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 GAL1P / Galactose-1-Phosphate (Gal-1-P), Erythrocytes in monitoring diet.

Method Name
Gas Chromatography/Mass Spectrometry (GC/MS)

NY State Available
Yes

Specimen

Specimen Type
Urine

Necessary Information
Patient’s age is required.

Specimen Required
Supplies: Urine Tubes, 10 mL (T068)

Container/Tube: Plastic, 10-mL urine tube (T068)

Specimen Volume: 2 mL

Collection Instructions:
1. Collect a random urine specimen.
2. No preservative.

Forms
If not ordering electronically, complete, print, and send an Inborn Errors of Metabolism Test Request (T798) with the specimen.

Specimen Minimum Volume
1 mL

Reject Due To
| Hemolysis | NA |

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Galactosemia is an autosomal recessive disorder that results from a deficiency of 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 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 females 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 (GAL1P) 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

4â€“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


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 (TMS) 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 1986157:277-294)

PDF Report

No

Day(s) and Time(s) Test Performed

Tuesday; 8 a.m.

Analytic Time

3 days

Maximum Laboratory Time

9 days

Specimen Retention Time

3 months

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 Definition: GATOL
Galactitol, QN, U

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
82542

LOINC® Information

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