Newborn Screening Quality Assurance Program

Quarterly Report
Volume 27, No.2

Sickle Cell and Other Hemoglobinopathies Proficiency Testing Program (HbPT)

2017 Quarter 2 May

Introduction

This report is the summary of HbPT data reported within the specified data-reporting period for Quarter 2, 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 misclassification 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*
217H1	FS	FSU	04	22,23
217H2	FS	FSU	04	22,23
217H3	FA	FA	01	
217H4	FA	FA	01	
217H5	FAS	FAS	02	

***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

On April 3, 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 72 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
217H1	FS	38	38	0	
	FSV	13	13	0	
	FSU	5	5	0	
	FAS	3	0	3	
	Other	13	13	0	
217H2	FS	55	55	0	
	FSa	4	4	0	
	FSE	4	4	0	
	FAS	1	0	1	4
	Other	8	8	0	
217H3	FA	71	71	0	
	FAA	1	1	0	
217H4	FA	71	71	0	
	FAA	1	1	0	
217H5	FAS	70	70	0	
	FA	1	0	1	
	FSA	1	0	1	

Table 3. Participant Reported Assessments

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Specimen	Presumptive Clinical Assessment*	Frequency	#Correctly Classified	#Misclassified	#Data Not Reported
217H1	04	48	48	0	4
	23	14	14	0	
	02	5	0	5	
	Other	5	5	0	
217H2	04	60	60	0	
	23	5	5	0	
	02	1	0	1	
	Other	6	6	0	
217H3	01	72	72	0	
217H4	01	72	72	0	
217H5	02	71	71	0	
	01	1	0	1	

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♦ 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	01	Electrophoresis —	5	0	0
	04	Isoelectric Focusing	130	3	4
	10	Bio-Rad Screening	185	3	3
	12	Other*	15	0	0
	14	Prima Ulta^2 HPLC	25	0	0
2	01	Electrophoresis— Cellulose Acetate	5	0	0
	04	Isoelectric Focusing	75	1	2
	10	Bio-Rad Screening	39	0	0
	11	Extended Gradient HPLC	8	0	0
	12	Other*	13	2	2
	13	PCR Amplification of DNA	3	0	0
	14	Prima Ulta^2 HPLC	16	0	0
3	02	Electrophoresis— Citrate Agar	5	0	0
	04	Isoelectric Focusing	2	0	0
	12	Other*	3	0	0
	13	PCR Amplification of DNA	2	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 six Presumptive Phenotype misclassifications and seven Presumptive Clinical Assessment misclassifications.

Future Shipments

The Newborn Screening Quality Assurance Program will ship next quarter's PT specimens for HbPT on October 2, 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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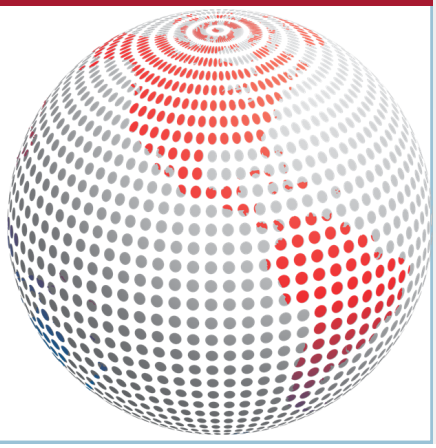

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