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 uridylytransferase (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 gene analysis (14-variant panel) is automatically performed*

- Concordant enzyme and variant results
  - Diagnostic of disease or carrier status
- Discordant enzyme and variant results
  - Consider GALTZ / GALT Gene, Full Gene Analysis, Varies to investigate for variants that are not identified by the 14-variant panel

Discordant enzyme and variant results

- 2 variants identified
  - Diagnostic of disease
- 1 variant identified and concordant enzyme results
  - Carrier status confirmed
- 0–1 variants identified and discordant enzyme results
  - To resolve discrepant enzyme and variant results, consider GALTP / Galactose-1-Phosphate Uridylytransferase 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
        - Rule-out galactokinase (GALK) deficiency; order GALK / Galactokinase, Blood
        - Rule-out uridine diphosphate (UDP) galactose-4′-epimerase (GALE) deficiency; order GALE / UDP-Galactose 4′ Epimerase (GALE), Blood
      - NO
        - Carrier status for classic galactosemia unlikely
          - NO
            - STOP
          - YES
            - Galactosemia Testing Algorithm

* The detection rate of this gene analysis is ~80%-90% for classic galactosemia variants.