

HOTLINE: Effective November 18, 2019

New Test **3001790** **Galactose-1-Phosphate Uridyltransferase (GALT Enzyme), RBC** **GALT ENZ**



Patient History For Galactosemia



Additional Technical Information

Methodology: Enzymatic/Liquid Chromatography-Tandem Mass Spectrometry
Performed: Mon, Wed, Fri
Reported: 2-5 days

Specimen Required: Collect: Green (Sodium or Lithium Heparin), Lavender (K₂EDTA), or Pink (K₂EDTA). Collect on ice.
Specimen Preparation: **DO NOT FREEZE.** Transport 7 mL whole blood. (Min: 3 mL)
Storage/Transport Temperature: Refrigerated.
Unacceptable Conditions: Hemolyzed specimens.
Stability (collection to initiation of testing): Ambient: Unacceptable; Refrigerated: 5 days; Frozen: Unacceptable

Reference Interval: Greater than or equal to 19.4 U/g Hb

Interpretive Data:

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

Genotype	Galactose-1-Phosphate Uridyltransferase activity (μmol h ⁻¹ gHb ⁻¹)
Classic galactosemia (G/G)	Less than or equal to 0.7
Duarte galactosemia (D/G)	3.1-7.8
Classic galactosemia carrier (G/N)	6.5-16.2
Duarte homozygous (D/D)	6.4-16.5
Duarte carrier (D/N)	12.0-24.0
Normal (N/N)	Greater than or equal to 19.4

See Compliance Statement B: www.aruplab.com/CS

Note: Values for enzyme activity only may not differentiate between variant form of galactosemia or carriers. For a more accurate evaluation of patients suspected to have galactosemia, the preferred test is Galactosemia (GALT) Enzyme Activity and 9 Mutations (ARUP test code 0051175). To monitor therapy in patients with galactosemia, order Galactose-1-Phosphate in Red Blood Cells (ARUP test code 0081296).

CPT Code(s): 82775

New York DOH Approved.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.