
Reporting Title: Galactosemia Gene Analysis**Performing Location: Rochester****Specimen Requirements:**

Forms:

1. Molecular Genetics: Congenital Inherited Diseases Patient Information (T521) in Special Instructions
2. New York Clients-Informed consent is required. Please document on the request form or electronic order that a copy is on file. An Informed Consent for Genetic Testing (T576) is available in Special Instructions.

Specimen preferred to arrive within 96 hours of draw.

Submit only 1 of the following specimens:

Preferred:

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 specimen in original tube.

Specimen Stability Information: Ambient (preferred)/Frozen/Refrigerated

Acceptable:

Specimen Type: Blood spot

Container/Tube: Whatman Protein Saver 903 Paper

Specimen Volume: 5 blood spots

Collection Instructions:

1. Let blood dry on the filter paper at ambient temperature in a horizontal position for 3 hours.

2. Do not expose specimen to heat or direct sunlight.

3. Do not stack wet specimens.

4. Keep specimen dry.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Specimen Type	Temperature	Time
Varies	Ambient (preferred)	
	Frozen	
	Refrigerated	

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
52878	Result Summary	Alphanumeric		50397-9
52879	Result	Alphanumeric		In Process
52880	Interpretation	Alphanumeric		69047-9
52881	Specimen	Alphanumeric		31208-2
52882	Source	Alphanumeric		31208-2
52883	Method	Alphanumeric		49549-9
52884	Released By	Alphanumeric		No LOINC Needed

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:
Enhanced**CPT Code:** 1 × 81401**Reference Values:**

An interpretive report will be provided.