

**Reporting Title:** Galactosemia Mutation Panel  
**Performing Location:** Rochester

**Ordering Guidance:**  
The recommended as a first-tier test is galactose-1-phosphate uridylyltransferase enzyme analysis; order GALT / Galactose-1-Phosphate Uridyltransferase, Blood.

This genetic variant panel is recommended for individuals with a GALT enzyme value less than 24.5 nmol/h/mg of hemoglobin.

**Shipping Instructions:**  
Specimen preferred to arrive within 96 hours of collection.

**Specimen Requirements:**  
Multiple whole blood tests for galactosemia can be performed on one specimen. Prioritize order of testing when submitting specimens. See [Galactosemia-Related Test List](#) for a list of tests that can be ordered together.

**Patient Preparation:** A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

**Specimen Type:** Whole blood  
**Container/Tube:**  
**Preferred:** Lavender top (EDTA) or yellow top (ACD)  
**Acceptable:** Any anticoagulant  
**Specimen Volume:** 3 mL  
**Collection Instructions:**  
1. Invert several times to mix blood.  
2. Send whole blood specimen in original tube. **Do not aliquot.**

**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. [Molecular Genetics: Biochemical Disorders Patient Information](#) (T527)

Specimen Type	Temperature	Time	Special Container
Varies	Ambient (preferred)		
	Frozen		
	Refrigerated		

**Result Codes:**

Result ID	Reporting Name	Type	Unit	LOINC®
-----------	----------------	------	------	--------

606344	Result Summary	Alphanumeric		50397-9
606345	Result	Alphanumeric		82939-0
606346	Interpretation	Alphanumeric		69047-9
606347	Additional Information	Alphanumeric		48767-8
606348	Method	Alphanumeric		85069-3
606349	Specimen	Alphanumeric		31208-2
606350	Source	Alphanumeric		31208-2
606351	Released By	Alphanumeric		18771-6

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

Supplemental Report:

No

CPT Code Information:

81401

Reference Values:

An interpretive report will be provided.