

TEST CHANGE

Galactosemia (*GALT*) Sequencing and Deletion/Duplication

3004716, *GALT* NGS

Specimen Requirements:

Patient Preparation:

Collect: Lavender or pink (EDTA) or yellow (ACD solution A or B).
New York State Clients: Lavender (EDTA)

Specimen Preparation: Transport **23** mL whole blood. (Min: **12** mL)
New York State Clients: Transport 3 mL whole blood. (Min. 3 mL)

Transport Temperature: Refrigerated.

Unacceptable Conditions: Serum or plasma; grossly hemolyzed or frozen specimens; saliva, buccal brush, or swab; FFPE tissue.

Remarks:

Stability: Ambient: 72 hours; Refrigerated: 1 week; Frozen: Unacceptable
New York State Clients: Ambient: 48 hours; Refrigerated: 2 weeks; Frozen: Unacceptable

Methodology: Massively Parallel Sequencing

Note:

Gene Tested: *GALT* (NM_000155)

When testing cord blood specimens, the presence of maternal cell contamination (MCC) is possible, which may impact result interpretation. If clinically warranted, testing for MCC is available, at a charge, through ARUP Laboratories.

CPT Codes: 81406; 81479

New York DOH Approval Status: Specimens from New York clients will be sent out to a New York DOH approved laboratory, if possible.

Interpretive Data:

Refer to report.

Reference Interval:

Refer to report