Newborn Screening Quality Assurance Program

Quarterly Report
Volume 7, No.1

Lysosomal Storage Disorders Proficiency Testing Program (LSDPT)

Issued: February 28, 2018

Introduction

This report is the Quarterly summary of data reported within the specified data-reporting period for the Quarter 1, 2018, 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 118L1, 118L2, 118L3, 118L4, and 118L5). 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*
118L1	7.12	1	6.38	1	6.72	1
118L2	4.34	1	5.48	1	16.21	1
118L3	5.68	1	18.36	1	0.13	2
118L4	4.60	1	10.49	1	8.66	1
118L5	4.25	1	0.19	2	4.44	1

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

Distribution of PT Specimens

On January 9, 2018, a PT panel of five unknown DBS specimens was distributed to 17 domestic laboratories.

Participant Results

- Quantitiative Data

We processed data from 15 participants. Laboratories were asked to report quantitative results for GALC, GAA, and IDUA in µmol/hr/L. For GALC, two laboratories reported using LC-MS/MS, seven used an FIA-MS/MS non-kit multiplexed enzyme reaction, and one used a fluorometric method. For GAA, two laboratories reported using LC-MS/MS, seven used an FIA-MS/MS non-kit multiplexed enzyme reaction, two reported using digital microfluidics, and one used a fluorometric method. For IDUA, two laboratories reported using LC-MS/MS, six reported using FIA-MS/MS non-kit multiplexed enzyme reaction, and two reported using digital microfluidics, and one used a fluorometric method. 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	118L1	10	4.98	2.21	0.68	0.15—1.50
	118L2	10	2.92	1.39		
	118L3	10	3.62	1.61		
	118L4	10	3.09	1.38		
	118L5	10	3.35	1.27		
GAA	118L1	15	10.17	7.40	2.92	0.95—9.50
	118L2	15	9.44	7.26		
	118L3	15	24.74	15.49		
	118L4	15	14.94	10.83		
	118L5	15	0.34	0.49		
IDUA	118L1	13	9.99	5.94	1.92	0.70—4.89
	118L2	13	23.02	14.41		
	118L3	13	0.17	0.12		
	118L4	13	12.26	8.16		
	118L5	13	5.06	1.59		

- 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	118L1	10	0	0
	118L2	10	0	0
	118L3	10	0	0
	118L4	10	0	0
	118L5	10	0	0
GAA	118L1	15	0	0
	118L2	15	0	0
	118L3	15	0	0
	118L4	15	0	0
	118L5	0	15	0
IDUA	118L1	13	0	0
	118L2	13	0	0
	118L3	0	13	0
	118L4	13	0	0
	118L5	12	0	1

Evaluations

Participants reported no False-negatives and no False-positives for Krabbe, Pompe or MPS-1.

Future Shipments

The Newborn Screening Quality Assurance Program will ship next quarter's LSDPT specimens on July 10, 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)

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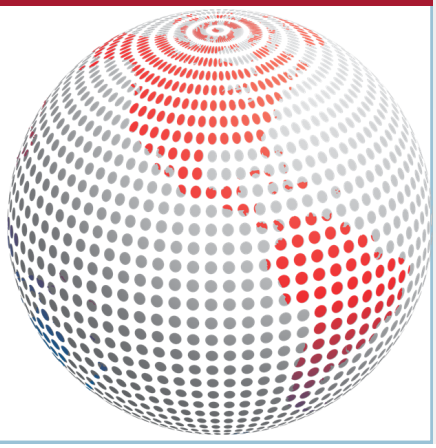
Direct inquiries to:

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