

Overview

Useful For

Monitoring dietary therapy of patients with galactosemia due to deficiency of galactose-1-phosphate uridyltransferase or uridine diphosphate galactose-4-epimerase

Genetics Test Information

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

Galactose-1-phosphate is elevated in patients with galactosemia due to GALT deficiency or uridine diphosphate galactose-4-epimerase (GALE) deficiency, therefore is a suitable analyte for monitoring dietary compliance.

Testing Algorithm

See [Galactosemia Testing Algorithm](#) in Special Instructions

Special Instructions

- [Galactosemia Testing Algorithm](#)
- [Biochemical Genetics Patient Information](#)
- [Galactosemia-Related Test List](#)

Method Name

Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)

NY State Available

Yes

Specimen

Specimen Type

Whole Blood EDTA

Ordering Guidance

This test is used to monitor dietary therapy of patients with galactosemia due to deficiency of galactose-1-phosphate uridyltransferase (GALT) or uridine diphosphate galactose-4-epimerase (GALE).

This test is **not** appropriate for the 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.

This test is **not** appropriate for the diagnosis of epimerase deficiency, the preferred test to evaluate this deficiency is GALE / UDP-Galactose 4' Epimerase, Blood.

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

Multiple whole blood tests for galactosemia can be performed on 1 specimen. Prioritize order of testing when

submitting specimens. See [Galactosemia-Related Test List](#) in Special Instructions for a list of tests that can be ordered together.

Patient Preparation: Specimens collected following a meal can exhibit postprandial elevations. For infants, collect a specimen immediately prior to feeding to avoid this.

Container/Tube:

Preferred: Lavender top (EDTA)

Acceptable: Green top (sodium heparin)

Specimen Volume: 3 mL

Forms

1. [Biochemical Genetics Patient Information](#) (T602) is recommended, 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

2 mL

Reject Due To

Gross hemolysis	Reject
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Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole Blood EDTA	Refrigerated	72 hours	

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). Galactose-1-phosphate (Gal-1-P) accumulates in the erythrocytes of patients with galactosemia due to either GALT or GALE deficiency. The quantitative measurement of Gal-1-P is useful for monitoring compliance with dietary therapy for either deficiency. Gal-1-P is thought to be the causative factor for development of liver disease in these patients and, because of this, patients should maintain low levels and be monitored on a regular basis. The concentration of Gal-1-P in erythrocytes is the most sensitive index of dietary control.

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 due to GALT deficiency 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.

Epimerase deficiency galactosemia can be categorized into 3 types: generalized, peripheral, and intermediate. Generalized epimerase deficiency galactosemia results in profoundly decreased enzyme activity in all tissues, whereas peripheral epimerase deficiency galactosemia results in decreased enzyme activity in red and white blood cells, but normal enzyme activity in all other tissues. This is compared with intermediate epimerase deficiency galactosemia, which results in decreased enzyme activity in red and white blood cells and less than 50% of normal enzyme levels in other tissues.

Clinically, infants with generalized epimerase deficiency galactosemia develop symptoms such as liver and renal dysfunction and mild cataracts when on a normal milk diet, while infants with peripheral or intermediate epimerase deficiency galactosemia do not develop any symptoms. Generalized epimerase deficiency galactosemia is treated by a galactose- and lactose-restricted diet, which can improve or prevent the symptoms of renal and liver dysfunction and mild cataracts. Despite adequate treatment from an early age, individuals with generalized epimerase deficiency galactosemia remain at increased risk for developmental delay and intellectual disability. Unlike patients with classic galactosemia resulting from a GALT deficiency, females with generalized epimerase deficiency galactosemia experience normal puberty and are not at increased risk for premature ovarian failure. Based upon reports by newborn screening programs, the frequency of epimerase deficiency galactosemia in the United States ranges from approximately 1 in 6700 in African American infants to 1 in 70,000 in infants of European ancestry.

See [Galactosemia Testing Algorithm](#) in Special Instructions.

Reference Values

Reference interval (normal range) : < or =0.9 mg/dL

Therapeutic range: < or =4.9 mg/dL

Interpretation

The concentration of galactose-1-phosphate (Gal-1-P) is provided along with reference values for patients with galactosemia and normal controls. The recommended Gal-1-P goal for patients with galactosemia is less than or equal to 4.9 mg/dL.

See [Galactosemia Testing Algorithm](#) in Special Instructions for additional information.

Cautions

No significant cautionary statements.

Clinical Reference

1. Berry GT: Classic Galactosemia and Clinical Variant Galactosemia. In GeneReviews. Edited by RA Pagon, MP Adam, HH Ardinger, et al. University of Washington, Seattle. Updated 2017 Mar 9. Accessed 03/08/2019. Available at www.ncbi.nlm.nih.gov/books/NBK1518/

2. Walter JH, Fridovich-Keil JL: Galactosemia. In The Online Metabolic and Molecular Bases of Inherited Disease. Edited by D Valle, AL Beaudet, B Vogelstein. McGraw-Hill. Accessed 03/08/2019. Available at <http://ommbid.mhmedical.com/content.aspx?bookid=971§ionid=62672411>

Performance

Method Description

Packed RBCs are diluted with cold water and vortexed to lyse the cells, creating a hemolysate. The hemolysate is extracted with acetonitrile/methanol containing internal standard and then is centrifuged prior to injection onto a liquid chromatography-tandem mass spectrometry (LC-MS/MS) system. The ratio of the extracted peak area of Gal-1-P to its internal standard (13)C2-Gal-1-P as determined by LC-MS/MS is used to calculate the concentration of analyte, in mg/dL, in the sample.(Unpublished Mayo method)

PDF Report

No

Day(s) Performed

Thursday

Report Available

8 to 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

84378

LOINC® Information

Test ID	Test Order Name	Order LOINC Value
GAL1P	Galactose-1-Phosphate, RBC	2312-7

Result ID	Test Result Name	Result LOINC Value
24101	Galactose-1-Phosphate, RBC	2312-7