

Patient ID 321	Patient Name TESTRNV, MSDBS ATLAS	Birth Date 1968-02-19	Sex M	Age 55
Order Number X100426957	Client Order Number X100426957	Ordering Physician TESTING	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 14 Aug 2023 05:00		

Multiple sulfatase deficiency, BS

Reason for Referral

Rule out MSD

Iduronate-2-sulfatase



1.55 nmol/mL/h

MCR

MCR

Reference Value
>4.30

Heparan-N-sulfatase



0.02 nmol/mL/h

MCR

Reference Value
>0.06

N-acetylgalactosamine-6-sulfatase



0.42 nmol/mL/h

MCR

Reference Value
>0.70

Interpretation

POSITIVE In this dried blood spot sample, the activities of several sulfatase enzymes iduronate-2-sulfatase, heparan-N-

sulfatase, and N-acetylgalactosamine-6-sulfatase were reduced indicating that this individual may be affected with multiple sulfatase deficiency (MSD). However, a sample integrity issue (e.g., prolonged exposure to heat) cannot be excluded. If clinically indicated, consider further testing to include urine sulfatide and glycosaminoglycan analyses (MCL test LDS) and/or molecular genetic analysis of the SUMF1 gene (MCL test CGPH, gene list ID: IEMCP-PCUBX1). Molecular genetic testing could be performed on the existing sample by calling MCL (1-800-533-1710) within one month. Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) if you have any questions.

ADDITIONAL INFORMATION

Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)

Reviewed By

Marie Quade

MCR

Received: 14 Aug 2023 16:06

Reported: 14 Aug 2023 17:35

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