

Newborn Screening Quality Assurance Program

PROFICIENCY TESTING

Sickle Cell Disease and Other Hemoglobinopathies

Volume 26, No. 1

Panel 1

February 2016

INTRODUCTION

On January 11, 2016 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 76 panels were sent by overnight mail to 48 domestic laboratories and 28 foreign laboratories. This PT report is a compilation of data reports received from 72 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 assess-

ments, 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.

The next shipment of materials for the Sickle Cell and Hemoglobinopathies PT program will be May 2, 2016.

MEETINGS AND TRAINING

Sickle Cell in Focus (SCiF) 2016, National Institutes of Health, Bethesda, Maryland, USA June 2-3, 2016. Website: <http://www.ststn.co.uk/scif/sicf2016/>

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

The 2015 guidance document by the CDC-supported, APHL Hemoglobinopathy Laboratory Workgroup, "Hemoglobinopathies: Current Practices for Screening, Confirmation and Follow-up" can be downloaded at http://www.aphl.org/AboutAPHL/publications/Documents/NBS_Hemoglobinopathy-Testing_122015.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: 2016 Panel: 1

Presumptive Clinical Phenotypes

	Specimen 116H1	Specimen 116H2	Specimen 116H3	Specimen 116H4	Specimen 116H5
Expected Presumptive Phenotype	FA	FAE	FAC	FAS	FA
Accepted Presumptive Phenotypes	FA	FAE, FAV	FAC	FAS	FA

Presumptive Clinical Assessments

	Specimen 116H1	Specimen1 116H2	Specimen 116H3	Specimen 116H4	Specimen 116H5
Expected Presumptive Clinical Assessment	01	09	03	02	01
Accepted Presumptive Clinical Assessments	01	09, 22	03	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

Newborn Screening Quality Assurance Program

Hemoglobinopathies Proficiency Testing Program

Panel 1 - FEBRUARY 2016

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
116H1	FA	71	71	0	0	5
116H2	FAE	56	1	0	0	5
	FAV	5	5			
	FAE/O	1	1			
	FA+Other	1	1			
	FAU	1	1			
	FA	7	0	7		
116H3	FAC	70	70	0	0	5
	FA	1	0	1		
116H4	FAS	70	70	0	0	5
	FA	1	0	1		
116H5	FA	70	70	0	1	5

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
116H1	01 - Normal	72	72	0	0	4
116H2	09 - HbE Carrier	58	58	0	0	4
	22 - Unidentified Variant Carrier	7	7	0		
	01 - Normal and 22 - Unidentified Variant Carrier	1	1	0		
	01 - Normal	6	0	6		
	16 - Bart's Hb (Reported as Other CA)	1	0	1		
116H3	03 - HbC Carrier	71	71	0	0	4
	01 - Normal	1	0	1		
116H4	02 - HbS Carrier	71	71	0	0	4
	01 - Normal	1	0	1		
116H5	01 - Normal	71	71	0	1	4
	21 - Unsatisfactory sample	1	0	0		

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	134	5	5
	10	Bio-Rad Screening HPLC	181	2	2
	12	Other*	10	0	0
	14	Primus Ultra ² HPLC	30	2	1
2	01	Electrophoresis- Cellulose Acetate	5	0	0
	04	Isoelectric focusing	85	2	2
	10	Bio-Rad Screening HPLC	35	0	0
	11	Extended Gradient HPLC	11	0	0
	12	Other*	8	0	0
	13	PCR Amplification of DNA	5	0	0
3	02	Electrophoresis- Citrate Agar	8	0	0
	04	Isoelectric focusing	2	0	0
	10	Bio-Rad Screening HPLC	5	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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