

Reporting Title: Sorbitol and Xylitol, QN, U**Performing Location:** Rochester**Ordering Guidance:**

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

Necessary Information:

1. Patient's age is required.
2. 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 Requirements:

Supplies: Urine Tubes, 10mL (T068)

Container/Tube: Plastic, 10-mL urine tube

Specimen Volume: 2 mL

Collection Instructions:

1. Collect a random urine specimen.
2. No preservative needed.

Specimen Minimum Volume:

1 mL

Forms:

Biochemical Genetics Patient Information (T602)

Specimen Type	Temperature	Time	Special Container
Urine	Refrigerated (preferred)	28 days	
	Frozen	28 days	

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
620921	Interpretation	Alphanumeric		59462-2
620922	Sorbitol	Numeric	mmol/mol Cr	48152-3
620923	Xylitol	Numeric	mmol/mol Cr	48132-5
620927	Reviewed By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

82542

Reference Values:

Sorbitol:

<35 mmol/mol creatinine

Xylitol:

<351 mmol/mol creatinine