MAYO CLINIC LABORATORIES

Test Definition: GALU

Galactose, QN, U

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

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: 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

Biochemical Genetics Patient Information (T602) is recommendation, see Special Instructions.

Reject Due To

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

Specimen Minimum Volume

0.5 mL

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Urine	Frozen (preferred)	365 days	
	Ambient	20 days	
	Refrigerated	20 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 (GALK), uridine diphosphate galactose-4-epimerase (GALE), and galactose mutarotase (GALM). 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. 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 (Gal-1-P) 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)	Gal-1-P (blood)
GALK	Elevated	Normal
GALT	Elevated	Elevated
GALE	Normal-Elevated	Elevated
GALM	Elevated	Normal-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

1. Berry GT: Classic galactosemia and clinical variant galactosemia. In: Adam MP, Ardinger HH, Pagon RA, et al. eds. GeneReviews [Internet]. University of Washington, Seattle; 2000. Updated March 9, 2017. Accessed May 3, 2021. Available at www.ncbi.nlm.nih.gov/books/NBK1518/

2. Walter JH, Fridovich-Keil JL: Galactosemia. In: Valle D, Beaudet AL, Vogelstein B, eds. The Online Metabolic and Molecular Bases of Inherited Disease. McGraw-Hill; 2019. Accessed May 3, 2021. Available at http://ommbid.mhmedical.com/content.aspx?bookid=971&sectionid=62672411

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

Specimen Retention Time

1 month

Performing Laboratory Location

Rochester

Test Definition: GALU

Galactose, QN, U

Fees & Codes

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 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	Reporting Name	LOINC®
8765	Galactose, QN, U	2310-1