

# Newborn Screening Quality Assurance Program

PROFICIENCY TESTING

LYSOSOMAL STORAGE DISORDER  
Quarterly Report

Volume 2, No. 1

February 2013

## INTRODUCTION

This report is the Quarterly summary of all data reported within the specified data-reporting period for the Quarter 1, 2013, 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 January 7, 2013, a panel of five unknown DBS specimens was distributed to six 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 113L1, 113L2, 113L3, 113L4, and 113L5).

We processed data from six 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.

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

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

The mean cutoff for the GALC methods was 0.413, with a range from 0.200 to 0.500  $\mu\text{mol/hr/L}$ ; the mean cutoff for the GAA methods was 3.732 with a range of 1.6 to 8.0  $\mu\text{mol/hr/L}$ .

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

The Newborn Screening Quality Assurance Program will ship next Quarter's pilot PT specimens for Krabbe and/or Pompe disease on April 1, 2013. ❖

## ACKNOWLEDGMENTS

We would like to thank Barbara Waters-Pick (Duke University Medical Center) for the supply of umbilical cord blood units. ❖

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 1. Specimen Certification

Specimen Number	Expected GALC ( $\mu\text{mol/hr/L}$ )	KRABBE Assessment Code
113L1	5.77	1
113L2	0.17	2
113L3	3.27	1
113L4	2.49	1
113L5	7.53	1
Specimen Number	Expected GAA ( $\mu\text{mol/hr/L}$ )	POMPE Assessment Code
113L1	19.25	1
113L2	32.76	1
113L3	17.24	1
113L4	0.23	2
113L5	24.83	1

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

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Table 2a. Frequency of reported Clinical Assessments: KRABBE DISEASE (GALC)

<b>Specimen Number</b>	<b>No follow-up required (Screen Negative)</b>	<b>Follow-up required</b>
113L1	5	0
113L2	0	5
113L3	5	0
113L4	5	0
113L5	5	0

Table 2b. 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>
113L1	5	0
113L2	5	0
113L3	5	0
113L4	0	5
113L5	5	0

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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%)
113L1	5	0	4.064	1.183	6.945
113L2	5	0	0.224	0.085	0.363
113L3	5	0	2.420	0.479	4.361
113L4	5	0	1.664	0.161	3.167
113L5	5	0	5.458	1.582	9.334

‡ 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 Methods‡

Specimen	N	Outliers*	Mean (µmol/hr/L)	UL (95%)	LL (95%)
113L1	4	1	25.868	15.743	35.992
113L2	4	1	45.588	23.147	68.028
113L3	4	1	21.268	14.961	27.574
113L4	4	1	1.015	0.000	3.327
113L5	4	1	30.140	18.472	41.808

‡ Summary of results from all methods reported

\* 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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