Test Definition: GATOL

Galactitol, Quantitative, Urine

Overview

Useful For

Monitoring effectiveness of treatment in patients with galactosemia

Establishing a baseline level prior to initiating treatment for galactosemia

Genetics Test Information

This test may be used as an aid in the diagnosis of galactosemia.

Urinary galactitol is often not affected by acute dietary ingestion of galactose; therefore, it is not a substitute for galactose-1-phosphate in monitoring diet.

Method Name

Gas Chromatography Mass Spectrometry (GC-MS)

NY State Available

Yes

Specimen

Specimen Type

Urine

Ordering Guidance

To monitor dietary ingestion of galactose, order GAL1P / Galactose-1-Phosphate, Erythrocytes.

Necessary Information

Patient's age is required.

Specimen Required

Supplies: Urine Tubes, 10 mL (T068)
Container/Tube: Plastic, 10-mL urine tube
Specimen Volume: 2 mL
Collection Instructions:
1. Collect a random urine specimen.
2. No preservative.

Forms

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

Specimen Minimum Volume

1 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	Refrigerated (preferred)	28 days	
	Frozen	28 days	

Clinical & Interpretive

Clinical Information

Galactosemia is an autosomal recessive disorder that results from a deficiency of 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 the GALT enzyme is life threatening. If left untreated, complications include liver failure, sepsis, cognitive and intellectual disabilities, and death.

Galactosemia is treated with a galactose-free diet, which allows for rapid recovery from the acute symptoms and a generally good prognosis. Despite adequate treatment from an early age, children with galactosemia remain at increased risk for developmental delays, speech problems, abnormalities of motor function, and female patients 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.

Galactose levels may be continuously elevated in individuals affected with galactosemia even with a galactose-restricted diet regimen due to an endogenous production of galactose. The reduction of galactose to galactitol is an alternate pathway of galactose disposition when galactose metabolism is impaired. The excretion of abnormal quantities of galactitol in the urine of patients is characteristic of this disorder, and patients may have abnormal levels of galactitol even with dietary compliance. Daily consumption of galactose may cause urine levels to rise thus providing information on effectiveness of or compliance with treatment, but unlike erythrocyte galactose-1-phosphate and plasma galactose, urine galactitol levels usually do not provide insight into acute and transient effects of galactose intake.

Reference Values

0-11 months: <109 mmol/mol creatinine
1-3 years: <52 mmol/mol creatinine
4-17 years: <16 mmol/mol creatinine
> or =18 years: <13 mmol/mol creatinine

Interpretation

The concentration of galactitol is provided along with reference ranges for patients with galactosemia and normal controls.

Cautions

No significant cautionary statements

Clinical Reference

1. Berry GT. Classic galactosemia and clinical variant galactosemia. In: Adam MP, Mirzaa GM, Pagon RA, et al, eds. GeneReviews [Internet]. University of Washington, Seattle; 2000. Updated March 11, 2021. Accessed September 25, 2025. Available at www.ncbi.nlm.nih.gov/books/NBK1518/
2. Walter JH, Fridovich-Keil JL. Galactosemia. In: Valle DL, Antonarakis S, Ballabio A, Beaudet AL, Mitchell GA. eds. The Online Metabolic and Molecular Bases of Inherited Disease. McGraw-Hill; 2019. Accessed September 25, 2025. Available at https://ommbid.mhmedical.com/content.aspx?sectionid=225081023&bookid=2709
3. OMIM entry 618881 Galactose mutarotase deficiency. Johns Hopkins University; 2020. Updated August 28, 2020. Accessed September 25, 2025. Available at https://omim.org/entry/618881
4. Pasquali M, Yu C, Coffee B. Laboratory diagnosis of galactosemia: a technical standard and guideline of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2018;20(1):3-11. doi:10.1038/gim.2017.172

Performance

Method Description

A total of 200 mcL of urine are spiked with a mixture of labeled internal standards, allowed to equilibrate, and evaporated. The dry residue is derivatized to form trimethylsilyl esters, then extracted with hexane. Specimens are analyzed by gas chromatography mass spectrometry, selected ion monitoring using ammonia chemical ionization and a stable isotope dilution method.(Jansen G, Muskiet F, Schierbeek H, et al. Capillary gas chromatography profiling of urinary, plasma, and erythrocyte sugars and polyols as their trimethylsilyl derivatives, preceded by a simple and rapid prepurification method. Clin Chim Acta. 1986;157:277-294; Marolt G, Kolar M. Analytical Methods for Determination of Phytic Acid and Other Inositol Phosphates: A Review. Molecules. 2020;26[1]:174)

PDF Report

No

Day(s) Performed

Tuesday, Friday

Report Available

3 to 7 days

Specimen Retention Time

3 months

Performing Laboratory Location

Mayo Clinic Laboratories - Rochester Main Campus

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

82542

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
GATOL	Galactitol, QN, U	47857-8

Result ID	Test Result Name	Result LOINC® Value
35831	Galactitol	47857-8
35832	Interpretation (GATOL)	59462-2
35833	Reviewed By	18771-6