



Patient ID SA00110717	Patient Name TESTINGRNA, SAMPLEREPORT	Birth Date 1982-06-29	Gender F	Age 36
Order Number SA00110717	Client Order Number SA00110717	Ordering Physician CLIENT, CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 02 Oct 2018 12:00		

Pompe Disease, BS

Interpretation

1 MCR

POSITIVE

In this blood spot sample, the following results were observed:

Acid Alpha-glucosidase (GAA) activity:

7.0 nmol/mL/hr (1%ile of reference population = 10.0)

(Cr/Crn)/GAA ratio:

2.4 (99%ile of reference population = 1.0)

Range of Pompe disease cases (2–5; N=4)

Range of false positive cases (6–7; N=2)

Score of 12 plex CLIR post-analytical tool for GAA deficiency: 35

(informative >20; percentile rank in comparison to known cases:

95%, N=4)

INTERPRETATION

These results are suggestive of a biochemical diagnosis of Pompe disease (glycogen storage disease type II), type to be determined by further biochemical and molecular studies. We recommend follow up investigations to include acid alpha-glucosidase activity in another dried blood spot or leukocytes, urine glucotetrasaccharides, and comprehensive molecular genetic analysis of the GAA gene.

ADDITIONAL INFORMATION

Flow Injection Analysis-Tandem Mass Spectrometry (FIA-MS/MS)

Reviewed By

MCR

Amy Breitlow

Received: 03 Oct 2018 12:56

Reported: 03 Oct 2018 13:16

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