

|                                                       |                                          |                                       |                 |                  |
|-------------------------------------------------------|------------------------------------------|---------------------------------------|-----------------|------------------|
| Patient ID<br><b>C7028846-0005478</b>                 | Patient Name<br><b>TEST, MPS3W A</b>     | Birth Date<br><b>1987-04-01</b>       | Sex<br><b>F</b> | Age<br><b>36</b> |
| Order Number<br><b>X100427055</b>                     | Client Order Number<br><b>X100427055</b> | Ordering Physician<br><b>unkdr</b>    | Report Notes    |                  |
| Account Information<br><b>C7028846 DLMP Rochester</b> |                                          | Collected<br><b>14 Aug 2023 23:15</b> |                 |                  |

## MPS III (Four) Panel, WBC

### Reason for Referral

Follow up of known MPS IIIB

### Heparan-N-sulfatase

1.35 nmol/h/mg Prot

### N-acetyl-alpha-D-glucosaminidase

 0.07 nmol/h/mg Prot  
Low

### Heparan-alpha-glucosaminide N-acetyltransferase

6.67 nmol/h/mg Prot

### N-acetylglucosamine-6-sulfatase

1.84 nmol/h/mg Prot

MCR

MCR

Reference Value  
>0.13

MCR

Reference Value  
>0.09

MCR

Reference Value  
>0.24

MCR

Reference Value  
>0.03

### Interpretation

1 MCR

**\*POSITIVE\*** In this sample, the activity of N-acetyl-alpha-D-glucosaminidase in leukocytes was reduced indicating that this individual may be affected with mucopolysaccharidosis type IIIB (Sanfilippo syndrome B). The other enzymes analyzed by this method were normal. If clinically indicated, consider further testing to include glycosaminoglycan analysis in blood (MCL test MPSBS or MPSEB) or urine (MCL test MPSQU) and/or confirmation by molecular genetic analysis of the NAGLU gene (MCL test CGPH, gene list ID: IEMCP-KG2Q99). 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

MCR

Marie Quade

Received: 15 Aug 2023 15:02

Reported: 15 Aug 2023 15:17

### 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 |
|------|--------------------------------------------------|------------------------------------------|-----------------------------|------------------|
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