

Newborn Screening Quality Assurance Program

PROFICIENCY TESTING

Sickle Cell Disease and Other Hemoglobinopathies

Volume 24, No. 2

Panel 2

June 2014

INTRODUCTION

On April 28, 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 70 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 number of additional methods that are used if a specimen is found to have abnormal hemoglobin(s). The individual data verification for each laboratory follows the acknowledgment page.

We included an educational specimen, EDU1, in this panel. It was prepared by mixing umbilical cord blood with normal hemoglobin (HbA) and blood from an anonymous adult EE donor to mimic an FAE newborn specimen. Of the 61 participants who reported results for this specimen, 57 reported a correct phenotype and 56 reported a correct clinical assessment (Tables 3a and 3b).

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 October 6, 2014. ❖

MEETINGS AND TRAINING

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 (UCB) samples supplied by Cleveland Cord Blood Center, Cleveland, Ohio and from residual excluded UCB units collected at Duke University Stem Cell Laboratory.

CDC/APHL

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

Specimen and Lab Certification

Year: 2014 Panel: 2

Presumptive Clinical Phenotypes

	Specimen 214H1	Specimen 214H2	Specimen 214H3	Specimen 214H4	Specimen 214H5
Expected Presumptive Phenotype	FA	FAS	FA	FAS	FA
Accepted Presumptive Phenotypes	FA	FAS	FA	FAS	FA

Presumptive Clinical Assessments

	Specimen 214H1	Specimen 214H2	Specimen 214H3	Specimen 214H4	Specimen 214H5
Expected Presumptive Clinical Assessment	01	02	01	02	01
Accepted Presumptive Clinical Assessments	01	02	01	02	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 | |
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Newborn Screening Quality Assurance Program
Hemoglobinopathies Proficiency Testing Program

Panel 2 – JUNE 2014

Total number of program participants = 70

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
214H1	FA	70	70	0	0	3
214H2	FAS	68	68	0	0	3
	FS	2	0	2	0	
214H3	FA	70	70	0	0	3
214H4	FAS	69	69	0	0	3
	FS	1	0	1	0	
214H5	FA	70	70	0	0	3

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

Specimen ID	Participant Reported Presumptive Clinical Assessment	Frequency	Correctly Classified	Mis-Classified	Non-Classified (no penalty)	Data Not Reported
214H1	01 - Normal	70	70	0	0	3
214H2	02 - Hgb S carrier	68	68	0	0	3
	04 - Hgb SS disease	2	0	2	0	
214H3	01 - Normal	70	70	0	0	3
214H4	02 - Hgb S carrier	68	68	0	0	3
	01 - Normal	1	0	1	0	
	04 - Hgb SS disease	1	0	1	0	
214H5	01 - Normal	70	70	0	0	3

Table 2. Number of samples reported per method by testing tier

	Method Code	Method	Samples Tested	Phenotype Errors	Assessment Errors
Primary Method	4	Isoelectric focusing	150	3	4
	7	Monoclonal Antibodies Method	1	0	0
	10	Bio-Rad Screening HPLC	170	0	0
	11	Extended Gradient HPLC	5	0	0
	12	Other*	10	0	0
	14	Primus Ultra ² HPLC	15	0	0
Secondary Method	1	Electrophoresis-Cellulose Acetate	5	0	0
	2	Electrophoresis-Citrate Agar	2	0	0
	4	Isoelectric focusing	43	0	0
	10	Bio-Rad Screening HPLC	29	0	0
	11	Extended Gradient HPLC	11	0	0
	12	Other*	22	0	0
	13	PCR Amplification of DNA	2	0	0
Tertiary Method	2	Electrophoresis-Citrate Agar	7	0	0
	4	Isoelectric focusing	7	0	0

Methods are designated as "Other" when less than 3 participants report results for a given method. Currently, those methods include:

IEC-HPLC	Capillarys - ALERE
MS/MS	Sebia capillarys Neonatal Haemoglobin FAST™ system

EDUCATIONAL SAMPLE - HBEDU1

Table 3a. Frequency distribution of participant reported presumptive clinical phenotype

Participant Reported Presumptive Clinical Phenotype	Frequency
FAE	51
FAE/O	2
FAC	3
FAV	2
FAS	1
FAU	1
FAE/A2	1

Table 3b. Frequency distribution of participant reported presumptive clinical assessments

Participant Reported Presumptive Clinical Assessment	Frequency
09 - Hgb E Carrier	52
22-Unidentified Variant Carrier	4
03 - Hgb C Carrier	3
20 - Assessment not listed	1
02- Hgb S Carrier	1

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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