

TEST CHANGE

Galactosemia (GALT) Enzyme Activity and 9 Mutations

0051175, GALTPAN

Specimen Requirements:

Patient Preparation:

Collect: Lavender (EDTA) or pink (K2EDTA)

Specimen Preparation: Transport 2 mL whole blood (Min: 1 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

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

HOTLINE NOTE: There is a prompt change associated with this test. Refer to the Hotline Test Mix for interface build information.