

Patient ID 321	Patient Name TESTRNV, IMPLEMENTATION	Birth Date 2008-06-06	Gender F	Age 11
Order Number X100310892	Client Order Number X100310892	Ordering Physician 321	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 27 Jan 2020 10:02		

Lysosomal Storage Disorders Scrn, U

Lysosomal Storage Disorders Interp

1 MCR

POSITIVE In this urine sample, the comprehensive profile of ceramide trihexosides, sulfatides, glycosaminoglycans, and oligosaccharides is suggestive of a biochemical diagnosis of alpha-mannosidosis. We recommend confirmatory testing of alpha-mannosidase activity (MCL test MANN) and/or molecular genetic analysis of the MAN2B1 gene (included on MCL test LPGD). 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

MCR

Marie Quade

Received: 27 Jan 2020 11:01

Reported: 27 Jan 2020 12:00

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