

# Newborn Screening Quality Assurance Program

PROFICIENCY TESTING

Lysosomal Storage Disorder  
Quarterly Report

Volume 5, No. 1

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## INTRODUCTION

This report is the Quarterly summary of all data reported within the specified data-reporting period for the Quarter 1, 2016, proficiency testing (PT) program for Lysosomal Storage Disorders (LSD) in dried blood spots (DBS) to detect Krabbe disease, Pompe disease, and Mucopolysaccharidosis Type I (MPS-I). The attached tables provide the certification profiles for the distributed specimens, the verification of your reported data, and the summary of reported analytical and categorical results.

On January 11, 2016 a panel of five unknown DBS specimens was distributed to eight laboratories in the United States to analyze Galactocerebrosidase (GALC) for Krabbe disease, Acid Alpha-Glucosidase (GAA) for Pompe disease, and alpha-L-iduronidase (IDUA) for MPS-I in whole blood.

## PARTICIPANT RESULTS

This panel of DBS specimens were prepared from human blood, including cord blood from unaffected individuals and leuko-depleted adult blood restored with lymphoblast cells derived from patients with LSD (specimens 116L1, 116L2, 116L3, 116L4, and 116L5).

We processed data from seven participants. Laboratories were asked to report quantitative results for GALC, GAA, and IDUA in  $\mu\text{mol/hr/L}$  units and qualitative results as “No follow-up required (Screen Negative)” or “Follow-up required”. In the statistical summary analysis, we included summary data for all methods.

For GALC, five laboratories reported using flow injection analysis MS/MS (FIA-MS/MS), non-kit and one used a flu-

orometric method. For GAA and IDUA, five laboratories reported using FIA-MS/MS, non-kit; one used LC-MS/MS and one reported using digital microfluidics.

The GALC, GAA, and IDUA expected values were based on CDC assayed values by FIA-MS/MS. The frequency distribution of participants' interpretations for categorical results is shown in Tables 1a–c. Specimen certification information is given in Table 2 and overall method statistics for GALC, GAA, and IDUA are given in Tables 3a–c.

The all-method mean cutoff for GALC was 0.6, with a range of 0.3 to 0.7  $\mu\text{mol/hr/L}$ ; the all-method mean cutoff for GAA was 3.3 with a range of 1.6 to 8.0  $\mu\text{mol/hr/L}$ ; and the all-method mean cutoff for IDUA was 2.0 with a range of 1.1 to 3.2  $\mu\text{mol/hr/L}$ .

No false negatives were reported for Krabbe (GALC), Pompe (GAA), or MPS-I (IDUA). One false positive assessment was reported for MPS-I for specimen 116L1. False-positive assessments should be monitored and kept as low as possible.

The Newborn Screening Quality Assurance Program will ship next Quarter's pilot PT specimens for Krabbe, Pompe, and/or and Mucopolysaccharidosis Type I disease on April 4, 2016.

## ACKNOWLEDGMENTS

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CDC/APHL

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**NEWBORN SCREENING QUALITY ASSURANCE PROGRAM**  
**LYSOSOMAL STORAGE DISORDERS TO DETECT KRABBE DISEASE, POMPE DISEASE**  
**AND MUCOPOLYSACCHARIDOSIS TYPE I**  
**IN DRIED BLOOD SPOTS**  
**QUARTER 1 – FEBRUARY 2016**

Table 1a. Frequency of reported Clinical Assessments:  
 RABBE DISEASE (GALC)

<b>Specimen Number</b>	<b>No follow-up required (Screen Negative)</b>	<b>Follow-up required</b>
116L1	0	6
116L2	6	0
116L3	6	0
116L4	6	0
116L5	6	0

Table 1b. Frequency of Reported Clinical Assessments:  
 POMPE DISEASE (GAA)

<b>Specimen Number</b>	<b>No follow-up required (Screen Negative)</b>	<b>Follow-up required</b>
116L1	7	0
116L2	7	0
116L3	7	0
116L4	0	7
116L5	7	0

NEWBORN SCREENING QUALITY ASSURANCE PROGRAM  
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QUARTER 1 – FEBRUARY 2016

Table 1c. Frequency of Reported Clinical Assessments:  
MUCOPOLYSACCHARIDOSIS TYPE I (IDUA)

<b>Specimen Number</b>	<b>No follow-up required (Screen Negative)</b>	<b>Follow-up required</b>
116L1	6	1
116L2	7	0
116L3	0	7
116L4	7	0
116L5	7	0

NEWBORN SCREENING QUALITY ASSURANCE PROGRAM  
 LYSOSOMAL STORAGE DISORDERS TO DETECT KRABBE DISEASE, POMPE DISEASE  
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 QUARTER 1 – FEBRUARY 2016

Table 2. Specimen Certification

Specimen Number	Expected GALC ( $\mu\text{mol/hr/L}$ )	KRABBE Assessment Code
116L1	0.41	2
116L2	8.40	1
116L3	4.50	1
116L4	3.49	1
116L5	8.52	1
Specimen Number	Expected GAA ( $\mu\text{mol/hr/L}$ )	POMPE Assessment Code
116L1	28.94	1
116L2	25.00	1
116L3	39.65	1
116L4	0.11	2
116L5	12.34	1

1 = No follow-up required (Screen Negative)

2 = Follow-up required

NEWBORN SCREENING QUALITY ASSURANCE PROGRAM  
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 QUARTER 1 – FEBRUARY 2016

Table 2. Specimen Certification, cont.

Specimen Number	Expected IDUA ( $\mu\text{mol/hr/L}$ )	MPS-I Assessment Code
116L1	2.70	1
116L2	26.59	1
116L3	0.66	2
116L4	5.83	1
116L5	17.43	1

1 = No follow-up required (Screen Negative)

2 = Follow-up required

## OVERALL STATISTICS

Table 3a. Screening Results for GALC – ALL Methods

Specimen	N	Mean ( $\mu\text{mol/hr/L}$ )	SD	%CV
116L1	6	0.25	0.1	40.4
116L2	6	7.54	3.1	41.7
116L3	6	3.84	1.6	42.8
116L4	6	3.02	0.9	29.7
116L5	6	7.08	3.2	45.4

Table 3b. Screening Results for GAA – ALL Methods

Specimen	N	Mean ( $\mu\text{mol/hr/L}$ )	SD	%CV
116L1	7	26.52	14.2	53.5
116L2	7	20.34	13.2	64.7
116L3	7	33.39	18.9	56.5
116L4	7	0.56	0.7	126.5
116L5	7	10.06	6.2	62.0

Table 3c. Screening Results for IDUA - ALL Methods

Specimen	N	Mean ( $\mu\text{mol/hr/L}$ )	SD	%CV
116L1	7	2.23	0.9	39.6
116L2	7	21.01	9.3	44.3
116L3	7	0.94	1.0	104.7
116L4	7	4.94	1.0	20.5
116L5	7	12.19	5.3	43.2

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