

Patient ID 321	Patient Name TESTRNV, MPS4B ATLAS	Birth Date 1968-02-19	Sex M	Age 55
Order Number X100426953	Client Order Number X100426953	Ordering Physician TESTING	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 14 Aug 2023 05:00		

MPS IV Panel, BS

Reason for Referral

Rule out MPS IV

N-acetylgalactosamine-6-sulfatase

1.22 nmol/mL/h

MCR

MCR

Reference Value
>0.70

Beta-galactosidase

 0.09 nmol/mL/h
Low

MCR

Reference Value
>1.30

Interpretation

POSITIVE In this dried blood spot sample, the activity of beta-galactosidase was reduced indicating that this individual may be affected with either mucopolysaccharidosis type IVB (Morquio syndrome B) or GM1 gangliosidosis. The other enzyme analyzed by this method, N-acetylgalactosamine-6-sulfatase, was normal.

1 MCR

Please note that beta-galactosidase may also be deficient in galactosialidosis. If clinically indicated, consider further testing to include glycosaminoglycan analysis in blood (MCL test MPSBS) or urine (MCL test MPSQU), urine oligosaccharide analysis (MCL test OLIGU) and/or confirmation by molecular genetic analysis of the GLB1 gene (MCL test CGPH, gene list ID: IEMCP-D5F3YS). If not already ordered, MPSBS and/or CGPH 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 16:05

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