

Lysosomal Storage Disorders (LSD) Proficiency Testing Program (PT)

2016 Quarter 4 November

Introduction

This report summarizes data submitted within the specified data-reporting period for the Quarter 4 2016 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 416L1, 416L2, 416L3, 416L4, and 416L5). 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 ($\mu\text{mol/hr/L}$)

Specimen	Expected GALC	Krabbe Assessment Code*	Expected GAA	Pompe Assessment Code	Expected IDUA	MPS-1 Assessment Code
416L1	3.17	1	0.16	2	5.07	1
416L2	0.24	2	35.59	1	2.65	1
416L3	6.59	1	31.47	1	31.00	1
416L4	3.47	1	48.52	1	0.39	2
416L5	7.94	1	14.69	1	15.95	1

1 = No follow-up required (Screen Negative)

2 = Follow-up required (Screen Positive)

3 = Borderline

Distribution of PT Specimens

On October 3, 2016 a PT panel of five unknown DBS specimens was distributed to nine domestic laboratories.

Participant Results

◆ Quantitative Data

We processed data from seven participants. Laboratories were asked to report quantitative results for GALC, GAA, and IDUA in $\mu\text{mol/hr/L}$. For GALC, five laboratories reported using flow injection analysis MS/MS (FIA-MS/MS) non-kit, and one used a fluorometric method. For GAA, five laboratories reported using FIA-MS/MS, non-kit; one used LC-MS/MS and one reported using digital microfluidics. For IDUA, five laboratories reported using FIA-MS/MS, non-kit; one used LC-MS/MS 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 ($\mu\text{mol/hr/L}$)	SD	Mean Reported Cutoff	Range of Reported Cutoff
GALC	416L1	7	3.22	0.9	0.6	0.4 - 0.7
	416L2	7	0.28	0.1		
	416L3	7	7.17	3.0		
	416L4	7	4.29	2.0		
	416L5	7	8.20	3.4		
GAA	416L1	8	0.68	0.9	2.8	1.0 - 9.6
	416L2	8	21.63	12.4		
	416L3	8	16.89	14.7		
	416L4	8	26.48	17.3		
	416L5	8	8.71	6.1		
IDUA	416L1	8	3.83	1.3	1.5	0.7 - 3.0
	416L2	8	1.67	0.4		
	416L3	8	19.02	9.7		
	416L4	8	0.31	0.3		
	416L5	8	10.67	5.4		

◆ 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	416L1	7	0	0
	416L2	1	6	0
	416L3	7	0	0
	416L4	7	0	0
	416L5	7	0	0
GAA	416L1	0	8	0
	416L2	8	0	0
	416L3	8	0	0
	416L4	8	0	0
	416L5	8	0	0
IDUA	416L1	8	0	0
	416L2	4	1	3
	416L3	8	0	0
	416L4	0	8	0
	416L5	8	0	0

Evaluations

Participants reported one False-positive assessment for MPS-1 and one False-negative assessment for Krabbe. Three laboratories reported assessments classified as Borderline for MPS-1 which was considered acceptable based on their laboratory grading algorithm.

Future Shipments

The Newborn Screening Quality Assurance Program will ship next quarter's LSDPT specimens on January 9, 2017.

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

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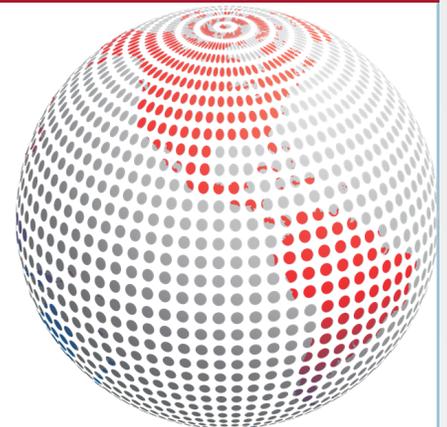
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