of Medical Genetics
Newborn Screening ACT Sheets and Confirmatory Algorithms
<http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm>

Cardinal Glennon Children's Hospital
St. Louis, Missouri 314-577-5639
Website: <http://pediatrics.slu.edu/index.phtml?page=geneticsdiv>

Children's Hospital at University Hospital and Clinics
Columbia, Missouri 573-882-6991
Website: <http://www.genetics.missouri.edu/>

Children's Mercy Hospital
Kansas City, Missouri
816-234-3290

Website: <http://www.childrens-mercy.org/content/view.aspx?id=155>

St. Louis Children's Hospital
St. Louis, Missouri
314-454-6093

Website: <http://www.peds.wustl.edu/genetics/>