Newborn Screening Quality Assurance Program T-Cell Receptor Circle in Dried Blood Spots Proficiency Testing Program (TRECPT)

In co-sponsorship with Association of Public Health Laboratories (APHL)
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Report Authorization

This report has been reviewed and authorized by Dr. Suzanne Cordovado, Laboratory Chief, Molecular Quality Improvement 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 all results submitted within the data-reporting period for the Quarter 4, 2019, proficiency testing (PT) program for T-cell receptor excision circle (TREC) analysis in dried blood spots (DBS) to detect severe combined immunodeficiency (SCID). The report is distributed to all participants, state laboratory directors, and program colleagues by request. The contents provide the certification profiles for the distributed specimens, screening methods, DNA extraction methods, reference genes used by participants, and the overall summary of reported clinical assessments. An evaluation of submitted data is attached to individual laboratory reports.

Certification of PT Specimens

This Quarter 4 panel consisted of five DBS specimens (specimens 419R1, 419R2, 419R3, 419R4, and 419R5) prepared from human blood, including umbilical cord blood from unaffected individuals and adult blood or modified adult blood depleted of mononuclear cells or leukocytes. Table 1 shows the certification and description of the specimens in the panel.

Table 1. Specimen Certification and Description

Specimen Number	Clinical Assessment*	Specimen Description
419R1	1	Normal sample; TREC and reference gene within acceptable range.
419R2	3	Unsatisifactory sample - both TREC and reference gene are out-of-range.
419R3	2	SCID-like sample with very low/undetectable TREC; reference gene within acceptable range.
419R4	1	Normal sample; TREC and reference gene within acceptable range.
419R5	2	SCID-like sample with very low/undetectable TREC; reference gene within acceptable range.

* Clinical Assessment Code Key:
1 – Screen Negative (no follow-up required)
2 – Screen Positive (TREC out-of-range, reference gene in-range)
3 – Unsatisfactory sample (both TREC and reference gene out-of-range)

Distribution of PT Specimens

On September 24, 2019, NSQAP distributed a panel of five unknown DBS specimens to 63 partcipants to analyze the TREC content in peripheral blood.

Participant Results

Data was received from 60 participants by the data reporting deadline. Participants tested specimens by the analytical schemes they routinely use in their laboratory. Reported data includes the laboratory method used to detect TREC levels, DNA extraction method, the reference gene and the clinical assessment.

Reported Method Data

Tables 2-5 summarize the reported frequency of methods used to assess TREC levels, DNA Extraction methods used, reference genes used, clinical assessments and misclassifications. Qualitative, categorical results of Screen Negative (no follow-up required), Screen Positive (TREC out-of-range, reference gene in-range), and Unsatisfactory sample (TREC and reference gene out-of-range) were requested for each specimen.

Table 2. Reported Laboratory Methods for TREC

Method	Number of Laboratories
Real Time PCR – TREC only (reference gene run separately)	8
Real Time PCR – TREC AND Reference Gene run in a single tube	21
Real Time PCR – TREC/SMN1 AND Reference Gene run in a single tube	9
EnLite™ Neonatal TREC kit	19
Digital PCR	1
Other	2

Table 3. Reported DNA Extraction Methods

Extraction Method	Number of Laboratories
In situ/on card (DNA is NOT extracted)	12
EnLite™ (DNA is NOT extracted)	19
Generations™ DNA Purification and Elution Solutions (S1/S2)	6
Generations™ Elution Solution (S2 only)	7
Extracta™ DBS with one wash	5
Other	10
No DNA Extraction Method Reported	1

Table 4. Reported Reference Genes

Reference Gene	Number of Laboratories
RNase P subunit (RPP30)	18
RNase P subunit (RPPH1)	5
Beta-actin (ACTB)	29
Other	5
No Reference Gene Reported	3

Table 5. Reported Clinical Assessments and Misclassifications

Specimen Number	1 - Screen Negative (no follow–up required)	2 - Screen Positive (TREC out-of-range reference gene in-range)	3 - Unsat Sample (TREC and reference gene out-of-range)	Clinical Assessment not Reported	Incorrect Clinical Assessments
419R1	59	0	0	1	0
419R2	1	0	59	0	1
419R3	0	57	3	0	3
419R4	58	1	0	1	1
419R5	0	59	1	0	1

Evaluations

Evaluations are based on the clinical assessment of the five specimens. Six incorrect clinical assessments were reported and two clinical assessments were not reported in this quarter.

Future Shipments

The Newborn Screening Quality Assurance Program will ship next quarter's PT specimens for TREC on January 14, 2020.

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

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