



Newborn Screening ACT Sheet [Elevated galactose-1-phosphate] Classical Galactosemia

Differential Diagnosis:

- Galactosemia (galactose-1-phosphate uridylyltransferase 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.

MEDICAL EMERGENCY: TAKE THE FOLLOWING IMMEDIATE ACTIONS:

- Consult with metabolic specialist; referral 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 state newborn screening program.

(See flowchart for information concerning the specific actions)

You Should Take the Following Actions:

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:

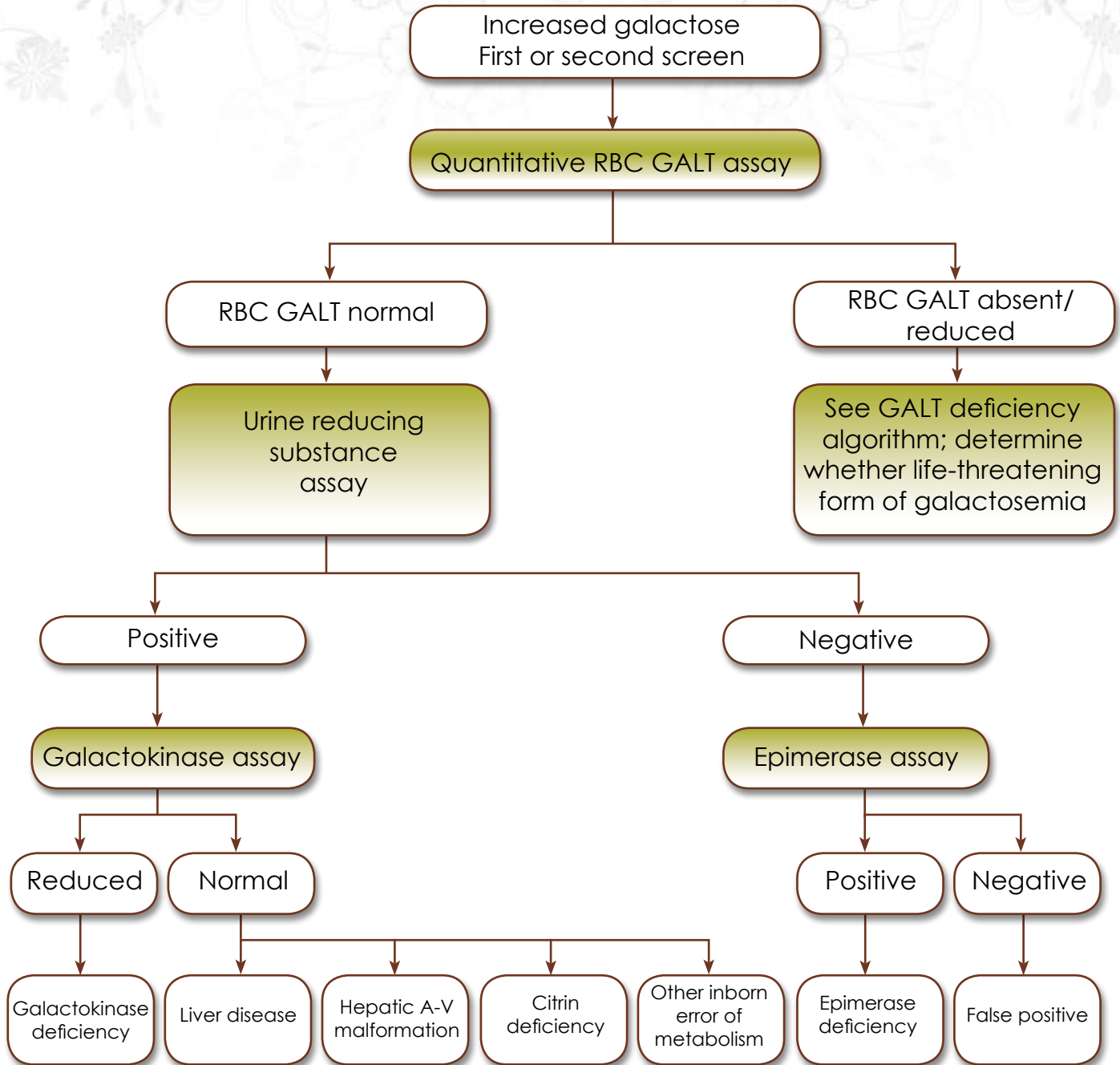
Classical galactosemia presents in the first few days of life and may be fatal without treatment. Signs include poor feeding, vomiting, jaundice and, sometimes, lethargy and/or bleeding. Neonatal E. coli sepsis can occur and is often FATAL. Treatment is withdrawal of milk and, if symptomatic, emergency measures

Disclaimer: These standards and guidelines are designed primarily as an educational resource for physicians to help them provide quality medical services. 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





Primary or Secondary Hypergalactosemia (Galactose Elevated)



Actions are shown in shaded boxes; results are in the unshaded boxes.

Abbreviations / Key:

RBC = Red blood cell
A-V = Arteriovenous

GALT = Galactose-1-phosphate uridylyltransferase

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