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**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

**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)	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