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), 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:

American College of Medical Genetics and Genomics
https://www.acmg.net/StaticContent/ACT/GalactosePlusGALT.pdf

National Center for Biotechnology Information
http://www.ncbi.nlm.nih.gov/books/NBK1518/

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