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.

SPECIALISTS:

Bryce Heese, MD  
Biochemical Genetics  
Children's Mercy Hospital- Kansas City, MO  
Clinic phone: 816-234-3771  
Hospital Operator: 816-234-3000  
Office Fax: 816-302-9963

Dr. Siddharthan Sivamurthy  
KU Wichita Pediatrics  
Wichita, KS  
Office: 316-962-7386

DISCLAIMER: These standards and guidelines were adapted from the American College of Medical Genetics ACT sheets. They are designed primarily as an educational resource for physicians to help them provide quality medical services. Adherence to these standards and guidelines does not necessarily ensure a successful medical outcome. These standards and guidelines should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonable directed to obtaining the same results. In determining the propriety of any specific procedure or test, the healthcare provider should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient’s record the rationale for any significant deviation from these standards and guidelines.