MAYO CLINIC LABORATORIES

Test Definition: SXDBS

Sorbitol and Xylitol, Quantitative, Blood Spot

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

Blood Spot

Ordering Guidance

This is a test for diagnosis and treatment monitoring for sorbitol dehydrogenase (SORD) 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

Fasting: 8 hours, required. Infants **should not fast** overnight; collect just before next feeding.
Supplies: Card-Blood Spot Collection (Filter Paper) (T493)
Container/Tube:
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.

Additional Information:

1. For collection instructions, see Blood Spot Collection Instructions
2. For collection instructions in Spanish, see Blood Spot Collection Card-Spanish Instructions (T777)
3. For collection instructions in Chinese, see Blood Spot Collection Card-Chinese Instructions (T800)

Forms

1. 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.
2. If not ordering electronically, complete, print, and send Neurology Specialty Testing Client Test Request (T732) with the specimen.

Specimen Minimum Volume

1 Blood spot

Reject Due To

Blood spot specimen that shows serum rings or has multiple layers	Reject
Insufficient specimen	Reject
Non-approved filter papers	Reject

Specimen Stability Information

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

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 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. Abnormal blood and urine (SORD / Sorbitol and Xylitol, Quantitative, Random, Urine) polyol results 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 =30.0 nmol/mL
Xylitol: < or =10.0 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. doi:10.1172/jci.insight.164954

Performance

Method Description

Blood spot specimens 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 (GC/MS) 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
Normal result: 1 month; Abnormal result: Indefinitely

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.

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
SXDBS	Sorbitol and Xylitol, Quant, DBS	In Process

Result ID	Test Result Name	Result LOINC® Value
623692	Interpretation	59462-2
623690	Sorbitol	In Process
623691	Xylitol	In Process
623693	Reviewed By	18771-6