

## Sorbitol and Xylitol, Quantitative, Whole Blood

**Test ID:** SORDB

**Useful for:**

Screening and treatment monitoring for sorbitol dehydrogenase deficiency-related neuropathy.

**Genetic Information:**

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

**Methods:**

Gas Chromatography Mass Spectrometry (GC-MS) Stable Isotope Dilution Analysis

**Reference Values:**

Sorbitol: < or =15.0 nmol/mL

Xylitol: < or =2.0 nmol/mL

**Specimen Requirements:**

**Patient Preparation:**            **Fasting: 8 hours, required**

**Preferred:**                        Lavender top (EDTA)

**Acceptable:**                    Green top (sodium heparin)

**Specimen Volume:**            1 mL

**Collection Instructions:**    1. Invert several times to mix blood.  
2. Freeze whole blood specimens in the original tube. Frozen aliquots from well-mixed specimens are also acceptable.  
3. Send frozen.

**Minimum Volume:**            0.5 mL

**Specimen Stability Information:**

Specimen Type	Temperature	Time
Whole blood	Frozen (preferred)	90 days
	Refrigerated	31 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 may be required to identify both causative variants.

**CPT Code:**

82542

**Day(s) Performed:** Friday**Report Available:** 5 to 11 days**Questions**

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