

Overview

Useful For

Screening for conditions associated with increased excretion of fructose, galactose, and xylose

This test is **not recommended** as a follow up test for abnormal newborn screening for galactosemia.

Reflex Tests

Test Id	Reporting Name	Available Separately	Always Performed
GALU	Galactose, QN, U	Yes	No

Testing Algorithm

Testing begins with carbohydrate analysis. If qualitative results are normal or abnormal but not indicative of galactose, testing is complete.

If qualitative results indicate the presence of galactose, then quantitative testing for galactose will be performed at an additional charge.

Special Instructions

- [Biochemical Genetics Patient Information](#)

Method Name

Thin-Layer Chromatography (TLC), Qualitative

NY State Available

Yes

Specimen

Specimen Type

Urine

Ordering Guidance

This test is not appropriate for evaluation of an abnormal newborn screen for galactosemia. For those cases, order GCT / Galactosemia Reflex, Blood and consider GAL1P / Galactose-1-Phosphate, Erythrocytes and GATOL / Galactitol, Quantitative, Urine.

Specimen Required

Supplies: Urine Tubes, 10 mL (T068)

Container/Tube: Plastic, 10-mL urine tube

Specimen Volume: 5 mL

Collection Instructions: Collect an early-morning (preferred) random urine specimen.

Forms

[Biochemical Genetics Patient Information](#) (T602) in Special Instructions.

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	Frozen (preferred)	21 days	
	Refrigerated	21 days	

Clinical & Interpretive

Clinical Information

Carbohydrates are a group of mono-, di-, and oligosaccharides of endogenous and exogenous sources. Their presence frequently reflects dietary consumption but can indicate specific pathology if either a particular saccharide or a particular excretory pattern is present. Most saccharides (except glucose) have low renal thresholds and are readily excreted in the urine.

In addition to several other saccharide species, chromatography of urinary saccharides identifies galactose and fructose and is, therefore, useful to screen for inborn errors of galactose and fructose metabolism. Xylose may also be detected in individuals with hereditary pentosuria, a benign trait with high frequency among individuals with Ashkenazi Jewish descent.

Reference Values

Negative

If positive, carbohydrate is identified.

Interpretation

An interpretive comment is provided.

Cautions

A number of compounds interfere with the assay and microbial contamination can lead to uninterpretable patterns of urinary saccharides. Retesting is recommended in these cases.

Clinical Reference

1. Steinmann B, Gitzelmann R, Van den Berghe G: Disorders of fructose metabolism. In: Valle D, Antonarakis S, Ballabio A, Beaudet A, Mitchell GA, eds. The Online Metabolic and Molecular Bases of Inherited Disease McGraw-Hill; 2019.

Accessed January 09, 2020. Available at

<http://ommbid.mhmedical.com/content.aspx?bookid=2709§ionid=225080452>

2. OMIM. #260800 Pentosuria; PNTSU. Johns Hopkins University; 1986. Updated July 9, 2016. Accessed April 23, 2021.

Available at <https://omim.org/entry/260800>

3. Gaughan S, Ayres L, Baker P II: Hereditary fructose intolerance. In: Adam MP, Ardinger HH, Pagon RA, et al, eds.

GeneReviews [Internet]. University of Washington, Seattle;2015. Updated February 18, 2021. Accessed April 23, 2021.

Available at www.ncbi.nlm.nih.gov/books/NBK333439

Performance

Method Description

The urine is chromatographed on a silica gel thin-layer plate. The sugars are located with naphthoresorcinol spray reagent and are identified by visual comparison with a sugar standard chromatographed on the same plate. If galactose is found, it is quantitated by an enzymatic method.(Prinz W, Meldrum W, Wilkinson L: A simple and rapid thin-layer chromatographic method for the identification of urinary carbohydrates. Clin Chim Acta. 1978;82:229-232; 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. Elsevier; 2017:1139-1158)

PDF Report

No

Day(s) Performed

Tuesday

Report Available

8 to 15 days

Specimen Retention Time

14 days

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

84377-Carbohydrate

82760-Galactose (if appropriate)

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
CHOU	Carbohydrate, U	16550-6

Result ID	Test Result Name	Result LOINC® Value
9255	Carbohydrate, U	16550-6