

Patient ID 321	Patient Name TESTRNV, GUSBW ATLAS	Birth Date 1968-02-19	Sex M	Age 55
Order Number X100426951	Client Order Number X100426951	Ordering Physician TESTING	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 14 Aug 2023 05:00		

Beta-Glucuronidase, WBC

Reason for Referral

Rule out MPS VII

Beta-glucuronidase


2.30 nmol/h/mg Prot

Reference Value
>3.50

MCR

MCR

Interpretation

1 MCR

POSITIVE In this sample, the activity of beta-glucuronidase in leukocytes was reduced indicating that this individual may be affected with mucopolysaccharidosis type VII (Sly syndrome). If clinically indicated, consider further testing to include glycosaminoglycan analysis in urine (MCL test MPSQU) and/or confirmation by molecular genetic analysis of the GUSB gene (MCL test CGPH, gene list ID: IEMCP-L613TF). 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 16:04

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