

### **Test Definition: GALTP**

Galactose-1-Phosphate Uridyltransferase Biochemical Phenotyping, Erythrocytes

# **Reporting Title:** Gal-1-Phos Urdyltrns Phenotype,RBC **Performing Location:** Rochester

#### Ordering Guidance:

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.

For monitoring of dietary compliance, order GAL1P / Galactose-1-Phosphate, Erythrocytes.

#### Necessary Information:

Patient's age is required.

A quantitative galactose-1-phosphate uridyltransferase level (GALT / Galactose-1-Phosphate Uridyltransferase, Blood) is required for accurate interpretation.

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: Lavender top (EDTA) Specimen Volume: 3 mL

#### **Specimen Minimum Volume:**

2 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
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Whole Blood EDTA	Refrigerated (preferred)	28 days	
	Ambient	14 days	

#### **Result Codes:**

Result ID	Reporting Name	Туре	Unit	LOINC®
80341	Gal-1-Phos Urdyltrns Phenotype,RBC	Alphanumeric		33780-8
34524	Reviewed By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

### Supplemental Report:

No

#### **CPT Code Information:**

82664 82775

#### **Reflex Tests:**

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
GALT	Gal-1-P Uridyltransferase, RBC			Yes	Yes

#### **Result Codes for Reflex Tests:**

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



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#### **Reference Values:**

An interpretative report will be provided.