Galactosemia (GALT Deficiency)

Absent or decreased GALT

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

Quantitative RBC GALT 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 (Clinical) Galactosemia

GALT Normal

No further action required

GALT genotype

Q188R/N314D
  Severe/N314D
  ‡ Variant/Variant
  †Variant/Variant
  Q188R/Normal
  Severe/Normal
  N314D/Normal
  †Variant/Normal

Variant (benign) Galactosemia or carrier

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
‡ = Clinically significant variant type
† = Benign variant type
* = Transfusions can invalidate results of RBC enzyme assays

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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