

TEST CHANGE

Galactosemia (GALT) Enzyme Activity and 9 Mutations

0051175, GALTPAN

Specimen Requirements:

Patient Preparation:

Collect: Lavender (EDTA) or, pink (K2EDTA), ~~or green (sodium heparin).~~

Specimen Preparation: ~~Do not freeze.~~ Transport 2-10 mL whole blood. (Min: 1-3 mL.)

Transport Temperature: Refrigerated.

Unacceptable Conditions: Frozen or room temperature specimens.

Remarks:

Stability: Room temperature: Unacceptable; Refrigerated: 5 days; Frozen: Unacceptable

Methodology: Enzymatic Assay/Polymerase Chain Reaction (PCR)/Single Nucleotide Extensions

Performed: Varies

Reported: 7-10 days

Note:

CPT Codes: 82775; 81401

New York DOH Approval Status: This test is New York DOH approved.

Interpretive Data:

One U/g Hb is equivalent to one umol/hour/gram of hemoglobin (umol/hr/g Hb).

Refer to report.

Reference Interval:

By report