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 mutation
  - Duarte mutation

GALT gene analysis (14-mutation panel) is automatically performed*

Concordant enzyme and mutation results

Diagnosis of disease or carrier status

Consider GALTZ / GALT Gene, Full Gene Analysis to investigate for mutations that are not identified by the 14-mutation panel

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 Uridylyltransferase Biochemical Phenotyping, Erythrocytes

Classic galactosemia excluded

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

YES

Classic galactosemia unlikely

NO

Carrier status for classic galactosemia unlikely

Was total galactose elevated on the newborn screen?

YES

Normal

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

Elevated

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.