

Newborn Screening Quality Assurance Program

PROFICIENCY TESTING

TREC
Quarterly Report

Volume 4, No. 4

November 2014

INTRODUCTION

This report is the Quarterly summary of all data reported within the specified data-reporting period for the Quarter 4, 2014, proficiency testing (PT) program for T-cell receptor excision circle (TREC) analysis in dried blood spots (DBS) to detect severe combined immunodeficiency (SCID). The attached tables provide the certification profiles for the specimens, summary of reported categorical results and the verification of your reported data. We distribute this PT report to all participants, state laboratory directors, and program colleagues by request.

On October 6, 2014 a panel of five unknown DBS specimens was distributed to 23 domestic and 15 international laboratories to analyze TREC content in peripheral blood.

PARTICIPANT RESULTS

This panel consisted of five DBS specimens prepared from human blood, including cord blood from unaffected individuals and modified adult blood depleted of mononuclear cells or leukocytes (specimens 414R1, 414R2, 414R3, 414R4, and 414R5).

Evaluations are based on the source of specimen and previously established consensus categorical results from core laboratories.

All participants used laboratory-developed tests. We requested only qualitative, categorical results: 'No follow-up required (Screen Negative)' or 'Follow-up

required' for each specimen since quantitative results vary significantly between laboratories using different test methods and calibrators.

We processed data from 32 participants. Five false-positive assessments were reported for specimen 414R4. The TREC level for this specimen, while in the normal range, was close to the cutoff for determining if follow-up was required. No false-negative assessments were reported. False-positive assessments should be monitored and kept as low as possible.

The Newborn Screening Quality Assurance Program will ship next quarter's pilot PT specimens for TREC on January 12, 2015. ❖

ACKNOWLEDGMENTS

We would like to thank Barbara Waters-Pick (Duke University Medical Center) for the supply of umbilical cord blood.

CDC/APHL

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NEWBORN SCREENING QUALITY ASSURANCE PROGRAM

T-CELL RECEPTOR EXCISION CIRCLE (TREC) ANALYSIS IN DRIED-BLOOD SPOTS

Quarter 4 – NOVEMBER 2014

SPECIMEN CERTIFICATION

Specimen Number	No follow-up required (Screen Negative)	Follow-up required	Specimen Description
414R1	1		'Normal specimen; medium TREC level, reference gene level within standard reference range
414R2		2	SCID-like specimen; low or no TREC, reference gene level within standard reference range
414R3		2	Blood with 'buffy-coat' removed - TREC and reference gene levels both below standard reference range.
414R4	1		Normal specimen; TREC level close to cutoff, reference gene level within standard reference range
414R5	1		Normal specimen; below average TREC level, reference gene level within standard reference range

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

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FREQUENCY OF REPORTED CLINICAL ASSESSMENTS

Specimen Number	No follow-up required (Screen Negative)	Follow-up required
414R1	32	0
414R2	0	32
414R3	0	32
414R4	27	5
414R5	32	0

LABORATORY METHODS

Method	Number of Laboratories
63 Real Time PCR - Singleplex	11
71 Real Time PCR - Multiplex	16
70 EnLite™ Neonatal TREC kit	4
Other	1

FREQUENCY OF DNA PREPARATION METHODS

DNA Preparation Method	Number of Laboratories
1 In situ/on card (no DNA extraction) with washing step(s)	5
2 EnLite™ (no DNA extraction)	4
3 DNA extracted at 99°C with washing step(s)	15
4 DNA extracted at 95°C with washing step(s)	2
5 DNA extracted at 70°C with washing step(s)	2
6 DNA extracted with no washing step	0
7 Other	3
Not provided	1

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FREQUENCY OF REFERENCE GENE ASSESSMENT CATEGORY
(for expected Follow-up Required Specimens)

Specimen Number	1- Within normal range	2- Outside normal range
414R2	31	1
414R3	0	32

FREQUENCY OF REFERENCE GENES

Reference Genes	Number of Laboratories
1 RNase P coding segments	14
2 Beta-actin	17
3 Serum albumin	1
4 TERT - Telomerase Reverse	0
5 Other	0

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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