
Lab Dept: Chemistry

Test Name: GALACTOSE-1-PHOSPHATE
URIDYLTRANSFERASE

General Information

Lab Order Codes: G1PH

Synonyms: GPUT; GALT

CPT Codes: 82775 – Galactose-1-phosphate uridyl transferase; quantitative

Test Includes: Galactose-1-phosphate uridyl transferase level reported as nmol/h/mg of hemoglobin.

Logistics

Test Indications: Diagnosis of galactose-1-phosphate uridyltransferase deficiency, the most common cause of galactosemia. Confirmation of abnormal state newborn screening results.

Lab Testing Sections: Chemistry - Sendouts

Referred to: Mayo Medical Laboratories (Mayo Test: GALT)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: 4-5 days; test set up Monday, Wednesday, Friday

Special Instructions: Biochemical Genetics Request Form (Supply T439) is required.

Specimen

Specimen Type: Whole blood

Container: Lavender top (EDTA) tube

Draw Volume: 5 mL (Minimum: 2 mL) blood

Processed Volume: Same as Draw Volume

Collection:	Routine venipuncture
Special Processing:	Lab Staff: Do Not centrifuge. Specimen should remain in original collection tube. Store and ship at refrigerated temperatures. Include Patient History for Galactosemia Form. Specimen must not be frozen. Forward promptly.
Patient Preparation:	None
Sample Rejection:	Specimens other than whole blood, anticoagulants other than EDTA; Heparin or ACD, gross hemolysis; frozen specimens; mislabeled or unlabeled specimens

Interpretive

Reference Range:	≥24.5 nmol/h/mg Hb An interpretive report will be provided.
Critical Values:	N/A
Limitations:	This assay is not useful for monitoring of dietary compliance by galactosemics. This assay will not detect galactokinase deficiency or uridine diphosphate galactose 4-epimerase deficiency.
Methodology:	Enzyme reaction followed by Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)
References:	Mayo Medical Laboratories (December 2017)
Updates:	11/21/2006: MML has removed the upper normal range value for this test. The reference range previously was stated as 18.5-28.5 U/g of hemoglobin. 2/14/2014: Test down at MML, temporary move to ARUP. 4/28/2014: Test back up at MML with new method, previously listed as enzymatic. Please note new reference range.