

## Overview

### Useful For

Monitoring effectiveness of treatment in patients with phosphomannomutase 2 deficiency (PMM2-CDG)

Establishing a baseline level prior to initiating treatment for PMM2-CDG

This test is **not useful for** diagnosing congenital disorders of glycosylation (CDG) in general or PMM2-CDG in particular

### Method Name

Gas Chromatography Mass Spectrometry (GC-MS)

### NY State Available

Yes

## Specimen

### Specimen Type

Urine

### Ordering Guidance

This is the preferred test for monitoring effectiveness of treatment in patients with phosphomannomutase 2 deficiency (PMM2-CDG). The preferred test for assessing sorbitol dehydrogenase (SORD) deficiency-related peripheral neuropathy is SORD / Sorbitol and Xylitol, Quantitative, Random, Urine

### 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**

Phosphomannomutase 2 deficiency, or PMM2-CDG, is the most common congenital disorder of glycosylation (CDG) accounting for about 50% of known CDG patients.

In many patients with PMM2-CDG, the urine polyols, sorbitol and mannitol, are elevated relative to controls. Sorbitol, in particular, has been shown to be positively correlated with severely affected patients in contrast to patients in the mild or moderate categories. It is also higher in patients with moderate peripheral neuropathy. Both mannitol and sorbitol were increased in patients with mild liver dysfunction.(1) Treatment options for PMM2-CDG remain limited however; current literature reports that the aldose reductase inhibitor, epalrestat, can correct the underlying enzyme deficiency in a majority of patients with PMM2-CDG.(2) Recent trials suggest that treatment with epalrestat, in addition to other therapeutic benefits, resulted in nearly normalized levels of sorbitol and mannitol relative to controls.(1)

**Reference Values**

Mannitol: <97 mmol/mol creatinine

Sorbitol: <35 mmol/mol creatinine

**Interpretation**

The quantitative results of sorbitol and mannitol are reported without added interpretation.

**Cautions**

No significant cautionary statements

**Clinical Reference**

1. Ligezka AN, Radenkovic S, Saraswat M, et al. Sorbitol is a severity biomarker for PMM2-CDG with therapeutic implications. *Ann Neurol*. 2021;90(6):887-900. doi:10.1002/ana.26245
2. Iyer S, Sam FS, DiPrimio N, et al. Repurposing the aldose reductase inhibitor and diabetic neuropathy drug epalrestat for the congenital disorder of glycosylation PMM2-CDG. *Dis Model Mech*. 2019;12(11):dmm040584. doi:10.1242/dmm.040584

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**Performance****Method Description**

A total of 200 mcL of urine is 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[3]: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**

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**

82542

**LOINC® Information**

## Test Definition: SORBU

Sorbitol and Mannitol, Quantitative, Random,  
Urine

Test ID	Test Order Name	Order LOINC® Value
SORBU	Sorbitol and Mannitol, QN, U	74447-4

Result ID	Test Result Name	Result LOINC® Value
614935	Sorbitol	48152-3
614936	Mannitol	47698-6
614937	Interpretation	59462-2
614938	Reviewed By	18771-6