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
- Discordant enzyme and variant results

Diagnostic of disease or carrier status

- 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 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
- NO

Was total galactose elevated on the newborn screen?

- YES
- NO

Carrier status for classic galactosemia unlikely

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 / UDP Galactose 4’ Epimerase, Blood

Order GAL1P / Galactose-1-Phosphate, Erythrocytes

Normal

Elevated

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, Blood

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