Newborn Screening Quality Assurance Program

Quarterly Report
Volume 6, No.2

Lysosomal Storage Disorders Proficiency Testing Program (LSDPT)

2017 Quarter 3 August

Introduction

This report is the Quarterly summary of data reported within the specified data-reporting period for the Quarter 3, 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 317L1, 317L2, 317L3, 317L4, and 317L5). 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*
317L1	10.95	1	6.24	1	8.79	1
317L2	11.19	1	6.48	1	8.97	1
317L3	0.25	2	17.93	1	1.82	3
317L4	4.30	1	0.35	2	5.08	1
317L5	10.90	1	10.99	1	15.96	1

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

Distribution of PT Specimens

On July 10, 2016 a PT panel of five unknown DBS specimens was distributed to 14 domestic laboratories.

Participant Results

- Quantitiative Data

We processed data from 11 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, six used an FIA-MS/MS non-kit multiplexed enzyme reaction, and one used a fluorometric method. For GAA, one laboratory reported using LC-MS/MS, five used an FIA-MS/MS non-kit multiplexed enzyme reaction, and one reported using digital microfluidics. For IDUA, one laboratory reported using LC-MS/MS, five reported using FIA-MS/MS non-kit multiplexed enzyme reaction, and one 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	317L1	9	9.23	3.3	0.73	0.44—1.45
	317L2	9	9.22	3.4		
	317L3	9	0.89	2.0		
	317L4	9	3.80	1.1		
	317L5	9	8.76	3.4		
GAA	317L1	10	7.90	5.0	2.81	0.73—10.0
	317L2	10	7.86	5.0		
	317L3	10	19.78	11.3		
	317L4	10	1.04	1.5		
	317L5	10	13.51	10.3		
IDUA	317L1	10	8.50	4.6	1.78	0.86—4.0
	317L2	10	8.47	4.5		
	317L3	10	1.60	0.5		
	317L4	10	4.35	1.7		
	317L5	10	15.47	7.7		

- 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	317L1	9	0	
	317L2	9	0	
	317L3	1	8	
	317L4	9	0	
	317L5	9	0	
GAA	317L1	10	0	
	317L2	10	0	
	317L3	10	0	
	317L4	1	9	
	317L5	10	0	
IDUA	317L1	10	0	
	317L2	10	0	
	317L3	3	4	3
	317L4	10	0	
	317L5	10	0	

Evaluations

Participants reported one False-negative for Krabbe and one False-negative for Pompe. No False-positive assessments for Krabbe, Pompe or MPS-1 were reported. No misclassifications were reported for IDUA specimen 317L3 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 October 2, 2017.

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

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NEWBORN SCREENING QUALITY ASSURANCE PROGRAM

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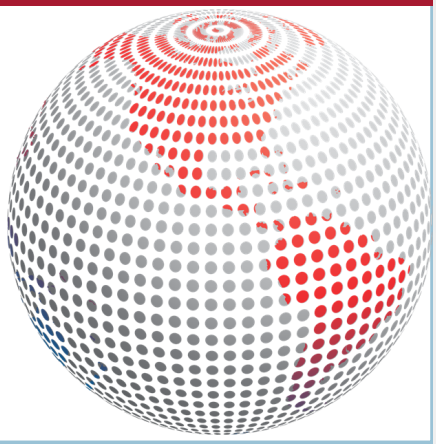

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