

**Reporting Title:** Galactokinase, B**Performing Location:** Rochester**Ordering Guidance:**

This test is for diagnosis of galactokinase (GALK) deficiency and does **not** detect either galactose-1-phosphate uridylyltransferase (GALT) deficiency, the most common cause of galactosemia, or uridine diphosphate-galactose 4' epimerase (GALE) deficiency. In most cases, GALT deficiency should be ruled out prior to evaluating for GALK deficiency.

- 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.
- To evaluate GALT deficiency only, order GALT / Galactose-1-Phosphate Uridyltransferase, Blood
- To evaluate for GALE deficiency only, order GALE / UDP-Galactose 4' Epimerase (GALE), Blood

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

**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](#).

**Container/Tube:**

**Preferred:** Lavender top (EDTA)  
**Acceptable:** Green top (sodium or lithium heparin) or yellow top (ACD)  
**Specimen Volume:** 4 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 | Refrigerated (preferred) | 10 days  |                   |
|                  | Ambient                  | 72 hours |                   |

**Result Codes:**

| Result ID | Reporting Name   | Type    | Unit         | LOINC®  |
|-----------|------------------|---------|--------------|---------|
| 38005     | Galactokinase, B | Numeric | nmol/h/mg Hb | 81143-0 |

|       |                       |              |  |         |
|-------|-----------------------|--------------|--|---------|
| 38007 | Interpretation (GALK) | Alphanumeric |  | 59462-2 |
| 38006 | Reviewed By           | Alphanumeric |  | 18771-6 |

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

Supplemental Report:

No

CPT Code Information:

82759

Reference Values:

> or =0.7 nmol/h/mg of hemoglobin