



|                                                |                                        |                                |              |           |
|------------------------------------------------|----------------------------------------|--------------------------------|--------------|-----------|
| Patient ID<br>321                              | Patient Name<br>IMPLEMENTATION, TEST T | Birth Date<br>1973-05-01       | Sex<br>F     | Age<br>51 |
| Order Number<br>X100481900                     | Client Order Number<br>X100481900      | Ordering Physician<br>123      | Report Notes |           |
| Account Information<br>C7028846 DLMP Rochester |                                        | Collected<br>06 Nov 2024 06:30 |              |           |

MPS I Biomarker, BS

Interpretation

1 MCR

\*POSITIVE\* In this sample, the MPS I specific glycosaminoglycan fragment is elevated and suggestive of a biochemical diagnosis of Mucopolysaccharidosis type I. We recommend follow up investigations to include urine mucopolysaccharides (MCL test MPSQU), alpha-L-iduronidase activity in leukocytes (MCL test IDUAW), and molecular genetic analysis of the IDUA gene (MCL test MPS1Z). 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

Amy Breitlow

Received: 06 Nov 2024 08:08

Reported: 06 Nov 2024 08:17

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 |
|------|--------------------------------------------------|------------------------------------------|----------------------|------------------|
| MCR  | Mayo Clinic Laboratories - Rochester Main Campus | 200 First Street SW, Rochester, MN 55905 | Nikola Baumann Ph.D. | 24D0404292       |