
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 uridylyltransferase (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

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
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