

Newborn Screening Quality Assurance Program

PROFICIENCY TESTING

Sickle Cell Disease and Other Hemoglobinopathies

Volume 24, No. 1

Panel 1

February 2014

INTRODUCTION

On January 13, 2014 we distributed five dried blood spot (DBS) specimens prepared from umbilical cord bloods to all active participants for the Panel 1 Sickle Cell Disease and Hemoglobinopathies Proficiency Testing (PT) event. A total of 73 panels were sent by overnight mail to 49 domestic laboratories and 24 foreign laboratories. This PT report is a compilation of data reports received from 71 of the participating laboratories by the designated deadline date. We distribute this PT report to all participants, state laboratory directors, and to program colleagues by request.

We requested that participants assay all survey specimens by the analytical schemes they routinely use and report for each specimen the presumptive phenotype, the presumptive clinical assessment, and any other clinical classifications that they deem consistent with their analytic results and program operations.

PARTICIPANTS' RESULTS

The certification report, listing hemoglobins (Hb) by phenotype and their presumptive clinical assessments, appears on page 2. The frequency distribution of reported presumptive phenotypes (Table 1a) and clinical assessments (Table 1b) appears on page 3. In Table 2, we provide the number of specimens reported per method by testing tier and number of sample errors. The testing tier corresponds to the level of confirmatory testing. To address the concern of a few participants regarding the appearance of Sample 114H3, in Table 3 we provide consensus data which support its integrity. Fifty eight of 71 laboratories were able to provide an acceptable phenotype and clinical assessment for Specimen 114H3. We also show the reported method frequency of the Not-Evaluated assessments. The individual data verification for each laboratory follows the acknowledgment page.

We will continue to ship three PT panels this year for Hemoglobinopathies. The next shipment of materials from the Sickle Cell and Hemoglobinopathies PT program will be on April 28, 2014. ❖

MEETINGS AND TRAINING

2014 Annual Sickle Cell Disease Research Meeting: April 11-14th, Miami, FL. For more information go to: <http://fscdr.org/>

2014 SCDA 42nd Annual Convention: October 1-4, Hyatt Regency Baltimore Inner Harbor, Baltimore, MD. For more information go to: <http://www.sicklecelldisease.org/>

CDC Web based Sickle Cell Resources - NEW Booklet: Download and share CDC's newest resource for teachers and caregivers on sickle cell disease (SCD): Tips for Supporting Students with Sickle Cell Disease. At http://www.cdc.gov/ncbddd/sicklecell/documents/tipsheet_supporting_students_with_scd.pdf

ACKNOWLEDGMENTS

The specimens for this survey were prepared from umbilical cord blood samples supplied by Cleveland Cord Blood Center, Cleveland, Ohio. They are an independent not-for-profit 501(c)3 organization that accepts donated cord blood for clinical use.

CDC/APHL

This program is cosponsored by the Centers for Disease Control and Prevention (CDC) and the Association of Public Health Laboratories (APHL).

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**Newborn Screening Quality Assurance Program
Hemoglobinopathies Proficiency Testing Program**

Specimen and Lab Certification

Year: 2014 Panel: 1

Presumptive Clinical Phenotypes

	Specimen 114H1	Specimen 114H2	Specimen 114H3	Specimen 114H4	Specimen 114H5
Expected Presumptive Phenotype	FA	FAC	FA Bart's	FA	FA
Accepted Presumptive Phenotypes	FA	FAC	FA FA Bart's	FA	FA

Presumptive Clinical Assessments

	Specimen 114H1	Specimen 114H2	Specimen 114H3	Specimen 114H4	Specimen 114H5
Expected Presumptive Clinical Assessment	01	03	16	01	01
Accepted Presumptive Clinical Assessments	01	03	01 16	01	01

NORMAL HEMOGLOBIN PATTERN

- 01 Normal - no abnormal Hb found
- 02 Hemoglobin S carrier
- 03 Hemoglobin C carrier
- 08 Hemoglobin D carrier
- 09 Hemoglobin E carrier

SICKLE CELL DISEASES

- 04 Hemoglobin SS disease (Sickle cell anemia)
- 05 Hemoglobin SC disease
- 06 Hemoglobin SD disease
- 12 Hemoglobin SE disease

OTHER REPORTABLE FINDINGS

- 16 Alpha thalassemia (Bart's Hb)
- 18 Hemoglobin E, E disease
- 19 Fast or aging bands (clinically insignificant)
- 20 Assessment not listed
- 21 Unsatisfactory sample
- 22 Unidentified variant carrier

LIST OF METHOD CODES

- | | |
|--|-----------------------------|
| 01 Electrophoresis - Cellulose Acetate | 10 Bio-Rad Screening HPLC |
| 02 Electrophoresis - Citrate Agar | 11 Extended Gradient HPLC |
| 04 Isoelectric focusing | 12 Other Methods |
| 07 Monoclonal antibody methods | 13 PCR amplification of DNA |
| 14 Primus Ultra ² HPLC | |

Newborn Screening Quality Assurance Program
Hemoglobinopathies Proficiency Testing Program

Panel 1 – February 2014

Total number of program participants = 73

Table 1a. Frequency distribution of participant reported presumptive clinical phenotypes

Specimen ID	Participant Reported Presumptive Clinical Phenotype	Frequency	#Correctly Classified	#Mis-Classified	#Non-Classified (no penalty)	#Data Not Reported
114H1	FA	71	71	0	0	2
114H2	FAC	69	69	0		2
	FA	1	0	1	0	
114H3	FAC	1	0	1		2
	FAE	1	0	1		
	FA	33	33	0	0	
	FA Bart's	25	25	0	0	
	Non-Evaluated	11	0	0	11	
114H4	FAC	1	0	1	0	2
	FAST	1	0	0	1	
	FA	70	70	0	0	
114H5	FA Bart's	1	0	1		2
	FA	71	71	0	0	

Table 1b. Frequency distribution of participant reported presumptive clinical assessments

Specimen ID	Participant Reported Presumptive Clinical Assessment	Frequency	#Correctly Classified	#Miss-Classified	#Non-Classified (no penalty)	#Data Not Reported
114H1	01 - Normal	71	71	0	0	2
114H2	03 - Hgb C carrier	69	69	0		2
	01 - Normal	1	0	1	0	
114H3	09 - Hgb E Carrier	1	0	1		2
	01 - Normal	33	33	0	0	
	16 - α -Thal Bart's	25	25	0	0	
	Non-Evaluated	11	0	0	11	
	03 - Hgb C carrier	1	0	1	0	
114H4	19 - Fast/aging bands	1	0	0	1	2
	01 - Normal	70	70	0	0	
114H5	16 - α -Thal Bart's	1	0	1		2
	01 - Normal	71	71	0	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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