

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

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

Specimen Type	Temperature	Time	Special Container
Urine	Frozen (preferred)	365 days	
	Ambient	20 days	
	Refrigerated	20 days	

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 uridylyltransferase (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.

	Galactose (Plasma/Urine)	Gal-1-P (Blood)
Deficiency		
GALK	Elevated	Normal
GALT	Elevated	Elevated
GALE	Normal-Elevated	Elevated

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

- Berry GT: Classic Galactosemia and Clinical Variant Galactosemia. In GeneReviews. Edited by MP Adam, HH Ardinger, RA Pagon, et al. Updated 2017 March 9. Accessed February 20, 2020. Available at www.ncbi.nlm.nih.gov/books/NBK1518/
- Walter JH, Fridovich -Keil JL: Galactosemia. In The Online Metabolic Bases of Inherited Disease. Edited by D Valle, AL Beaudet, B Vogelstein, et al. McGraw-Hill. Accessed June 18, 2019. Available at <http://ommbid.mhmedical.com/content.aspx?bookid=971§ionid=62672411>

Performance

Method Description

The formation of nicotinamide adenine dinucleotide (NADH; reduced form) measured by the increase in absorbance at 340 nm is proportional to the amount of D-galactose in the sample. (Kurz G, Wallenfels K: Methods of Enzymatic Analysis. Vol. 3. Second edition. Edited by HV Bergmeyer, V Chemie, Weinheim. Academic Press, 1974 pp 1279-1282); Cowan T, Pasquali M: Laboratory Investigations of Inborn Errors of Metabolism. In Pediatric

Endocrinology and Inborn Errors of Metabolism. Second Edition. Edited by K Sarafoglou, GF Hoffman, KS Roth. 2017 pp 1139-1158)

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

Test ID	Test Order Name	Order LOINC Value
GALU	Galactose, QN, U	2310-1

Result ID	Test Result Name	Result LOINC Value
8765	Galactose, QN, U	2310-1