



KANSAS DEPARTMENT OF HEALTH AND ENVIRONMENT

NEWBORN SCREENING ACT SHEET

SCREEN FOR: ABSENT/REDUCED GALACTOSE-1-PHOSPHATE URIDYLTRANSFERASE (GALT)

CONDITION: 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.

METABOLIC DESCRIPTION: In galactosemia, GALT deficiency results in accumulation of galactose-1-phosphate (Gal-1-P) and galactose, causing multi-organ disease.

MEDICAL EMERGENCY - ACTION TO BE TAKEN IMMEDIATELY:

- ◆ Contact the family to inform them of the newborn screening result and ascertain clinical status.
- ◆ Arrange immediate clinical evaluation; for reduced GALT with symptoms or absent GALT result, stop breast or cow milk based infant formula and initiate non-lactose feeding (powder-based soy formula)
- ◆ If reduced GALT result and infant is asymptomatic, repeat the newborn screening test.
- ◆ Consult with a specialist; refer if considered appropriate.
- ◆ Evaluate the infant (jaundice, poor feeding, vomiting, lethargy, bulging fontanel, and bleeding) and arrange diagnostic testing as directed by the specialist.
- ◆ Emergency treatment as recommended by the specialist. If baby is sick, stop breast milk and/or cow milk based formula and initiate non-lactose feedings.
- ◆ 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 EXPECTATIONS: 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 the withdrawal of human milk and cow milk based formula; if symptomatic, the necessary emergency management.

REPORTING: Report diagnostic result to family and Kansas NBS program.

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