

Patient ID C7028846-0005471	Patient Name TEST, MPS3B A	Birth Date 1977-03-05	Sex M	Age 46
Order Number X100426880	Client Order Number X100426880	Ordering Physician unkdr	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 07 Aug 2023 23:15		

MPS III (Three) Panel, BS

Reason for Referral

Follow up of known MPS IIIA

Heparan-N-sulfatase



0.02 nmol/mL/h

MCR

MCR

Reference Value
>0.06

may be affected with mucopolysaccharidosis type IIIA (Sanfilippo syndrome A). The other enzymes analyzed by this method were normal. If clinically indicated, consider further testing to include glycosaminoglycan analysis in blood (MCL test MPSBS) and/or confirmation by molecular genetic analysis of the SGSH gene (MCL test CGPH, gene list ID: IEMCP-BXML7T). If not already ordered, these 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.

N-acetyl-alpha-D-glucosaminidase

0.93 nmol/mL/h

MCR

Reference Value
>0.70

ADDITIONAL INFORMATION

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

Heparan-alpha-glucosaminide N-acetyltransferase

0.75 nmol/mL/h

MCR

Reference Value
>0.50

Reviewed By

Marie Quade

MCR

Interpretation

1 MCR

POSITIVE In this dried blood spot sample, the activity of heparan-N-sulfatase was reduced indicating that this individual

Received: 14 Aug 2023 15:30

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

Report Status: Final