Galactosemia Testing Algorithm

- Clinical suspicion of classic galactosemia
- Positive newborn screen for galactosemia
- Investigation of possible carrier status

Order GCT / Galactosemia Reflex, Blood

Galactose-1-phosphate uridylyltransferase (GALT) enzyme analysis performed

GALT <24.5 nmol/h/mg of hemoglobin

- Possible causes:
  - Classic galactosemia
  - Duarte variant galactosemia
  - Carrier:
    - Classic galactosemia variant
    - Duarte variant

  GALT full gene analysis (sequencing and deletion/duplication) is automatically performed

  - 2 variants identified
  - 1 variant identified and concordant enzyme results
  - 0–1 variants identified and discordant enzyme results

  Diagnostic of disease
  Carrier status confirmed

  To resolve discrepant enzyme and variant results, consider GALTP / Galactose-1-Phosphate Uridylyltransferase Biochemical Phenotyping, Erythrocytes

GALT ≥24.5 nmol/h/mg of hemoglobin

- Classic galactosemia excluded

Was testing ordered to follow-up an abnormal newborn screen?

- YES
  - Was total galactose elevated on the newborn screen?
    - YES
      - Carrier status for classic galactosemia unlikely
      - STOP
    - NO
      - Order GAL1P / Galactose-1-Phosphate, Erythrocytes
      - Normal
      - Elevated
      - Rule-out galactokinase (GALK) deficiency; order GALK
      - Rule-out uridine diphosphate (UDP) galactose-4-epimerase (GALE) deficiency; order GALE
  - NO
    - Carrier status confirmed

- NO
  - Order GAL1P / Galactose-1-Phosphate, Erythrocytes
  - Normal
  - Elevated
  - Rule-out galactokinase (GALK) deficiency; order GALK
  - Rule-out uridine diphosphate (UDP) galactose-4-epimerase (GALE) deficiency; order GALE

If clinically indicated, rule-out galactosemia due to galactokinase (GALK) deficiency; order GALK / Galactokinase, Blood or uridine diphosphate (UDP) galactose-4-epimerase (GALE) deficiency; order GALE / Uridine Diphosphate-Galactose 4’ Epimerase, Blood

STOP