

Newborn Screening Quality Assurance Program

PROFICIENCY TESTING

Lysosomal Storage Disorder
Quarterly Report

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INTRODUCTION

This report is the Quarterly summary of all data reported within the specified data-reporting period for the Quarter 3, 2015, 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.

On July 13, 2015, 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 315L1, 315L2, 315L3, 315L4, and 315L5).

We processed data from eight 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 summary data for methods reported by less than two laboratories.

For GALC six laboratories reported using flow injection analysis MS/MS (FIA-MS/MS), non-kit; one used LC-MS/MS; and one used digital microfluidic technology. For GAA six laboratories reported using FIA-MS/MS, non-kit; one used LC-MS/MS and one used digital microfluidic technology.

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 MS/MS overall statistics for GALC and GAA are given in Tables 3a and 3b.

The all-method mean cutoff for the GALC methods was 0.41, with a range of 0.1 to 0.60 $\mu\text{mol/hr/L}$; the all-method mean cutoff for the GAA methods was 2.79 with a range of 0.8 to 6.50 $\mu\text{mol/hr/L}$.

No false negatives were reported for Krabbe (GALC) or Pompe (GAA). Two false positive assessments were reported for Pompe (GAA) only. 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 and/or Pompe disease on October 5, 2015.

ACKNOWLEDGMENTS

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CDC/APHL

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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
315L1	0	8
315L2	8	0
315L3	8	0
315L4	8	0
315L5	8	0

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

Specimen Number	No follow-up required (Screen Negative)	Follow-up required
315L1	8	0
315L2	6	2
315L3	0	8
315L4	8	0
315L5	8	0

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Table 2. Specimen Certification

Specimen Number	Expected GALC ($\mu\text{mol/hr/L}$)	KRABBE Assessment Code
315L1	0.23	2
315L2	2.79	1
315L3	3.53	1
315L4	5.31	1
315L5	10.53	1
Specimen Number	Expected GAA ($\mu\text{mol/hr/L}$)	POMPE Assessment Code
315L1	29.09	1
315L2	4.64	1
315L3	0.06	2
315L4	15.34	1
315L5	37.74	1

1 = No follow-up required (Screen Negative) 2 = Follow-up required

OVERALL STATISTICS

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

Specimen	N	Mean ($\mu\text{mol/hr/L}$)	SD	%CV
315L1	7	0.22	0.04	19.9
315L2	7	2.89	0.86	29.6
315L3	7	3.07	0.44	14.3
315L4	7	4.76	1.46	30.7
315L5	7	10.56	2.73	25.8

* Data from methods where N<2 are not included.
UL = upper limit LL = lower limit

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

Specimen	N	Mean ($\mu\text{mol/hr/L}$)	SD	%CV
315L1	7	27.18	8.03	29.6
315L2	7	4.13	1.96	47.6
315L3	7	0.21	0.21	98.3
315L4	7	12.58	5.18	41.1
315L5	7	29.92	13.67	45.7

‡ Data from methods where N<2 are not included.
UL = upper limit LL = lower limit

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