Newborn Screening Quality Assurance Program X-linked Adrenoleukodystrophy in Dried Blood Spots Proficiency Testing Program (XALDPT)

In co-sponsorship with Association of Public Health Laboratories (APHL)
Provided by the Newborn Screening and Molecular Biology Branch
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Report Authorization

This report has been reviewed and authorized by Dr. Joanne Mei, Laboratory Chief, Newborn Screening Quality Assurance Program.

Confidentiality Statement

NSQAP participant information and evaluations are strictly confidential and shared only with individual participants, unless written authorization for release is received.

Introduction

This report summarizes data collected within the specified period for Quarter 3, 2019, for the detection of X-ALD by analysis of the biomarkers 24:0-Lysophosphatidylcholine (24LPC) and 26:0-Lysophosphatidylcholine (26LPC) in dried blood spots (DBS). 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, statistical analysis of participant quantitative data, and frequency of clinical assessments. An evaluation of your laboratory's data is attached to this summary.

Certification of PT Specimens

DBS specimens were prepared from Type A+ human whole blood, which was adjusted to a hematocrit of 50 ± 1% and enriched with the biomarkers 24LPC and 26LPC. Expected values for each were determined by LC-MS/MS in units of µmol/L blood. Clinical assessments were based on the NSQAP cut-off of 0.47 µmol/L blood for 24LPC and 0.39 µmol/L blood for 26LPC. Table 1 shows the NSQAP expected values and clinical assessments for each specimen.

Table 1. Expected Values – 24LPC and 26LPC (µmol/L blood)

Specimen	Expected 24LPC	24LPC Assessment Code*	Expected 26LPC	26LPC Assessment Code*
31921	0.20	1	0.05	1
31922	1.10	2	0.95	2
31923	0.20	1	0.05	1
31924	0.20	1	0.05	1
31925	0.20	1	0.05	1

*1 = Within Normal Limits
2 = Outside Normal Limits

Distribution of PT Specimens

On June 25, 2019 a PT panel of five unknown DBS specimens was distributed to 17 domestic laboratories and 5 foreign laboratories.

Participant Results

Quantitative Data

We processed data from 18 participants, with one participant submitting two method assessments. Laboratories were asked to report concentrations of 24LPC and 26LPC results in µmol/L blood. Data not submitted in the requested units were not accepted. The conversion factor from µg/mL to µmol/L blood is provided on the XALDPT Data Report Form.

Overall statistics from MS/MS methods were combined so as to not identify an individual laboratory. We also did not include data that were outside the 99% confidence interval. The statistical summary analysis for all methods is provided in Tables 2a-b.

Seven participants reported using Flow Injection Analysis (FIA) MS/MS non-kit, twelve reported using LC-MS/MS and one reported a two-tier assessment scheme utilizing both FIA– and LC-MS/MS. Fifteen participants submitted quantitative results for 24LPC, and nine did not report a clinical assessment. Nineteen participants reported quantitative results and clinical assessments for 26LPC. One participant reported cutoffs for 24LPC using multi-variate analysis by a post-analytic tool and 2nd tier testing when indicated. Tables 3a and 3b show the cutoffs reported for 24LPC and 26LPC sorted by method. The frequency distribution of clinical assessments are shown in Tables 4a-b.

Table 2a. Screening Results for 24LPC– All MS/MS Methods

Analyte/Specimen	N	Mean (µmol/L)	SD
31921	15	0.18	0.07
31922	15	0.85	0.29
31923	15	0.18	0.07
31924	15	0.19	0.08
31925	15	0.19	0.09

Table 2b. Screening Results for 26LPC – All MS/MS Methods

Analyte/Specimen	N	Mean (µmol/L)	SD
31921	19	0.14	0.16
31922	19	1.08	0.29
31923	19	0.14	0.17
31924	19	0.14	0.15
31925	19	0.14	0.16

Table 3a. Analyte Cutoffs Sorted by Method (µmol/L)

Method 53 - LC-MS/MS	24LPC	26LPC
N	6	12
Mean	0.45	0.28
Max	0.85	0.47
Min	0.14	0.12
Median	0.43	0.25
Mode	N/A	0.40

Table 3b. Analyte Cutoffs Sorted by Method (µmol/L)

Method 67 - FIA-MS/MS	24LPC	26LPC
N	3	6
Mean	0.41	0.44
Max	0.50	0.50
Min	0.33	0.36
Median	0.40	0.45
Mode	NA	NA

Clinical Assessments

Laboratories were asked to report qualitative results as “Within Normal Limits” or “Outside Normal Limits”. Qualitative assessments may differ because of specific assessment practices. The frequency distribution of participants’ clinical assessments is shown in Tables 4a-b.

Table 4a. Frequency Distribution of Clinical Assessments for 24LPC

Specimen	Within Normal Limits (WNL)	Borderline	Outside Normal Limits (ONL)
31921	10	0	0
31922	0	0	10
31923	10	0	0
31924	10	0	0
31925	9	1	0

Table 4b. Frequency Distribution of Reported Clinical Assessments for 26LPC

Specimen	Within Normal Limits (WNL)	Borderline	Outside Normal Limits (ONL)
31921	19	0	0
31922	0	0	19
31923	19	0	0
31924	19	0	0
31925	19	0	0

Evaluations

No misclassifications were reported for 24LPC or 26LPC.

Future Shipments

The Newborn Screening Quality Assurance Program will ship next quarter's PT specimens on September 24, 2019.

Direct Inquiries

If you have any comments or questions about XALDPT MS/MS analysis, contact Dr. Christopher A. Haynes at 770-488-7019 or by e-mail at cph7@cdc.gov

For data reporting questions, contact Irene Williams at nsqapdmt@cdc.gov

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

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.

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