Test Definition: SORD

Sorbitol and Xylitol, Quantitative, Random, Urine

Overview

Useful For
Screening for sorbitol dehydrogenase deficiency-related neuropathy

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

Method Name
Gas Chromatography Mass Spectrometry (GC-MS)

NY State Available
Yes

Specimen

Specimen Type
Urine

Ordering Guidance
This is the preferred test for assessing sorbitol dehydrogenase deficiency-related peripheral neuropathy. The preferred test for monitoring effectiveness of treatment in patients with phosphomannomutase 2 deficiency (PMM2-CDG) is SORBU / Sorbitol and Mannitol, Quantitative, Random, Urine.

Necessary Information
1. Patient's age is required.
2. 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
Supplies: Urine Tubes, 10mL (T068)
Container/Tube: Plastic, 10-mL urine tube
Specimen Volume: 2 mL
Collection Instructions:
1. Collect a random urine specimen.
2. No preservative.
3. Refrigerate immediately.

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 1 of the following forms with the specimen:
-Neurology Specialty Testing Client Test Request (T732)
-Biochemical Genetics Test Request (T798

Specimen Minimum Volume
1 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
Urine	Refrigerated (preferred)	28 days	
	Frozen	28 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 when compared to controls. Polyols are sugar alcohols that have been identified in blood, urine, and cerebrospinal fluid. An abnormal urine and blood polyols 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:
<35 mmol/mol creatinine

Xylitol:
<351 mmol/mol creatinine

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. 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
2. 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
3. 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
4. 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
5. 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

A total of 200 mcL of urine is spiked with a mixture of labeled internal standards, allowed to equilibrate, and evaporated. The dry residue is derivatized to form trimethylsilyl esters, then extracted with hexane. Specimens are analyzed by gas chromatography mass spectrometry, selected ion monitoring using ammonia chemical ionization and a stable isotope dilution method.(Jansen G, Muskiet F, Schierbeek H, Berger R, van der Slik W. Capillary gas chromatography profiling of urinary, plasma, and erythrocyte sugars and polyols as their trimethylsilyl derivatives, preceded by a simple and rapid prepurification method. Clin Chim Acta. 1986; 157[3]:277-294; Marolt G, Kolar M. Analytical methods for determination of phytic acid and other inositol phosphates: A review. Molecules. 2020;26[1]:174)

PDF Report

No

Day(s) Performed

Tuesday, Friday

Report Available

3 to 7 days

Specimen Retention Time

3 months

Performing Laboratory Location

Mayo Clinic Laboratories - Rochester Main Campus

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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
SORD	Sorbitol and Xylitol, QN, U	74447-4

Result ID	Test Result Name	Result LOINC® Value
620921	Interpretation	59462-2
620922	Sorbitol	48152-3
620923	Xylitol	48132-5
620927	Reviewed By	18771-6