
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.

Specimen Minimum Volume:

1 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. 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.