

Newborn Screening Quality Assurance Program

LYSOSOMAL STORAGE DISORDER Quarterly Report

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May 2014

INTRODUCTION

This report is the Quarterly summary of all data reported within the specified data-reporting period for the Quarter 2, 2014, pilot proficiency testing (PT) program for Lysosomal Storage Disorders (LSD) in dried-blood spots (DBS) to detect Krabbe and/or Pompe disease. 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. We distribute this PT report to all participants, state laboratory directors, and program colleagues by request.

On April 7, 2014, a panel of five unknown DBS specimens was distributed to eight laboratories in the United States to analyze Galactocerebrosidase (GALC) for Krabbe disease and/or Acid Alpha-Glucosidase (GAA) for Pompe disease in whole blood.

PARTICIPANT RESULTS

This panel consisted of five DBS specimens 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 214L1, 214L2, 214L3, 214L4, and 214L5).

We processed data from seven participants. Laboratories were asked to report quantitative results for GALC and/or GAA 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 did not include data that were outside the 99% confidence interval, nor did we include summary data for methods reported by

less than two laboratories. Two laboratories that normally report data for both enzymes did not report GAA for this quarter.

For GALC, five laboratories reported using flow injection analysis MS/MS (FIA-MS/MS), and one used LC-MS/MS. For GAA, three laboratories reported using FIA-MS/MS, one used LC-MS/MS. One laboratory used digital microfluidic technology and therefore was not included in the statistical evaluation below.

The expected GALC and GAA 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-b. Specimen certification information is given in Table 2 and overall statistics for GALC and GAA are given in Tables 3a-b.

The mean cutoff for the GALC methods ($n>2$) was 0.50 with a range of 0.30 to 0.70 $\mu\text{mol/hr/L}$; the mean cutoff for the GAA methods ($n>2$) was 3.03 with a range of 2.10 to 4.00 $\mu\text{mol/hr/L}$.

No false positive and no false negative assessments were reported for Krabbe (GALC) and no false positive and no false negative assessments were reported for Pompe (GAA).

The Newborn Screening Quality Assurance Program will ship next Quarter's pilot PT specimens for Krabbe and/or Pompe disease on July 14, 2014. ❖

ACKNOWLEDGMENTS

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CDC/APHL

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NEWBORN SCREENING QUALITY ASSURANCE PROGRAM
 LYSOSOMAL STORAGE DISORDERS TO DETECT KRABBE AND/OR POMPE DISEASE
 IN DRIED-BLOOD SPOTS
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Table 1a. Frequency of reported Clinical Assessments: KRABBE DISEASE (GALC)

Specimen Number	No follow-up required (Screen Negative)	Follow-up required
214L1	6	0
214L2	6	0
214L3	0	6
214L4	6	0
214L5	6	0

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

Specimen Number	No follow-up required (Screen Negative)	Follow-up required
214L1	5	0
214L2	5	0
214L3	5	0
214L4	5	0
214L5	0	5

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Table 2. Specimen Certification

Specimen Number	Expected GALC ($\mu\text{mol/hr/L}$)	KRABBE Assessment Code
214L1	8.70	1
214L2	3.55	1
214L3	0.29	2
214L4	5.61	1
214L5	2.51	1
Specimen Number	Expected GAA ($\mu\text{mol/hr/L}$)	POMPE Assessment Code
214L1	22.59	1
214L2	16.75	1
214L3	30.77	1
214L4	18.77	1
214L5	0.02	2

1 = No Follow-Up Required (Screen Negative)
 2 = Follow-Up Required

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OVERALL STATISTICS

Table 3a. Screening Results for GALC – Mass Spectrometry Methods‡

Specimen	N	Outliers*	Mean (µmol/hr/L)	UL (95%)	LL (95%)
214L1	5	0	5.85	8.64	3.07
214L2	5	0	3.33	4.53	2.14
214L3	5	0	0.18	0.29	0.07
214L4	5	0	4.53	6.23	2.83
214L5	5	0	2.26	3.01	1.51

‡ Data from methods where N<2 are not included.

* Outliers are not included in N

UL = upper limit LL = lower limit

Table 3b. Screening Results for GAA – Mass Spectrometry Methods‡

Specimen	N	Outliers*	Mean (µmol/hr/L)	UL (95%)	LL (95%)
214L1	3	0	21.39	30.30	12.49
214L2	3	0	14.15	22.20	6.10
214L3	3	0	33.18	41.59	24.77
214L4	3	0	19.96	26.44	13.48
214L5	3	0	0.54	2.01	0.00

‡ Data from methods where N<2 are not included.

* Outliers are not included in N

UL = upper limit LL = lower limit

This **NEWBORN SCREENING QUALITY ASSURANCE PROGRAM** report is an internal publication distributed to program participants and selected program colleagues. The laboratory quality assurance program is a project cosponsored by the **Centers for Disease Control and Prevention (CDC)** and the **Association of Public Health Laboratories**.

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