

Reporting Title: Gal-1-P Uridyltransferase, RBC
Performing Location: Rochester

Ordering Guidance:

This assay is not appropriate for monitoring dietary compliance. If dietary monitoring is needed, order GAL1P / Galactose-1-Phosphate, Erythrocytes.

This test is for galactose-1-phosphate uridyltransferase (GALT) enzyme testing only. The preferred test to evaluate for possible diagnosis of galactosemia, routine carrier screening, and follow-up of abnormal newborn screening results is GCT / Galactosemia Reflex, Blood.

This assay will not detect galactokinase (GALK) deficiency or uridine diphosphate-galactose 4' epimerase (GALE) deficiency.

- To evaluate for GALK deficiency, order GALK / Galactokinase, Blood.
- To evaluate for GALE deficiency, order GALE / Uridine Diphosphate -Galactose 4' Epimerase, Blood.
- To evaluate for GALT deficiency, order GALP / Galactose, Plasma and molecular analysis of the *GALT* gene.

Necessary Information:
Patient's age is required.

[Biochemical Genetics Patient Information](#) (T602) is recommended, but not required, to be filled out and sent with the specimen to aid in the interpretation of test results.

Specimen Requirements:

Multiple whole blood tests for galactosemia can be performed on 1 specimen. Prioritize order of testing when submitting specimens. For a list of tests that can be ordered together see [Galactosemia-Related Test List](#).

Container/Tube:
Preferred: Lavender top (EDTA)
Acceptable: Green top (sodium heparin) or yellow top (ACD)
Specimen Volume: 5 mL

Forms:

- 1. New York Clients-Informed consent is required.** Document on the request form or electronic order that a copy is on file. The following documents are available:
 - [Informed Consent for Genetic Testing](#) (T576)
 - [Informed Consent for Genetic Testing-Spanish](#) (T826)
- 2. [Biochemical Genetics Patient Information](#) (T602)** is recommended.
- 3. If not ordering electronically, complete, print, and send a [Biochemical Genetics Test Request](#) (T798) with the specimen.**

Specimen Type	Temperature	Time	Special Container
Whole Blood EDTA	Ambient	14 days	
	Refrigerated (preferred)	28 days	

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
8333	Gal-1-P Uridyltransferase, RBC	Numeric	nmol/h/mg Hb	24082-0
2296	Interpretation (GALT)	Alphanumeric		59462-2
58115	Reviewed By	Alphanumeric		18771-6

LOINC® and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

82775

Reference Values:

> or =24.5 nmol/h/mg of hemoglobin