MAYO CLINIC LABORATORIES

Test Definition: SORDB

Sorbitol and Xylitol, Quantitative, Whole Blood

Overview

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

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

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

NY State Available
Yes

Specimen

Specimen Type
Whole blood

Ordering Guidance
This is a test for diagnosis and treatment monitoring for sorbitol dehydrogenase deficiency-related peripheral neuropathy.

Necessary Information
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 Required
Patient Preparation:
Fasting: 8 hours, required
Container/Tube:
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.

Forms
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 Minimum Volume

0.5 mL

Reject Due To

All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

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

Clinical & Interpretive

Clinical Information

Sorbitol dehydrogenase (SORD) deficiency is an autosomal recessive condition caused by biallelic variants in the *SORD* gene resulting in peripheral neuropathy, which may present as clinically similar to Charcot-Marie-Tooth disease type 2 or distal hereditary motor neuropathy. The SORD enzyme catalyzes the breakdown of sorbitol to fructose. In patients with SORD deficiency-related peripheral neuropathy, two urine polyols, sorbitol and xylitol, are elevated in both blood and urine when compared to controls. Polyols are sugar alcohols that have been identified in blood, urine, and cerebrospinal fluid. An abnormal blood and urine polyol result suggestive of SORD deficiency-related peripheral neuropathy should be confirmed with molecular genetic analysis. For molecular confirmation, genetic testing for *SORD* can be performed (CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies; specify gene list ID: NEUROLOGY-S3NL4H).

Reference Values

Sorbitol: < or =15.0 nmol/mL
Xylitol: < or =2.0 nmol/mL

Interpretation

An interpretive report will be provided.

All profiles are reviewed by the laboratory director and interpretation is based on pattern recognition. A detailed interpretation is given, including an overview of the results and of their significance, a correlation to available clinical information, and recommendations for in vitro confirmatory studies (molecular analysis).

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.

Clinical Reference

1. Cortese A, Zhu Y, Rebelo AP, et al. Biallelic mutations in SORD cause a common and potentially treatable hereditary neuropathy with implications for diabetes. Nat Genet. 2020;52(5):473-481. doi:10.1038/s41588-020-0615-4
2. Lassuthova P, Mazanec R, Stanek D, et al. Biallelic variants in the SORD gene are one of the most common causes of hereditary neuropathy among Czech patients. Sci Rep. 2021;11(1):8443. doi:10.1038/s41598-021-86857-0
3. Pons N, Fernandez-Eulate G, Pegat A, et al. SORD-related peripheral neuropathy in a French and Swiss cohort: Clinical features, genetic analyses, and sorbitol dosages. Eur J Neurol. 2023;30(7):2001-2011. doi:10.1111/ene.15793
4. Zhu Y, Lobato AG, Rebelo AP, et al. Sorbitol reduction via govorestat ameliorates synaptic dysfunction and neurodegeneration in sorbitol dehydrogenase deficiency. JCI Insight. 2023;8(10):e164954. Published 2023 May 22. doi:10.1172/jci.insight.164954.
5. Bontrager JE, White AL, Brigatti KW, et al. Urine Sorbitol and Xylitol for the Diagnosis of Sorbitol Dehydrogenase Deficiency-Related Neuropathy. Neurology. 2025;105(11):e214425. doi:10.1212/WNL.0000000000214425

Performance

Method Description
Fifty microliters of lysed whole blood are spiked with a mixture of labeled internal standards and evaporated. The dry residue is derivatized to form trimethylsilyl ethers, then extracted with hexane. Specimens are analyzed by gas chromatography mass spectrometry, selected ion monitoring using positive ammonia chemical ionization and stable isotope dilution.(Unpublished Mayo method)

PDF Report
No

Day(s) Performed
Friday

Report Available
5 to 11 days

Specimen Retention Time
3 months

Performing Laboratory Location
Mayo Clinic Laboratories - Rochester Main Campus

Fees & Codes

Fees
- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact Customer Service.

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Test Classification

This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. It has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information

82542

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
SORDB	Sorbitol and Xylitol, QN, WB	In Process

Result ID	Test Result Name	Result LOINC® Value
623504	Interpretation	59462-2
623502	Sorbitol	In Process
623503	Xylitol	In Process
623505	Reviewed By	18771-6