

# **Test Definition: GALT**

Galactose-1-Phosphate Uridyltransferase,
Blood

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.

## **Necessary Information:**

Patient's age is required.

<u>Biochemical Genetics Patient Information</u> (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 <u>Galactosemia-Related Test List</u>.

#### **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 <u>Biochemical Genetics Test Request</u> (T798) with the specimen.

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



# **Test Definition: GALT**

Galactose-1-Phosphate Uridyltransferase, Blood

#### **Result Codes:**

Result ID	Reporting Name	Туре	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