## 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, $10-\mathrm{mL}$ urine tube
Specimen Volume: 2 mL
Collection Instructions:

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

## 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 ${ }^{\circledR}$ |
| :--- | :--- | :--- | :--- | :--- |
| 620921 | Interpretation | Alphanumeric |  | $59462-2$ |
| 620922 | Sorbitol | Numeric | $\mathrm{mmol} / \mathrm{mol} \mathrm{Cr}$ | $48152-3$ |
| 620923 | Xylitol | Numeric | $\mathrm{mmol} / \mathrm{mol} \mathrm{Cr}$ | $48132-5$ |
| 620927 | Reviewed By | Alphanumeric |  | $18771-6$ |

LOINC ${ }^{\circledR}$ 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

