

Newborn Screening Quality Assurance Program

PROFICIENCY TESTING

Sickle Cell Disease and Other Hemoglobinopathies

Volume 25, No. 2

Panel 2

June 2015

INTRODUCTION

On May 4, 2015 we distributed five dried blood spot (DBS) specimens prepared from umbilical cord bloods to all active participants for the Panel 2 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 66 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. 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.

We included an educational specimen, 22015, in this panel. The educational specimen was created by pooling residual blood collected for hemoglobin confirmatory testing of newborns and spotting the pooled blood onto filter paper. The specimens were from individuals with the FSC phenotype, including one from a baby that was transfused. Consequently, a small amount of Hb A was detected by some methods. Of the 65 participants who reported results for this specimen, 64 reported the correct

phenotype and 61 reported the correct clinical assessment (Tables 3a and 3b, page 5). On page 6, Table 3c we also show the frequency of the reported presumptive phenotypes by method.

The next shipment of materials from the Sickle Cell and Hemoglobinopathies PT program will be on October 5, 2015.

MEETINGS AND TRAINING

Sickle Cell in Focus (SCiF), London, UK, 6/15-16, 2015. For more information go to: www.ststn.co.uk

SCDAA 43rd Annual Convention, 9/23-26, 2015 – Baltimore, MD. For more information go to: <http://www.sicklecelldisease.org/index.cfm?page=annual-convention>

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: 2

Presumptive Clinical Phenotypes

	Specimen 215H1	Specimen 215H2	Specimen 215H3	Specimen 215H4	Specimen 215H5	EDU 22015
Expected Presumptive Phenotype	FAC	FAS	FA	FA	FAS	FSC
Accepted Presumptive Phenotypes	FAC	FAS	FA	FA	FAS	FSC, FASC

Presumptive Clinical Assessments

	Specimen 215H1	Specimen 215H2	Specimen 215H3	Specimen 215H4	Specimen 215H5	EDU 22015
Expected Presumptive Clinical Assessment	03	02	01	01	02	05
Accepted Presumptive Clinical Assessments	03	02	01	01	02	05

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 2 – June 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
215H1	FAC	60	60	0	0	5
	FCA	6	0	6		
215H2	FAS	65	65	0	0	5
	FS	1	0	1		
215H3	FA	65	65	0	0	5
	FA IND	1	0	1		
215H4	FA	65	65	0	0	5
	FA IND	1	0	1		
215H5	FAS	65	65	0	0	5
	FS	1	0	1		

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
215H1	03	65	65	0	0	5
	08	1	0	1		
215H2	02	65	65	0	0	5
	04	1	0	1		
215H3	01	65	65	0	0	5
	22	1	0	1		
215H4	01	65	65	0	0	5
	22	1	0	1		
215H5	02	65	65	0	0	5
	04	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	139	0	1
	10	Bio-Rad Screening HPLC	157	0	0
	12	Other*	10	0	0
	14	Primus Ultra ² HPLC	20	2	2
2	01	Electrophoresis-Cellulose Acetate	5	0	0
	02	Electrophoresis-Citrate Agar	3	0	0
	04	Isoelectric focusing	59	4	5
	10	Bio-Rad Screening HPLC	35	0	0
	11	Extended Gradient HPLC	9	0	0
	12	Other*	13	0	0
	13	PCR Amplification of DNA	3	0	0
	14	Primus Ultra ² HPLC	16	0	0
3	02	Electrophoresis-Citrate Agar	8	0	0
	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

EDUCATIONAL SAMPLE - 22015

Total number of participants who reported results for EDU 22015 = 65

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

Participant Reported Presumptive Clinical Phenotype	Frequency	Reviewer Comment
FSC	50	Certified Presumptive Phenotype
FSCA	3	Acceptable Phenotypes due to the presence of F, A, S and C hemoglobins
FSCa	3	
FCS	3	
FASC	3	
FaSC	1	
FCSA	1	
FV	1	Incorrect Phenotype

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

Participant Reported Presumptive Clinical Assessment	Frequency	Acceptable	Participant comment used in evaluation
05 Hemoglobin SC disease	59	59	Acceptable: A is <1%
			Acceptable: 1.3% A on HPLC
			Acceptable: HPLC result was FSCa. With 5.8%S, 5.2%C, and 1.3%A
			Acceptable: This sample would be unsatisfactory if it was a patient; due to the presence of Hgb a. We would ask for another sample.
			Acceptable: pattern - FSCa A= 1.2% S= 5.4% S= 4.3%
20 Assessment not listed	2	1	Acceptable: Specimen appears to be SC disease, but has Hb A present on both IEF and HPLC which is not consistent with SC disease unless the baby is transfused.
			Unacceptable: Both IEF and HPLC showed presence of Adult hemoglobin. (SC disease not mentioned in clinical assessment or comment)
22 Unidentified variant carrier	1	0	Unacceptable: Cannot ID. Got FCSU, FAS and FSC
02 Hb S carrier 03 Hb C carrier	1	0	
05 Hb SC disease 20 Assessment not listed	1	1	
Not Reported	1	1	

Table 3c. Frequency of Reported Phenotypes by Method.

Method	Phenotypes Reported	Frequency
IEF	FSC	24
	FSCA	2
	FASC	2
	FCS	1
HPLC	FSC	26
	FSCA	2
	FSCa	3
	FCS	2
	FASC	1
	FaSC	1
Other	FCSA	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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