Galactosemia (GALT Deficiency)

Assess clinically
LFTs, Quant. RBC Gal-1-P, Urine reducing substance assay

- GALT < 1%
  - GALT genotype
    - Q188R/Q188R
    - Other
    - Severe/Severe
  - Classical galactosemia
    (potentially life-threatening)

- GALT = 1 – 10%
  - GALT genotype
    - S135L/S135L
    - Variant/Variant
    - Severe/Variant
  - Variant (Clinical)
    Galactosemia

- GALT >10 – 75%
  - GALT genotype
    - Q188R/Variant
    - Other
    - Variant/Variant
  - Variant (benign)
    Galactosemia or carrier

GALT Normal
- No further action required

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

Abbreviations/Key
GALT = Galactose-1-phosphate uridyl transferase
LFT: Liver Function Tests
RBC = Red blood cell
a = Abnormal 120 min. [13C] galactose breath test
b = Normal 120 min. [13C] galactose breath test and 10% GALT in liver
* = Transfusions can invalidate results of RBC enzyme assays
† = Benign variant type
‡ = Clinically significant variant type

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

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