TEXAS Department of State Health Services

Newborn Screening ACT Sheet

Absent/Severely Reduced Galactose-1-phosphate Uridyltransferase (GALT) **Classical Galactosemia**

Differential Diagnosis: Galactosemia (galactose-1-phosphate uridyltransferase deficiency); GALT heterozygotes; GALT variants; artifactual reductions due to enzyme inactivation by high temperature and/or humidity.

Condition Description: In galactosemia, GALT deficiency results in accumulation of galactose-1-phosphate (Gal-1-P), and galactose, causing multiorgan disease.

Medical Emergency: Take the Following IMMEDIATE Actions

- **Contact family to inform them of the newborn screening result, ascertain clinical status, arrange immediate clinical evaluation, stop breast or cow's milk, and initiate non-lactose feeding (powder-based soy formula).**
- **Consult with metabolic specialist; refer if considered appropriate.**
- **Evaluate the infant (jaundice, poor feeding, vomiting, lethargy, bulging fontanel, and bleeding), and arrange diagnostic testing as directed by metabolic specialist.**
- **Initiate emergency treatment as recommended by metabolic specialist. If baby is sick, admit to hospital.**
- **Repeat newborn screen if second screen has not yet been done.**
- **Educate family about importance of diet change.**
- **Report findings to newborn screening program.**

Confirmation of Diagnosis: Quantification of erythrocyte galactose-1-phosphate (gal-1-P) and GALT. Classical galactosemia shows <1% GALT activity and markedly increased gal-1-P.

Transfusions in infant can invalidate the results of erythrocyte enzyme assays. Enzyme variants may be distinguished by GALT electrophoresis or mutation analysis.

Clinical Considerations: Classical galactosemia presents in the first few days of life and may be fatal without treatment. Signs include poor feeding, vomiting, jaundice and, sometimes, lethargy and/or bleeding. Neonatal E. coli sepsis can occur and is often FATAL. Treatment is withdrawal of milk and, if symptomatic, emergency measures.

Additional Information:

New England Metabolic Consortium
http://www.childrenshospital.org/newenglandconsortium/NBS/gal/gal_protocol.htm

Gene Tests/Gene Clinics
http://www.genetests.org/servlet/access?db=geneclinics&site=gt&id=8888891&key=0JvJTPALRrmnH&gry=&fcn=y&fw=SCnL&filename=/profiles/galactosemia/index.html

Genetics Home Reference
http://ghr.nlm.nih.gov/ghr

TEXAS Department of State Health Services

Newborn Screening FACT Sheet

Galactosemia
(GALT Deficiency)

What is Galactosemia?

Galactosemia, GALT deficiency, is a rare, inherited problem. It is caused when the body can't break down galactose. Galactose is a sugar found in milk and milk products.

What Causes Galactosemia?

Breast milk and most infant formulas have a sugar called lactose. The body breaks lactose down into sugars called glucose and galactose. Galactose must be broken down more before the body can use it for energy. An enzyme called galactose-1-phosphate uridyl transferase (GALT) helps do this. Enzymes help start chemical reactions in the body. Most people with galactosemia don't have this special enzyme. This causes galactose to build up in the body.

What Symptoms or Problems Occur with Galactosemia?

[Symptoms are something out of the ordinary that a parent notices.]

High levels of galactose poison the body and cause these serious problems:

- swollen liver
- kidney failure
- stunted growth and mental retardation
- cataracts in the eyes

Children and young adults treated for galactosemia may still have problems over the years with:

- speech
- language
- hearing
- clumsiness with hands
- bleeding in the gel-like part of the eye
- tremors (shaking)
- stunted growth
- learning disabilities

What is the Treatment for Galactosemia?

Special Diet – The treatment for galactosemia, GALT deficiency, is to limit galactose and lactose from the diet for life. All milk and all foods that have milk in them must not be used at all. This includes any kind of milk, such as cow's milk, goat's milk, and human breast milk. Your child should also not eat dairy products like butter, cheese, and yogurt. Other foods with small amounts of milk products must also not be eaten. These include foods with whey, casein, and curds.

Things to Remember

Children with galactosemia should be in the care of a doctor who specializes in the treatment of galactosemia. You will also have a dietitian who will teach you about special diets for your child. Dietitians know what are the right foods to eat.

Read labels carefully when you shop for your child's food. Many prepared foods have hidden ingredients that contain galactose.

Many medicines contain fillers that include galactose. It is important to ask the doctor and pharmacist about this for any medicines prescribed for your child.

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