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.

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**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.

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**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**

Disclaimer: This information is adapted from the American College of Medical Genetics and Genomics (ACMG) 05/2015