Newborn Screening Quality Assurance Program

Quarterly Report
Volume 27, No.1

Sickle Cell and Other Hemoglobinopathies Proficiency Testing Program (HbPT)

2017 Quarter 1 February

Introduction

This report is the summary of HbPT data reported within the specified data-reporting period for Quarter 1, 2017. It is distributed to all participants, state laboratory directors, and program colleagues by request. The content includes specimen certification profiles, material distribution information, and misclassifica-tion frequency tables for presumptive phenotypes, clinical assessments, and reported methods. A evaluation of your reported data is attached to this summary.

Certification of PT Specimens

The dried blood spot (DBS) specimens in this panel were prepared from purchased umbilical cord blood. Table 1 lists the detected hemoglobin phenotypes and their presumptive clinical assessments.

Table 1. Specimen Certification

Specimen	Expected Presumptive Phenotype	Accepted Presumptive Phenotype	Expected Presumptive Clinical Assessment *	Accepted Presumptive Clinical Assessment*
117H1	FS	FSU	04	23
117H2	FAE	FAV, FAU	09	22
117H3	FAC	FAV, FAU	03	22
117H4	FAS		02	
117H5	FA		01	

***Clinical Assessment Codes**

Normal Hemoglobin Pattern
01 Normal - no abnormal Hb found

Hemoglobin Variant Carriers
02 Hemoglobin S trait
03 Hemoglobin C trait
08 Hemoglobin D trait
09 Hemoglobin E trait

Sickle Cell Diseases
04 Hemoglobin SS disease
05 Hemoglobin SC disease
06 Hemoglobin SD disease
12 Hemoglobin SE disease
23 Hemoglobin S with an uncommon variant
24 Hemoglobin C with an uncommon variant
25 Hemoglobin D with an uncommon variant
26 Hemoglobin E with an uncommon variant

Other Reportable Findings
16 Alpha thalassemia (Bart's Hb)
17 F only (Beta Thalessemia Major)
18 Hemoglobin E, E disease
19 Aging bands (clinically insignificant)
20 Assessment not listed
21 Unsatisfactory sample
22 Unidentified variant trait

Distribution of PT Specimens

January 11, 2017 a PT panel of five DBS specimens was distributed to all program participants. A total of 76 panels were sent to 48 domestic and 28 foreign laboratories.

Participant Results

We received data from 76 participants by the data reporting deadline. We requested that participants assay all survey specimens by the analytical schemes they routinely use and report for each specimen to determine the presumptive phenotype, presumptive clinical assessment, and any other clinical classifications they deem consistent with their analytic results and program operations. Laboratories were asked to report one presumptive phenotype derived from results of all analytic methods used by their laboratory. When reporting the phenotype, we require that participants list the hemoglobins in the order of their abundance using standard phenotypic nomenclature. We also recommend that participants supplement unusual phenotype reports with comments in the Phenotype Comments section of the HbPT Data Report Form. Table 2 shows the frequency distribution of participant reported presumptive clinical phenotypes along the frequency of misclassifications. Table 3 shows the frequency distribution of participant reported presumptive clinical assessments and the frequency of misclassifications.

Table 2. Frequency Distribution of Reported Phenotypes

Specimen	Presumptive Clinical Phenotype	Frequency	#Correctly Classified	#Misclassified	#Data Not Reported
117H1	FS	55	55	0	0
	FSS	2	2	0	
	FSA	2	2	0	
	FSa	3	3	0	
	Fsa	2	2	0	
	FSE	3	3	0	
	FSV	2	2	0	
	FAS	4	0	4	
	Other	3	2	1	
117H2	FAE	63	63	0	0
	FAV	5	5	0	
	FA	3	0	3	
	Other	5	2	3	

Table 2. Frequency Distribution of Reported Phenotypes (cont.)

Specimen	Presumptive Clinical Phenotype	Frequency	#Correctly Classified	#Misclassified	#Data Not Reported
117H3	FAC	71	71	0	0
	FCA	3	3	0	
	Other	2	0	2	
117H4	FAS	71	71	0	0
	Other	5	0	5	
117H5	FA	74	74	0	0
	Other	2	1	1	

Table 3. Participant Reported Assessments

Specimen	Presumptive Clinical Assessment*	Frequency	#Correctly Classified	#Misclassified	#Data Not Reported
117H1	04	63	63	0	0
	02	4	0	4	
	20	3	3	0	
	23	3	3	0	
	12	3	3	0	
117H2	09	66	66	0	0
	22	5	5	0	
	01	3	0	3	
	Other	2	0	2	
117H3	03	75	75	0	0
	Other	1	0	1	
117H4	02	72	72	0	0
	Other	4	0	4	
117H5	01	76	76	0	0

♦ Reported Methods Data

Table 4 shows the number of specimens reported per method by testing tier and number of phenotype and assessment misclassifications. The testing tier corresponds to the level of confirmatory testing.

Table 4. Number of Samples Reported Per Method By Testing Level

Testing Level	Method Code	Method	# Samples	# Phenotype Misclassifications	# Assessment Misclassifications
1	04	Isoelectric Focusing	156	10	10
	10	Bio-Rad Screening HPLC	181	8	3
	12	Other*	15	0	0
	14	Prima Ulta^2 HPLC	30	1	1
2	01	Electrophoresis—Cellulose Acetate	5	0	0
	04	Isoelectric Focusing	87	0	0
	10	Bio-Rad Screening HPLC	45	0	0
	11	Extended Gradient HPLC	9	0	0
	12	Other*	14	5	0
	13	PCR Amplification of DNA	4	0	0
	14	Prima Ulta^2 HPLC	21	0	0
3	02	Electrophoresis—Citrate Agar	5	0	0
	04	Isoelectric Focusing	2	0	0
	10	Bio-Rad Screening HPLC	10	0	0
	12	Other*	4	0	0
	13	PCR Amplification of DNA	1	0	0

*Methods are designated as “Other” when less than three participants report results for a given method. Currently those methods include:

IEC-HPLC

MS/MS

Capillarys—ALERE

Sebia capillarys Neonatal Haemoglobin

Evaluations

Overall, participants reported 19 Presumptive Phenotype misclassifications and 14 Presumptive Clinical Assessment misclassifications.

Future Shipments

The Newborn Screening Quality Assurance Program will ship next quarter's PT specimens for HbPT on April 3, 2017.

Acknowledgements

The specimens for this program were prepared from umbilical cord blood samples supplied by Cleveland Cord Blood Center, Cleveland, Ohio; CORD:USE Cord Blood Bank, Orlando, FL; Carolinas Cord Blood Bank, Raleigh, NC; and Life Line Stem Cell, New Haven, IN. Patient specimens were provided by Children's Hospital Oakland Research Institute (CHORI).

The content of this report may also be located on our website at:
http://www.cdc.gov/labstandards/nsqap_reports.html

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NEWBORN SCREENING QUALITY ASSURANCE PROGRAM

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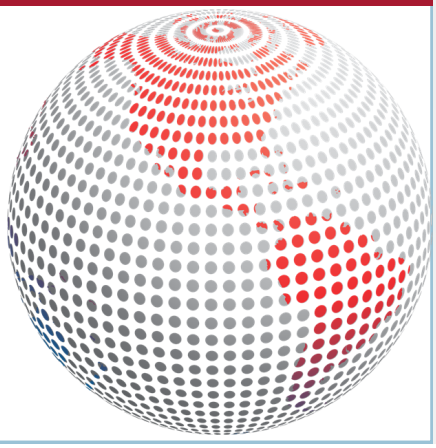

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