

Newborn Screening Quality Assurance Program

PROFICIENCY TESTING

Sickle Cell Disease and Other Hemoglobinopathies

Volume 25, No. 1

Panel 1

February 2015

INTRODUCTION

On January 12, 2015 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 69 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. The individual data verification for each laboratory follows the acknowledgment page.

We will continue to ship three PT panels next year for Hemoglobinopathies. The next shipment of materials from the Sickle Cell and Hemoglobinopathies PT program will be on May 4, 2015.

MEETINGS AND TRAINING

9th Annual Sickle Cell Disease Research and Educational Symposium and 38th National Sickle Cell Disease Scientific Meeting. (April 10-13, 2015) <http://fscdr.org/Meetings>

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

ACKNOWLEDGMENTS

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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: 1

Presumptive Clinical Phenotypes

	Specimen 115H1	Specimen 115H2	Specimen 115H3	Specimen 115H4	Specimen 115H5
Expected Presumptive Phenotype	FA	FAS	FAE	FA	FA
Accepted Presumptive Phenotypes	FA	FAS	FAE FAV FAC for IEF <i>only labs</i> FA + Other	FA	FA

Presumptive Clinical Assessments

	Specimen 115H1	Specimen1 115H2	Specimen 115H3	Specimen 115H4	Specimen 115H5
Expected Presumptive Clinical Assessment	01	02	09	01	01
Accepted Presumptive Clinical Assessments	01	02	09 20 03 for IEF <i>only labs</i> 22	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

Newborn Screening Quality Assurance Program

Hemoglobinopathies Proficiency Testing Program

Panel 1 - February 2015

Total number of program participants = 73

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
115H1	FA	65	65	0	0	8
115H2	FAS	65	65	0	0	8
115H3	FAE	52	52	0	0	8
	FEE	1	0	1		
	FE	1	0	1		
	FA	3	0	3		
	FAC*	2	2	0		
	FAV	3	3	0		
	FAE/O	2	2	0		
	FA+Other	1	1	0		
115H4	FA	65	65	0	0	8
115H5	FA	65	65	0	0	8

*FAC considered correct only in laboratories who do not have either an E control or a more sensitive method for confirmation.

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
115H1	01	69	69	0	0	4
115H2	02	69	69	0	0	4
115H3	01	3	0	3	0	4
	03	2	2	0		
	09	58	58	0		
	18	2	0	2		
	22	4	4	0		
115H4	01	69	69	0	0	4
115H5	01	68	68	0	0	4
	02	1	0	1		

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

Testing Level	Method Code	Method	# Samples	# Phenotype Errors**	# Assessment Errors
1	04	Isoelectric focusing	144	2	3
	10	Bio-Rad Screening HPLC	177	2	2
	12	Other*	5	0	0
	14	Primus Ulta ² HPLC	25	1	1
2	01	Electrophoresis- Cellulose Acetate	5	0	0
	02	Electrophoresis- Citrate Agar	2	0	0
	04	Isoelectric focusing	61	1	1
	10	Bio-Rad Screening HPLC	25	0	0
	11	Extended Gradient HPLC	12	0	0
	12	Other*	17	0	0
	13	PCR Amplification of DNA	2	0	0
3	02	Electrophoresis- Citrate Agar	7	0	0
	04	Isoelectric focusing	1	0	0
	10	Bio-Rad Screening HPLC	2	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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