Newborn Screening Quality Assurance Program

Quarterly Report
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Lysosomal Storage Disorders Proficiency Testing Program (LSDPT)

2017 Quarter 1 February

Introduction

This report is the Quarterly summary of data reported within the specified data-reporting period for the Quarter 1, 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 117L1, 117L2, 117L3, 117L4, and 117L5). 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 MPS-I in whole blood. The expected values were based on NSQAP assayed values by FIA-MS/MS.

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

Specimen	Expected GALC	Krabbe Assessment Code*	Expected GAA	Pompe Assessment Code	Expected IDUA	MPS-1 Assessment Code
117L1	7.03	1	27.57	1	24.31	1
117L2	4.01	1	1.04	2	5.01	1
117L3	4.39	1	25.14	1	16.72	1
117L4	7.56	1	13.09	1	12.71	1
117L5	4.42	1	44.11	1	1.04	2

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

Distribution of PT Specimens

On January 11, 2016 a PT panel of five unknown DBS specimens was distributed to 12 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, five 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, four 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, one reported an FIA-MS/MS non-kit individual enzyme reaction, four 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	117L1	8	7.76	2.99	0.6	0.4 - 0.7
	117L2	8	3.53	1.44		
	117L3	8	8.08	2.54		
	117L4	8	8.47	2.99		
	117L5	8	4.65	1.41		
GAA	117L1	9	14.10	12.4	2.9	0.9 - 12.0
	117L2	9	0.59	0.9		
	117L3	9	14.36	12.4		
	117L4	9	7.82	5.6		
	117L5	9	24.90	18.5		
IDUA	117L1	10	19.46	11.50	2.1	0.7 - 6.0
	117L2	10	5.43	2.15		
	117L3	10	17.97	11.01		
	117L4	10	9.60	4.95		
	117L5	10	0.36	0.58		

♦ 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	117L1	8	0	0
	117L2	8	0	0
	117L3	8	0	0
	117L4	8	0	0
	117L5	8	0	0
GAA	117L1	9	0	0
	117L2	0	9	0
	117L3	9	0	0
	117L4	9	0	0
	117L5	9	0	0
IDUA	117L1	10	0	0
	117L2	10	0	0
	117L3	10	0	0
	117L4	10	0	0
	117L5	0	10	0

Evaluations

Participants reported no False-positive assessments and no False-negative assessments for Krabbe, Pompe or MPS-1.

Future Shipments

The Newborn Screening Quality Assurance Program will ship next quarter's LSDPT specimens in July 10, 2017.

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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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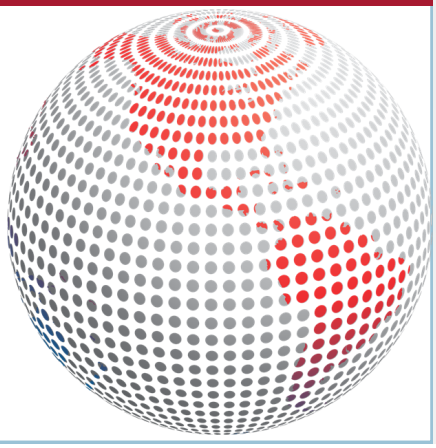

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