

Newborn Screening Quality Assurance Program

PROFICIENCY TESTING

Sickle Cell Disease and Other Hemoglobinopathies

Volume 25, No. 3A

Panel 3

November 2015

INTRODUCTION

This is an amended report for the Quarterly summary of all data reported within the specified data-reporting period. On October 5, 2015 we distributed five dried blood spot (DBS) specimens prepared from umbilical cord bloods to all active participants for the Panel 3 Sickle Cell Disease and Hemoglobinopathies Proficiency Testing (PT) event. A total of 71 panels were sent by overnight mail to 48 domestic laboratories and 23 foreign laboratories. This PT report is a compilation of data reports received from 67 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 (amended)) and clinical assessments (Table 1b) appears on page 3. Table 2 shows the number of specimens reported per method by testing tier and number of phenotype and assessment errors (page 4). The testing tier corresponds to the level of confirmatory testing. The individual data verification for each laboratory follows the acknowledgment page. For this panel, we determined that two specimens, 315H2 and 315H4, would not be graded. The expected phenotype for both specimens was FA; however, many participants reported that the specimens contained unidentified variants, were unsatisfactory, had fast or aging bands, or contained Bart's Hb (Tables 1a and 1b). Because these specimens did not meet the 80% consensus for clinical assessments needed among domestic participants, they were not graded.

The next shipment of materials for the Sickle Cell and Hemoglobinopathies PT program will be in January, 2016.

MEETINGS AND TRAINING

4th Caribbean Conference on SCD to be held in Kingston, Jamaica during January 20-22, 2016. For information: <http://www.carest-network.org/2/projects/article/iv-caribbean-conference-on-sickle-cell-disease-advances-in-clinical-care>
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/tip-sheet_supporting_students_with_scd.pdf

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CDC/APHL

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**Newborn Screening Quality Assurance Program
Sickle Cell Disease and Other Hemoglobinopathies**

Specimen and Lab Certification

Year: 2015 Panel: 3

Presumptive Clinical Phenotypes

	Specimen 315H1	Specimen 315H2	Specimen 315H3	Specimen 315H4	Specimen 315H5
Expected Presumptive Phenotype	FAE	FA	FAC	FA	FAS
Accepted Presumptive Phenotypes	FAV				

Presumptive Clinical Assessments

	Specimen 315H1	Specimen1 315H2	Specimen 315H3	Specimen 315H4	Specimen 315H5
Expected Presumptive Clinical Assessment	09	01	03	01	02
Accepted Presumptive Clinical Assessments	19, 22, 21	19, 21	19, 21	19, 21	19, 21

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

Newborn Screening Quality Assurance Program

Hemoglobinopathies Proficiency Testing Program

Panel 3 - NOVEMBER 2015

Amended December 2015

Total number of program participants = 71

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

Specimen ID	Participant Reported Presumptive Clinical Phenotype	Frequency	#Correctly Classified	#Mis-Classified	#Non-Classified (no penalty)	#Data Not Reported
315H1	FAE or FEA	18	18	0	11	4
	FAV or FVA	29	29	0		
	FAVD	1	1	0		
	Other	8	8	0		
315H2	FA	7	UNGRADED		14	4
	FAD	35				
	Other	11				
315H3	FAC	65	65	0	0	4
	FCA	2	2	0		
315H4	FA	28	UNGRADED		9	4
	FA Bart's	28				
	FAV	2				
315H5	FAS	67	67	0	0	4

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
315H1	Hemoglobin E carrier	19	19	0	0	4
	Normal	3	3	0		
	Assessment not listed	2	2	0		
	Unsatisfactory sample	8	8	0		
	Unidentified variant carrier	35	35	0		
315H2	Normal	7	UNGRADED		1	4
	Hemoglobin S carrier	1				
	Hemoglobin D carrier	36				
	Assessment not listed	2				
	Unsatisfactory sample	16				
	Unidentified variant carrier	4				
315H3	Hemoglobin C carrier	66	66	0	0	4
	Unidentified variant carrier	1	0	1		
315H4	Normal	25	UNGRADED		0	4
	α -Thalassemia (Bart's Hb)	24				
	Assessment not listed	1				
	Unsatisfactory sample	9				
	Unidentified variant carrier	2				
	Fast or aging bands	6				
315H5	Hemoglobin S carrier	67	67	0	0	4

LIST OF PRESUMPTIVE CLINICAL ASSESSMENT CODES	
01 Normal - No abnormal HGB found	NORMAL
02 Hemoglobin S carrier 03 Hemoglobin C carrier 08 Hemoglobin D carrier 09 Hemoglobin E carrier	HEMOGLOBIN VARIANT CARRIERS
04 Hemoglobin SS disease (Sickle cell anemia) 05 Hemoglobin SC disease 06 Hemoglobin SD disease 12 Hemoglobin SE disease	SICKLE CELL DISEASES
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	OTHER REPORTABLE FINDINGS

Table 2. Number of samples reported per method by testing level
(For graded samples only)

Testing Level	Method Code	Method	# Samples	# Phenotype Errors**	# Assessment Errors
1	04	Isoelectric focusing	74	0	0
	10	Bio-Rad Screening HPLC	104	0	0
	12	Other*	6	0	0
	14	Primus Ulta ² HPLC	15	0	0
2	01	Electrophoresis- Cellulose Acetate	3	0	0
	04	Isoelectric focusing	59	0	0
	10	Bio-Rad Screening HPLC	10	0	0
	11	Extended Gradient HPLC	9	0	0
	12	Other*	12	0	0
	13	PCR Amplification of DNA	3	0	0
	14	Primus Ulta ² HPLC	12	0	0
3	02	Electrophoresis- Citrate Agar	6	0	0
	10	Bio-Rad Screening HPLC	6	0	0
4	10	Bio-Rad Screening HPLC	3	0	0

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

IEC-HPLC
MS/MS

Capillarys - ALERE
Sebia capillarys Neonat Haemoglobin
FAST™ system

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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