



Patient ID SA00092799	Patient Name TESTINGRNV, SAMPLEREPORT	Birth Date 2017-06-19	Gender F	Age 2 D
Order Number SA00092799	Client Order Number SA00092799	Ordering Physician CLIENT, CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 21 Jun 2017 12:00		

Pompe Disease 2ND Tier NBS, BS

Interpretation

1 MCR

POSITIVE

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

Acid Alpha-glucosidase (GAA) activity:
1.0 nmol/mL/hr (abnormal <6.1)

(Cr/Crn)/GAA ratio:
1.6 (99%ile of reference population = 1.0)

Range of Pompe disease cases (2.0–5.0; N=100)
Range of false positive cases (1.0–6.0; N=25)

Score of 12 plex CLIR post-analytical tool for GAA deficiency: 65
(informative >20; percentile rank in comparison to known cases:
95%, N=100)

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

Birth Weight (grams, XXXX)

MCR

2350 g

Time of Birth (24hr time, XX:XX)

MCR

13:45 Time

Gestational Age (weeks, XX.X)

MCR

38.2 wk

Received: 22 Jun 2017 12:21

Reported: 22 Jun 2017 12:32

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