

**Reporting Title:** UDP-galactose 4' epimerase, RBC  
**Performing Location:** Rochester

**Ordering Guidance:**

This test is appropriate for diagnosis of uridine diphosphate-galactose 4' epimerase (GALE) deficiency, but it will **not** detect galactokinase (GALK) deficiency or galactose-1-phosphate uridytransferase (GALT) deficiency or galactose mutarotase (GALM) deficiency.

- To evaluate for GALK deficiency, order GALK / Galactokinase, Blood.
- To evaluate for GALT deficiency, order GALT / Galactose-1-Phosphate Uridyltransferase, Blood.
- To evaluate for GALM deficiency, order GALP / Galactose, Plasma and molecular analysis of the *GALM* gene.

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

**Necessary Information:**  
**Patient's age is required.**

[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:** 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 [Biochemical Genetics Test Request](#) (T798) with the specimen.

Specimen Type	Temperature	Time	Special Container
Whole Blood EDTA	Refrigerated (preferred)	14 days	
	Ambient	6 days	

**Result Codes:**

Result ID	Reporting Name	Type	Unit	LOINC®
64372	UDP-galactose 4' epimerase, RBC	Numeric	nmol/h/mg Hb	79469-3
37979	Interpretation (GALE)	Alphanumeric		59462-2
37978	Reviewed By	Alphanumeric		18771-6

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

Supplemental Report:

No

CPT Code Information:

82542

Reference Values:

> or =3.5 nmol/h/mg of hemoglobin