

Patient ID SA00123661	Patient Name TESTINGRNV, GAAW SAMPLE A	Birth Date 2019-10-01	Gender M	Age 2 W
Order Number SA00123661	Client Order Number SA00123661	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 17 Oct 2019 08:00		

Acid Alpha-Glucosidase, Leukocytes

Acid Alpha-Glucosidase, Leukocytes


0.30 nmol/h/mg Prot
MCR
Reference Value
 ≥1.50

Reviewed By

Marie Quade

MCR
Received: 17 Oct 2019 13:28

Reported: 17 Oct 2019 13:40

Interpretation

1 MCR

In this sample, the activity of acid alpha-glucosidase (GAA) was markedly reduced indicating that this individual is affected with early onset Pompe disease. We recommend follow up investigations to include urine glucotetrasaccharides (MCL test HEX4) and molecular genetic analysis of the GAA gene (MCL test GAAZ; could be performed on existing specimen by calling MCL within one month). 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)

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