

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  


MCR  
Reference Value  
≤262

#### Total/Free Sialic Acid Ratio

 19.30  


MCR  
Reference Value  
3.35–15.81

#### Ceramide Trihexosides

Negative

MCR  
Reference Value  
Negative

#### Sulfatides

Negative

MCR  
Reference Value  
Negative

#### Oligosaccharides

 Positive  


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