

Sorbitol and Xylitol, Quantitative, Blood Spot

Test ID: SXDBS

Useful for:

Screening and treatment monitoring for sorbitol dehydrogenase deficiency-related neuropathy

Methods:

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

Reference Values:

Sorbitol: < or =30.0 nmol/mL

Xylitol: < or =10.0 nmol/mL

Specimen Requirements:

Fasting: 8 hours, required. Infants should not fast overnight; collect just before next feeding.

Supplies:	Card-Blood Spot Collection (Filter Paper) (T493)
Preferred:	Card-Blood Spot Collection (Filter Paper)
Acceptable:	PerkinElmer 226 filter paper, Munktell filter paper, Whatman protein Saver 903 paper, local newborn screening card, or blood collected in tubes containing EDTA (preferred) or sodium heparin and dried on filter paper.
Specimen Volume:	2 Blood spots
Collection Instructions:	1. An alternative blood collection option for a patient older than 1 year is a fingerstick. For detailed instructions, see How to Collect a Dried Blood Spot Sample. 2. Completely fill at least 2 circles on the filter paper card (approximately 100 microliters blood per circle). 3. Let blood dry on filter paper at ambient temperature in a horizontal position for a minimum of 3 hours. 4. Do not expose specimen to heat or direct sunlight. 5. Do not stack wet specimens. 6. Keep specimen dry.
Minimum Volume:	1 Blood spot

Specimen Stability Information:

Specimen Type	Temperature	Time	Special Container
Whole blood	Ambient (preferred)	31 days	FILTER PAPER
	Frozen	90 days	FILTER PAPER
	Refrigerated	90 days	FILTER PAPER

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.