

Patient ID 321	Patient Name TESTRNV, MSDW ATLAS	Birth Date 1968-02-19	Sex M	Age 55
Order Number X100426959	Client Order Number X100426959	Ordering Physician TESTING	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 14 Aug 2023 05:00		

Multiple sulfatase deficiency, WBC

Reason for Referral

Rule out MSD

MCR

Interpretation

1 MCR

Iduronate-2-sulfatase

 **1.23 nmol/h/mg Prot**
 Low

MCR

Reference Value
>2.20

POSITIVE In this sample, the activities of several sulfatase enzymes (iduronate-2-sulfatase, N-acetylglucosamine-6-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 LSDS) and/or molecular genetic analysis of the SUMF1 gene (MCL test CGPH, gene list ID: IEMCP-PCUBX1). Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) if you have any questions.

Heparan-N-sulfatase

2.45 nmol/h/mg Prot

MCR

Reference Value
>0.13

ADDITIONAL INFORMATION

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

N-acetylglucosamine-6-sulfatase

 **0.02 nmol/h/mg Prot**
 Low

MCR

Reference Value
>0.03

N-acetylgalactosamine-6-sulfatase

 **0.55 nmol/h/mg Prot**
 Low

MCR

Reference Value
>1.60

Reviewed By

Marie Quade

MCR

Received: 14 Aug 2023 16:08

Reported: 14 Aug 2023 18:31

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