

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 uridylyltransferase 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

Forms:

1. [Biochemical Genetics Patient Information](#) (T602) is recommended.
2. 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	Refrigerated	72 hours	

Result Codes:

Result ID	Reporting Name	Type	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