TEXAS Health and Human Services | 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.

Conditions associated with this analyte have been identified by the Society of Inherited Metabolic Disorders (SIMD) as critical and require immediate action.

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);**
- **LAB: Collect GALT enzyme testing (Quantitative RBC Galactose -1 Phosphate Uridyltransferase level). Consider obtaining liver function tests and glucose levels. 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; and**
- **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

American College of Genetics and Genomics – Classical Galactosemia ACT Sheet
National Center for Biotechnology Information – Classic Galactosemia and Clinical Variant Galactosemia
U.S. National Library of Medicine, Medline Plus – Galactosemia

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