

Patient ID 321	Patient Name IMPLEMENTATION, TEST T	Birth Date 1973-05-01	Sex F	Age 51
Order Number X100477427	Client Order Number X100477427	Ordering Physician 123	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 25 Sep 2024 08:15		

Lysosomal Disorders Scrn, U

Lysosomal Disorders Interpretation

Dermatan Sulfate

0.93 mg/mmol Cr

MCR
Reference Value
≤1.00

Heparan Sulfate

0.24 mg/mmol Cr

MCR
Reference Value
≤0.25

Chondroitin-6 Sulfate

1.02 mg/mmol Cr

MCR
Reference Value
≤1.50

Keratan Sulfate

0.04 mg/mmol Cr

MCR
Reference Value
≤0.30

Free Sialic Acid

30 mmol/mol Cr

MCR
Reference Value
≤38

Total Sialic Acid

 575 mmol/mol Cr
High

MCR
Reference Value
≤262

Total/Free Sialic Acid Ratio

 19.30
High

MCR
Reference Value
3.35–15.81

Ceramide Trihexosides

Negative

MCR
Reference Value
Negative

Sulfatides

Negative

MCR
Reference Value
Negative

Oligosaccharides

 Positive
Abn

MCR
Reference Value
Negative

Lysosomal Disorders Interpretation

POSITIVE In this urine sample, the comprehensive screen for lysosomal disorders is suggestive of a biochemical diagnosis of sialidosis. We recommend confirmatory testing by molecular genetic analysis of the NEU1 gene (MCL test CGPH, gene list ID: IEMCP-1HNNPQ). 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) and Matrix-assisted laser desorption ionization time-of-flight mass spectrometry (MALDI-TOF MS)

Reviewed By

Marie Quade

MCR

Received: 25 Sep 2024 09:55

Reported: 25 Sep 2024 13:30

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