

# Newborn Screening Quality Assurance Program

## PROFICIENCY TESTING

## Sickle Cell Disease and Other Hemoglobinopathies

Volume 24, No. 3

Panel 3

November 2014

### INTRODUCTION

On October 6, 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 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 January 12, 2015. ❖

### MEETINGS AND TRAINING

Global Sickle Cell Diseases Network Conference: Rio De Janeiro, Brazil, 2014. At: [www.globalsickleceldisease.org](http://www.globalsickleceldisease.org) November 11-14, 2014.

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](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  
Hemoglobinopathies Proficiency Testing Program**

***Specimen and Lab Certification***

Year: 2014 Panel: 3

**Presumptive Clinical Phenotypes**

	<b>Specimen 314H1</b>	<b>Specimen 314H2</b>	<b>Specimen 314H3</b>	<b>Specimen 314H4</b>	<b>Specimen 314H5</b>
<b>Expected Presumptive Phenotype</b>	FAS	FAC	FA	FAS	FAC
<b>Accepted Presumptive Phenotypes</b>	FAS	FAC	FA	FAS	FAC

**Presumptive Clinical Assessments**

	<b>Specimen 314H1</b>	<b>Specimen 314H2</b>	<b>Specimen 314H3</b>	<b>Specimen 314H4</b>	<b>Specimen 314H5</b>
<b>Expected Presumptive Clinical Assessment</b>	02	03	01	02	03
<b>Accepted Presumptive Clinical Assessments</b>	02	03	01	02	03

**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

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**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 <sup>2</sup> HPLC      |                             |
-

Newborn Screening Quality Assurance Program  
Hemoglobinopathies Proficiency Testing Program

Panel 3 – November 2014

*Total number of program participants = 74*

**Table 1a.** Frequency distribution of Reported Presumptive Phenotypes

Specimen ID	Participant Reported Presumptive Clinical Phenotype	Frequency	#Correctly Classified	#Mis-Classified	#Non-Classified (no penalty)	#Data Not Reported
314H1	FA	2	0	2	0	4
	FAS	68	68	0		
314H2	FAC	68	68	0	0	4
	FAC/FAC Bart's	1	0	1		
	FACS	1	0	1		
314H3	FA	70	70	0	0	4
	FAS	1	0	1		
314H4	FA	2	0	2	0	4
	FAS	68	68	0		
314H5	FAC	70	70	0	0	4

**Table 2a.** Frequency distribution of Reported Presumptive Clinical Assessments

Specimen ID	Participant Reported Presumptive Clinical Assessment	Frequency	#Correctly Classified	#Mis-Classified	#Non-Classified (no penalty)	#Data Not Reported
314H1	FA	2	0	2	0	4
	FAS	68	68	0		
314H2	FAC	68	68	0	0	4
	FAC/FAC Bart's	1	0	1		
	FACS	1	0	1		
314H3	FA	70	70	0	0	4
	FAS	1	0	1		
314H4	FA	2	0	2	0	4
	FAS	68	68	0		
314H5	FAC	70	70	0	0	4

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

Testing Tier	Method Code	Method	# Samples	# Phenotype Errors	# Assessment Errors
1	04	Isoelectric focusing	155	4	4
	10	Bio-Rad Screening HPLC	167	3	3
	11	Extended Gradient HPLC	5	0	0
	12	Other*	5	0	0
	14	Primus Ultra <sup>2</sup> HPLC	5	0	0
2	01	Electrophoresis-Cellulose Acetate	5	0	0
	02	Electrophoresis-Citrate Agar	4	0	0
	04	Isoelectric focusing	66	1	0
	10	Bio-Rad Screening HPLC	39	0	0
	11	Extended Gradient HPLC	13	0	0
	12	Other*	19	0	0
	13	PCR Amplification of DNA	3	0	0
	14	Primus Ultra <sup>2</sup> HPLC	16	0	0
3	02	Electrophoresis-Citrate Agar	9	0	0
	10	Bio-Rad Screening HPLC	4	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

Capillars - ALERE  
Sebia capillars 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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