



Pompe Disease Second-Tier Newborn Screening,
Blood Spot

Patient ID SA00106722	Patient Name TESTINGRN, SAMPLEREPORT	Birth Date 2018-05-01	Gender F	Age 2 W
Order Number SA00106722	Client Order Number SA00106722	Ordering Physician CLIENT, CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 15 May 2018 07:00		

Pompe Disease 2ND Tier NBS, BS

Interpretation

1 MCR

NEGATIVE

Acid Alpha-glucosidase (GAA) activity:
6.4 nmol/mL/hr (1%ile of reference population = 5.3)

However, the integrated interpretation of Acid Alpha-glucosidase (GAA) activity and of the (creatinine)/GAA ratio was not indicative of a possible biochemical diagnosis of Pompe disease (glycogen storage disease type II) and the reduced activity of GAA is likely related to either carrier status or presence of pseudodeficiency alleles.

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: 16 May 2018 06:55

Reported: 16 May 2018 07:02

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