

Galactose-1-Phosphate, Erythrocytes

## **Reporting Title:** Galactose-1-Phosphate, RBC **Performing Location:** Rochester

### Ordering Guidance:

This test is used to monitor dietary therapy of patients with galactosemia due to deficiency of galactose-1-phosphate uridyltransferase or uridine diphosphate galactose-4-epimerase.

This test is not appropriate for the diagnosis of galactosemia. 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 test is not appropriate for the diagnosis of epimerase deficiency, the preferred test to evaluate this deficiency is GALE / Uridine Diphosphate-Galactose 4' Epimerase, Blood.

If GAL1P / Galactose-1-Phosphate, Erythrocytes testing is needed, the test can be added to existing specimens if they were received in the testing laboratory within 72 hours of collection.

### **Necessary Information:**

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.

Patient Preparation: Specimens collected following a meal can exhibit postprandial elevations. For infants, collect a specimen immediately prior to feeding to avoid this. Container/Tube: Preferred: Lavender top (EDTA) Acceptable: Green top (sodium heparin) Specimen Volume: 3 mL

### **Specimen Minimum Volume:**

2 mL

### Forms:

Biochemical Genetics Patient Information (T602) is recommended.
If not ordering electronically, complete, print, and send a Biochemical Genetics Test Request (T798) with the specimen.

# LABORATORIES

### **Test Definition: GAL1P**

Galactose-1-Phosphate, Erythrocytes

Specimen Type	Temperature	Time	Special Container	
Whole Blood EDTA	Refrigerated	72 hours		

### **Result Codes:**

Result ID	Reporting Name	Туре	Unit	LOINC®
24101	Galactose-1-Phosphate, RBC	Numeric	mg/dL	2312-7

LOINC and CPT codes are provided by the performing laboratory.

### **Supplemental Report:**

No

### **CPT Code Information:**

84378

### **Reference Values:**

Reference interval (normal range): < or =0.9 mg/dL Therapeutic range: < or =4.9 mg/dL