



Patient ID C7028846-0005917	Patient Name ABNORMAL, SORD	Birth Date 1991-05-15	Sex F	Age 32
Order Number X100452012	Client Order Number X100452012	Ordering Physician unknown	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 18 Jan 2024 06:30		

Sorbitol and Xylitol, QN, U

Interpretation

1 MCR

POSITIVE In this sample, the excretions of sorbitol and xylitol were elevated. This result is suggestive of a biochemical diagnosis of sorbitol dehydrogenase (SORD) deficiency with peripheral neuropathy. We recommend molecular genetic analysis of the SORD gene (MCL test CGPH, gene list ID: NEUROLOGY-S3NL4H). Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) if you have any questions.

ADDITIONAL INFORMATION

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

Sorbitol



High

72 mmol/mol Cr

MCR

Reference Value
<35

Xylitol



High

684 mmol/mol Cr

MCR

Reference Value
<351

Reviewed By

Amy Breitlow

MCR

Received: 18 Jan 2024 13:56

Reported: 18 Jan 2024 14:03

Laboratory Notes

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

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292