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## **Galactosemia**

### **Information for Physicians and Other Health Care Professionals**

**Definition** Galactosemia is an inherited defect of galactose metabolism caused by an enzyme deficiency that prevents proper metabolism and utilization of galactose, or milk sugar. The main dietary source of galactose is lactose, the principle carbohydrate found in all forms of milk.

**Clinical Symptoms** Although infants with galactosemia may appear normal at birth, within a few days to two weeks after initiating milk 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 and decreased immunity.

**Newborn Screening and Definitive Diagnosis** In Illinois, newborn screening for galactosemia is designed to detect classical galactosemia due to a deficiency of the galactose-1-phosphate uridyl transferase (GALT) enzyme; primary screening is performed by fluorometric assay. This test determines the level of galactose in the blood specimen and, when an elevated level is detected, Beutler assay of the specimen is performed to measure GALT enzyme activity. False negative and false positive results are possible with these tests. Infants who have not yet received a lactose (milk) feeding or those who are on soy formula may not have elevated galactose levels and should receive Beutler assay regardless of the galactose level. Infants who have received transfusions prior to specimen collection may have false negative Beutler enzyme results due to the GALT activity of transfused red blood cells. **The type of infant feeding and transfusion status should always be indicated on the filter paper specimen. Infants with a presumptive positive screening test (no GALT activity detected or a seriously elevated galactose level) require prompt follow-up. When receiving a presumptive positive result, the clinician should immediately check on the clinical status of the baby and refer the infant to a metabolic disease specialist. The infant's feeding should be changed to soy formula.** If screening results indicate a mildly elevated galactose level with GALT activity present, filter paper screening should be promptly repeated and, if the second screening is abnormal, the infant should be referred to a metabolic disease specialist. The GALT enzyme is susceptible to damage from heat and filter paper specimens require prompt testing.

**Treatment** Early diagnosis and treatment of classical galactosemia is imperative to prevent life threatening complications of sepsis and liver failure and to prevent additional developmental delays. Without early treatment, sepsis due to *Escherichia coli* may prove fatal in the neonatal period. 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 certain drugs that may contain lactose is also necessary.

**Incidence** The incidence of classical galactosemia is one in 60,000 births. Illinois began testing for galactosemia in 1984 and more than 70 cases of classical galactosemia, 170 carriers and 80 cases with a variant form of the disorder have been identified.

**Inheritance Pattern** 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 and 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.

**Physiology** Galactose is present in all milk sources and must be metabolized to glucose for absorption from the intestine. Individuals with classical galactosemia have a severe inherited deficiency of galactose-1-phosphate uridyl transferase (GALT). The GALT enzyme is one of three enzymes necessary for galactose metabolism. The biochemical consequence of GALT deficiency is abnormally high concentrations of galactose and its metabolites in body tissues and fluids. Classical galactosemia may result in life threatening crisis in the early neonatal period due to liver dysfunction, bleeding tendencies and septicemia.

**Key Points for Parents** Avoid overly alarming the child's parents if the diagnosis has not yet been confirmed. If the child needs additional testing or diagnostic evaluation, make certain the parents understand the importance of following the pediatrician's and/or specialist's recommendations for additional testing and referrals.

**Follow-up After Confirmation of Diagnosis** These guidelines should be followed after a diagnosis of galactosemia has been confirmed:

1. Infants and children with galactosemia should have regular follow-up appointments with a metabolic disease specialist.
2. Parents should understand that treatment is not curative and that 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.
3. Genetic counseling services may be indicated. A list of counselors and geneticists, whose services are available through the Illinois Department of Public Health, should be given to the parents if they have not already seen a geneticist.
4. Provide a list of support services available within the community, such as the local health department, Early Intervention service providers and the University of Illinois at Chicago, Division of Specialized Care for Children (DSCC).
5. Additional information about newborn screening can be found at:
  - Baby's First Test: <http://www.babysfirsttest.org/>  
Health Resource and Service Administration (HRSA), Grant no. U36MC16509, Quality Assessment of the Newborn Screening System.
  - National Center for Biotechnology Information: <http://www.ncbi.nlm.nih.gov/gtr/>  
National Center for Biotechnology Information, U.S. National Library of Medicine, 8600 Rockville Pike, Bethesda MD, 20894 USA.