

Notification Date: February 13, 2024 Effective Date: February 20, 2024

Sorbitol and Xylitol, Quantitative, Random, Urine

Test ID: SORD

Useful for:

Screening for sorbitol dehydrogenase deficiency--related neuropathy.

Genetic Information:

This test is used to aid in the diagnosis of patients with sorbitol dehydrogenase-related peripheral neuropathy.

Methods:

Gas Chromatography Mass Spectrometry (GC-MS)

Reference Values:

Sorbitol:

<35 mmol/mol creatinine

Xylitol:

<351 mmol/mol creatinine

Specimen Requirements:

Supplies: Urine Tubes, 1mL (T068)

Container/Tube: Plastic, 10-mL urine tube

Specimen Volume: 2 mL

Collection Instructions: 1. Collect a random urine specimen.

2. No preservative needed.

Minimum Volume: 1 mL

Necessary Information:

1. Patient's age is required.

Specimen Stability Information:

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

Cautions:

A positive test result is diagnostic of sorbitol dehydrogenase (SORD) deficiency-related neuropathy; however, it is strongly recommended to follow-up with molecular analysis. Molecular analysis of the *SORD* gene is complicated by the *SORD2P* pseudogene and specific molecular testing approaches maybe required.

CPT Code:

82542

Day(s) Performed: Tuesday Report Available: 3 to 9 days

Questions

Contact Melissa Tricker-Klar, Laboratory Resource Coordinator at 800-533-1710.