MAYO CLINIC LABORATORIES

Test Definition: GALU

Galactose, Quantitative, Random, Urine

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 GALT deficiency or galactokinase deficiency.

Classic galactosemia can be diagnosed by analysis of GALT enzyme.

Testing Algorithm
For information, see Galactosemia Testing Algorithm.

Special Instructions
- Galactosemia Testing Algorithm
- Biochemical Genetics Patient Information

Method Name
Spectrophotometric/Kinetic

NY State Available
Yes

Specimen

Specimen Type
Urine

Ordering Guidance
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 follow-up 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: Sarstedt Aliquot Tube, 5 mL (T914)
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

Biochemical Genetics Patient Information (T602) is recommended.

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	Ambient	20 days	
	Refrigerated	20 days	
	Frozen (preferred)	365 days	

Clinical & Interpretive

Clinical Information

Galactosemia is an autosomal recessive disorder that results from a deficiency of any 1 of the 4 enzymes catalyzing the conversion of galactose to glucose: galactose-1-phosphate uridyltransferase (GALT), galactokinase, uridine diphosphate galactose-4-epimerase, and galactose mutarotase. 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 motor function abnormalities. Female patients 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 (Gal1P) levels may be useful in distinguishing among the 4 forms of galactosemia; however, these are only general patterns and further confirmatory testing would be required to make a diagnosis.

Deficiency	Galactose (plasma/urine)	Gal1P (blood)
GALK	Elevated	Normal
GALT	Elevated	Elevated
GALE	Normal-Elevated	Elevated
GALM	Elevated in plasma	Normal-Elevated

For more information, see Galactosemia Testing Algorithm.

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 for patients with galactose-1-phosphate uridyltransferase and uridine diphosphate galactose-4-epimerase deficiencies.

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 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. For more information see Ordering Guidance.

Cautions

No significant cautionary statements

Clinical Reference

1. Berry GT: Classic galactosemia and clinical variant galactosemia. In: Adam MP, Everman DB, Mirzaa GM, et al. eds. GeneReviews [Internet]. University of Washington, Seattle; 2000. Updated March 11, 2021. Accessed September 10, 2024. Available at www.ncbi.nlm.nih.gov/books/NBK1518/

2. Walter JH, Fridovich-Keil JL: Galactosemia. In: Valle D, Antonarakis S, Ballabio A, Beaudet AL, Mitchell GA, eds. The Online Metabolic and Molecular Bases of Inherited Disease. McGraw-Hill; 2019. Accessed September 10, 2024. Available at https://ommbid.mhmedical.com/content.aspx?bookid=2709&sectionid=%20225081023

3. Yazici H, Canda E, Altinok YA, Ucar SK, Coker M. Two siblings with galactose mutarotase deficiency: Clinical differences. JIMD Rep. 2021;63(1):25-28. doi:10.1002/jmd2.12263

Performance

Method Description

The formation of reduced nicotinamide adenine dinucleotide (NADH) measured by the increase in absorbance at 340 nm is proportional to the amount of D-galactose in the sample.(Kurz G, Wallenfels K: In: Bergmeyer HV, ed: Methods of Enzymatic Analysis. Vol. 3. 2nd ed. Verlag Chemie, Weinheim, Academic Press; 1974:1279-1282; Cowan T, Pasquali M: Laboratory investigations of inborn errors of metabolism. In: Sarafoglou K, Hoffman GF, Roth KS, eds. Pediatric Endocrinology and Inborn Errors of Metabolism. 2nd ed. McGraw-Hill; 2017:1139-1158)

PDF Report
No

Day(s) Performed
Tuesday

Report Available
4 to 10 days

Specimen Retention Time
1 month

Performing Laboratory Location
Rochester

Fees & 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 account representative. 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. It has not been cleared or approved by the US 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