

Reporting Title: Galactosemia Reflex, B**Performing Location:** Rochester**Ordering Guidance:**

This test is appropriate for the diagnosis of, and routine carrier screening for, galactose-1-phosphate uridylyltransferase deficiency.

This assay is not appropriate for monitoring dietary compliance. For dietary monitoring, order GAL1P / Galactose-1-Phosphate, Erythrocytes.

Necessary Information:

Patient's age is required.

Specimen Requirements:

Multiple whole blood tests for galactosemia can be performed on one 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 heparin) or yellow top (ACD)

Specimen Volume: 5 mL

Collection Instructions:

1. Invert several times to mix blood.
2. Send whole blood specimen in original tube. Do not aliquot.

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. If not ordering electronically, complete, print, and send an Biochemical Genetics Test Request (T798) with the specimen.

| Specimen Type | Temperature | Time | Special Container |
|------------------|--------------------------|---------|-------------------|
| Whole Blood EDTA | Refrigerated (preferred) | 28 days | |

| | | | |
|--|---------|---------|--|
| | Ambient | 14 days | |
|--|---------|---------|--|

Result Codes:

| Result ID | Reporting Name | Type | Unit | LOINC® |
|-----------|---|--------------|--------------|---------|
| 8333 | Gal-1-P Uridyltransferase, RBC Also used by tests: GALT | Numeric | nmol/h/mg Hb | 24082-0 |
| 2296 | Interpretation (GALT) Also used by tests: GALT | Alphanumeric | | 59462-2 |
| 58115 | Reviewed By Also used by tests: GALT | Alphanumeric | | 18771-6 |

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

82775

81406 (if appropriate)

Reflex Tests:

| Test ID | Reporting Name | CPT Units | CPT Code | Always Performed | Orderable Separately |
|---------|-------------------------------------|-----------|----------|------------------|----------------------|
| GALZ | Galactosemia, Full Gene Analysis | | | No | Yes |
| CULFB | Fibroblast Culture for Genetic Test | | | No | Yes |

Result Codes for Reflex Tests:

| Test ID | Result ID | Reporting Name | Type | Unit | LOINC® |
|---------|-----------|------------------|--------------|------|---------|
| GALZ | 608596 | Test Description | Alphanumeric | | 62364-5 |
| GALZ | 608597 | Specimen | Alphanumeric | | 31208-2 |

| Test ID | Result ID | Reporting Name | Type | Unit | LOINC® |
|---------|-----------|------------------------|--------------|------|---------|
| GALZ | 608598 | Source | Alphanumeric | | 31208-2 |
| GALZ | 608599 | Result Summary | Alphanumeric | | 50397-9 |
| GALZ | 608600 | Result | Alphanumeric | | 82939-0 |
| GALZ | 608601 | Interpretation | Alphanumeric | | 69047-9 |
| GALZ | 608602 | Resources | Alphanumeric | | 99622-3 |
| GALZ | 608603 | Additional Information | Alphanumeric | | 48767-8 |
| GALZ | 608604 | Method | Alphanumeric | | 85069-3 |
| GALZ | 608605 | Genes Analyzed | Alphanumeric | | 48018-6 |
| GALZ | 608606 | Disclaimer | Alphanumeric | | 62364-5 |
| GALZ | 608607 | Released By | Alphanumeric | | 18771-6 |
| CULFB | 52327 | Result Summary | Alphanumeric | | 50397-9 |
| CULFB | 52329 | Interpretation | Alphanumeric | | 69965-2 |
| CULFB | 52328 | Result | Alphanumeric | | 82939-0 |
| CULFB | CG770 | Reason for Referral | Alphanumeric | | 42349-1 |
| CULFB | CG899 | Specimen | Alphanumeric | | 31208-2 |
| CULFB | 52331 | Source | Alphanumeric | | 31208-2 |
| CULFB | 52332 | Method | Alphanumeric | | 85069-3 |
| CULFB | 54625 | Additional Information | Alphanumeric | | 48767-8 |
| CULFB | 52333 | Released By | Alphanumeric | | 18771-6 |

Reference Values:

> or =24.5 nmol/h/mg of hemoglobin