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 uridytransferase (GALT) enzyme analysis performed

GALT <24.5 nmol/h/mg of hemoglobin

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

Concordant enzyme and mutation results

Diagnosis of disease or carrier status

2 mutations identified

Diagnostic of disease

1 mutation identified and concordant enzyme results

Carrier status confirmed

0–1 mutations identified and discordant enzyme results

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

GALT ≥24.5 nmol/h/mg of hemoglobin

Classical galactosemia excluded

Was total galactose elevated on the newborn screen?

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

Carrier status for classic galactosemia unlikely

Normal

Elevated

Order GAL1P / Galactose-1-Phosphate (Gal-1-P), Erythrocytes

Rule-out galactokinase (GALK) deficiency by ordering GALK / Galactokinase, Blood

Rule-out Uridine Diphosphate (UDP) galactose-4-epimerase (GALE) deficiency

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