Newborn Screening ACT Sheet

[Absent/Reduced Galactose-1-Phosphate Uridyltransferase (GALT)]

Classical Galactosemia

Differential Diagnosis: Galactosemia (galactose-1-phosphate uridyltransferase [GALT] 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 multi-organ disease.

YOU SHOULD TAKE THE FOLLOWING ACTIONS IMMEDIATELY:

- 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.
- Emergency treatment as recommended by metabolic specialist. If baby is sick, stop cow's milk and initiate non-lactose feedings.
- Educate family about importance of diet change.
- Report findings to newborn screening program.

Diagnostic Evaluation: 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:
- Gene Reviews
- Genetics Home Reference

Referral (local, state, regional and national):
- Testing
- Clinical Services
- Find Genetic Services
LOCAL RESOURCES: Insert State newborn screening program web site links

State Resource site (insert state newborn screening program website information)

Name
URL
Comments

Local Resource Site (insert local and regional newborn screening website information)

Name
URL
Comments

APPENDIX: Resources with Full URL Addresses

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Find Genetic Services
http://www.acmg.net/GIS/Disclaimer.aspx