Newborn Screening Quality Assurance Program

Quarterly Report
Volume 6, No.3

Lysosomal Storage Disorders Proficiency Testing Program (LSDPT)

Issued: November 8, 2017

Introduction

This report is the Quarterly summary of data reported within the specified data-reporting period for the Quarter 4, 2017, 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-1). It is distributed to all participants, state laboratory directors, and program colleagues by request. The tables within this report provide certification profiles for the distributed specimens, and a summary of reported analytical and categorical results. An evaluation of your laboratory's data is attached to this summary.

Certification of PT Specimens

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 417L1, 417L2, 417L3, 417L4, and 417L5). Table 1 shows the expected specimen values and clinical assessments for Galactocerebrosidase (GALC) for Krabbe disease, Acid Alpha-Glucosidase (GAA) for Pompe disease, and alpha-L-iduronidase (IDUA) for Mucopolysaccharidosis Type I in whole blood. The expected values were based on NSQAP assayed values by FIA-MS/MS.

Table 1. Expected Values – GALC, GAA and IDUA (μmol/hr/L)

Specimen	Expected GALC	Krabbe Assessment Code*	Expected GAA	Pompe Assessment Code*	Expected IDUA	MPS-1 Assessment Code*
417L1	5.44	1	19.37	1	0.13	2
417L2	0.21	2	17.04	1	1.50	3
417L3	11.09	1	10.57	1	15.16	1
417L4	10.95	1	6.26	1	7.95	1
417L5	10.55	1	10.61	1	15.08	1

*1 = No follow-up required (Screen Negative)
2 = Follow-up required (Screen Positive)
3 = Borderline

Distribution of PT Specimens

On October 2, 2017 a PT panel of five unknown DBS specimens was distributed to 16 domestic laboratories.

Participant Results

- Quantitiative Data

We processed data from 14 participants. Laboratories were asked to report quantitative results for GALC, GAA, and IDUA in µmol/hr/L. For GALC, one laboratory reported using LC-MS/MS, seven used an FIA-MS/MS non-kit multiplexed enzyme reaction, and one used a fluorometric method. For GAA, three laboratories reported using LC-MS/MS, eight used an FIA-MS/MS non-kit multiplexed enzyme reaction, and three reported using digital microfluidics. For IDUA, three laboratories reported using LC-MS/MS, seven reported using FIA-MS/MS non-kit multiplexed enzyme reaction, and three reported using digital microfluidics. The statistical summary analysis and cutoff information for all methods is provided in Table 2.

Table 2. Screening Results for GALC, GAA and IDUA —All methods

Analyte	Specimen	N	Mean (µmol/hr/L)	SD	Mean Reported Cutoffs	Range of Reported Cutoffs
GALC	417L1	9	3.95	1.41	0.73	0.44—1.45
	417L2	9	0.33	0.22		
	417L3	9	8.77	2.82		
	417L4	9	9.66	3.61		
	417L5	9	8.42	2.77		
GAA	417L1	14	24.36	15.70	2.81	0.73—10.0
	417L2	14	22.51	12.29		
	417L3	14	16.02	12.17		
	417L4	14	8.40	5.53		
	417L5	14	15.74	11.48		
IDUA	417L1	13	0.37	0.74	1.78	0.86—4.0
	417L2	13	1.92	0.72		
	417L3	13	19.10	9.81		
	417L4	13	10.46	5.80		
	417L5	13	19.28	9.74		

- Clinical Assessments

Laboratories were asked to report qualitative results as “No follow-up required (Screen Negative)” or “Follow-up required (Screen Positive)”. A “Borderline” assessment category is included to more accurately assess those labs that identify milder disease forms, carriers, or pseudo deficiencies. The frequency distribution of participants’ clinical assessments is shown in Table 3.

Table 3. Frequency Distribution of reported Clinical Assessments

Analyte	Specimen	No follow-up required (Screen Negative)	Follow-up required (Screen Positive)	Borderline
GALC	417L1	9	0	0
	417L2	0	9	0
	417L3	9	0	0
	417L4	9	0	0
	417L5	9	0	0
GAA	417L1	14	0	0
	417L2	14	0	0
	417L3	14	0	0
	417L4	14	0	0
	417L5	14	0	0
IDUA	417L1	0	13	0
	417L2*	3	4	6
	417L3	13	0	0
	417L4	13	0	0
	417L5	13	0	0

*Specimen 417L2 was not evaluated for IDUA.

Evaluations

Participants reported no False-negatives or False-positives for Krabbe, Pompe or MPS-1. No misclassifications were reported for IDUA specimen 417L2 due to the wide variety of reported grading schemes.

Future Shipments

The Newborn Screening Quality Assurance Program will ship next quarter's LSDPT specimens in January 2018.

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

The content of this report may also be located on our website at:
http://www.cdc.gov/labstandards/nsqap_reports.html

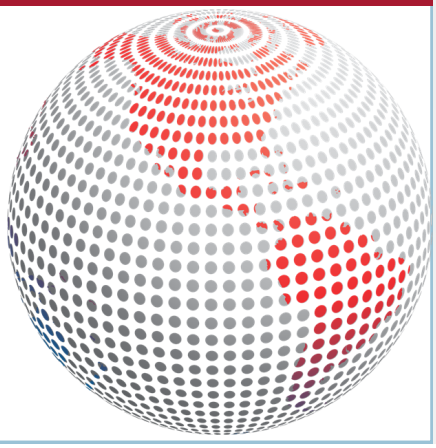
This program is co-sponsored by the Centers for Disease Control and Prevention (CDC) and The Association of Public Health Laboratories (APHL)

NEWBORN SCREENING QUALITY ASSURANCE PROGRAM

Direct inquiries to:

Centers for Disease Control and Prevention
4770 Buford Highway NE, MS/F19
Atlanta, GA 30341-3724
Phone: 404-488-7945 Email: jvm0@cdc.gov

Editors
Joanne Mei
Irene Williams



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.

INQUIRIES TO:

Irene Williams, Editor • Centers for Disease Control and Prevention (CDC) • Newborn Screening Quality Assurance Program
Mailstop F-24 • 4770 Buford Highway, N.E. • Atlanta, GA 30341-3724
Phone (770) 488-4582 • NSQAPDMT@cdc.gov
E-mail: IWilliams1@cdc.gov