

Patient ID SA00167611	Patient Name TESTINGRNV, SAMPLEREPORT	Birth Date 2024-04-07	Sex F	Age 2 D
Order Number SA00167611	Client Order Number SA00167611	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 09 Apr 2024 08:00		

LSD/X-ALD Newborn Screen, BS

LSD/X-ALD Newborn Screen Result

1 MCR

POSITIVE

In this blood spot sample, the following abnormal result was observed:

Acid Alpha-glucosidase (GAA) activity:
1.0 nmol/mL/hr (1st percentile of reference population: 3.5;
N=10000)

The score of CLIR post-analytical tool for GAA deficiency: 65
(informative >30; percentile rank in comparison to known cases
95%, N=500)

INTERPRETATION

These results are indicative of a biochemical diagnosis of Pompe disease (glycogen storage disease type II). We recommend follow up investigations to include urine glucotetrasaccharides (MCL test HEX4), CK, ALT, AST, LDH, and comprehensive molecular genetic analysis of the GAA gene (MCL test GAAZ). Please contact the Biochemical Genetics consultant or genetic

counselor on call (Tel. 1-800-533-1710) if you have any questions.

ADDITIONAL INFORMATION

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

Reviewed By

Amy Breitlow

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Birth Weight (grams, XXXX)

2750 g

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

13:14 Time

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Gestational Age (weeks, XX.X)

37.2 wk

MCR

Received: 10 Apr 2024 07:32

Reported: 10 Apr 2024 07:45

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