Overview

Useful For
Screening test for galactosemia using urine specimens

Genetics Test Information
Galactose-1-phosphate uridyltransferase (GALT) deficiency is the most common cause of galactosemia and requires lifelong restriction of dietary galactose.

Urine galactose can be elevated in patients with galactosemia caused by either galactose-1-phosphate uridyltransferase (GALT) deficiency or galactokinase (GALK) deficiency.

Classic galactosemia can be diagnosed by analysis of GALT enzyme.

Testing Algorithm
See Galactosemia Testing Algorithm in Special Instructions.

Special Instructions
- Galactosemia Testing Algorithm
- Biochemical Genetics Patient Information

Method Name
Spectrophotometric, Kinetic

NY State Available
Yes

Specimen

Specimen Type
Urine

Advisory Information
This test is not recommended for follow-up of positive newborn screening results, or for diagnosis of galactosemia. The preferred test to evaluate for possible diagnosis of galactosemia, routine carrier screening, and followup of abnormal newborn screening results is GCT / Galactosemia Reflex, Blood along with GAL1P / Galactose-1-Phosphate, Erythrocytes.

This test is not appropriate for monitoring of galactosemia. For monitoring, 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 Required
Supplies: Aliquot Tube, 5 mL (T465)

Collection Container/Tube: Clean, plastic urine collection container
Test Definition: GALU
Galactose, QN, U

Submission Container/Tube: Plastic, 5-mL tube

Specimen Volume: 1 mL

Collection Instructions: Collect a random urine specimen.

Forms
1. Biochemical Genetics Patient Information (T602) is recommendation, see Special Instructions.
2. If not ordering electronically, complete, print, and send an Inborn Errors of Metabolism Test Request (T798) with the specimen.

Specimen Minimum Volume
0.5 mL

Reject Due To
All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

<table>
<thead>
<tr>
<th>Specimen Type</th>
<th>Temperature</th>
<th>Time</th>
<th>Special Container</th>
</tr>
</thead>
<tbody>
<tr>
<td>Urine</td>
<td>Frozen (preferred)</td>
<td>365 days</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Ambient</td>
<td>20 days</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Refrigerated</td>
<td>20 days</td>
<td></td>
</tr>
</tbody>
</table>

Clinical and Interpretive

Clinical Information

Galactosemia is an autosomal recessive disorder that results from a deficiency of any 1 of the 3 enzymes catalyzing the conversion of galactose to glucose: galactose-1-phosphate uridyltransferase (GALT), galactokinase (GALK), and uridine diphosphate galactose-4-epimerase (GALE). GALT deficiency is the most common cause of galactosemia and is often referred to as classic galactosemia. The complete or near-complete deficiency of GALT enzyme is life-threatening if left untreated. Complications in the neonatal period include failure to thrive, liver failure, sepsis, and death.

Galactosemia is treated by a galactose-restricted diet, which allows for rapid recovery from the acute symptoms and a generally good prognosis. Despite adequate treatment from an early age, individuals with galactosemia remain at increased risk for developmental delays, speech problems, and abnormalities of motor function. Females with galactosemia are at increased risk for premature ovarian failure. Based upon reports by newborn screening programs, the frequency of classic galactosemia in the United States is approximately 1 in 30,000, although literature reports range from 1 in 10,000 to 1 in 60,000 live births.

A comparison of plasma and urine galactose and blood galactose-1-phosphate (Gal-1-P) levels may be useful in distinguishing among the 3 forms of galactosemia; however, these are only general patterns and further confirmatory testing would be required to make a diagnosis.
Test Definition: GALU
Galactose, QN, U

<table>
<thead>
<tr>
<th>Deficiency</th>
<th>Galactose (Plasma/Urine)</th>
<th>Gal-1-P (Blood)</th>
</tr>
</thead>
<tbody>
<tr>
<td>GALK</td>
<td>Elevated</td>
<td>Normal</td>
</tr>
<tr>
<td>GALT</td>
<td>Elevated</td>
<td>Elevated</td>
</tr>
<tr>
<td>GALE</td>
<td>Normal-Elevated</td>
<td>Elevated</td>
</tr>
</tbody>
</table>

See Galactosemia Testing Algorithm in Special Instructions for additional information.

Reference Values
<30 mg/dL

Interpretation
Additional testing is required to investigate the cause of abnormal results.

In patients with galactosemia, elevated galactose in plasma or urine may suggest ineffective dietary restriction or compliance; however, the concentration of galactose-1-phosphate in erythrocytes (GAL1P / Galactose-1-Phosphate, Erythrocytes) is the most sensitive index of dietary control.

Increased concentrations of galactose may also be suggestive of severe hepatitis, biliary atresia of the newborn and, in rare cases, galactose intolerance.

If galactosemia is suspected, additional testing to identify the specific enzymatic defect is required. See Galactosemia Testing Algorithm in Special Instructions for follow-up of abnormal newborn screening results, comprehensive diagnostic testing, and carrier testing. Results should be correlated with clinical presentation and confirmed by specific enzyme or molecular analysis.

Cautions
No significant cautionary statements

Clinical Reference


Performance

Method Description
Test Definition: GALU
Galactose, QN, U


PDF Report
No

Day(s) and Time(s) Test Performed
Tuesday; a.m.

Analytic Time
8 days

Maximum Laboratory Time
15 days

Specimen Retention Time
1 month

Performing Laboratory Location
Rochester

Fees and Codes

Fees
- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their Regional Manager. For assistance, contact Customer Service.

Test Classification
This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

CPT Code Information
82760

LOINC® Information

<table>
<thead>
<tr>
<th>Test ID</th>
<th>Test Order Name</th>
<th>Order LOINC Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>GALU</td>
<td>Galactose, QN, U</td>
<td>2310-1</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Result ID</th>
<th>Test Result Name</th>
<th>Result LOINC Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>8765</td>
<td>Galactose, QN, U</td>
<td>2310-1</td>
</tr>
</tbody>
</table>