Newborn Screening Quality Assurance Program

Quarterly Report
Volume 28, No.1

Sickle Cell and Other Hemoglobinopathies Proficiency Testing Program (HbPT)

Issued: March 12, 2018

Introduction

This report is the summary of HbPT data reported within the specified period for Quarter 1, 2018. It is distributed to all participants, state laboratory directors, and program colleagues by request. The content includes specimen certification profiles, material distribution information, 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 hemoglobin presumptive 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*
118H1	FAS		02 Hemoglobin S trait	01 Normal - no abnormal Hb found
118H2	FA		01 Normal - no abnormal Hb found	
118H3	FAC		03 Hemoglobin C trait	
118H4	FAB	FA	16 Alpha thalassemia (Bart's Hb)	30 Alpha thalassemia — silent carrier 01 Normal - no abnormal Hb found
118H5	FA		01 Normal - no abnormal Hb found	

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
30 Alpha thalassemia — silent carrier

Distribution of PT Specimens

On January 9, 2018 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 73 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 the presumptive phenotype, presumptive clinical assessment, and any other clinical classifications they deem consistent with their analytic results and program operations.

Participants should be mindful of reporting presumptive phenotypes and presumptive clinical assessments as directed on the Hemoglobinopathies Data Report Form to avoid point deductions from their overall score. Laboratories should:

- Report one presumptive phenotype derived from results of all methods used by their laboratory for each specimen. We also recommend that participants supplement unusual phenotype reports with comments in the Phenotype Comments section of the HbPT Data Report Form.
- List the hemoglobins in the order of their abundance using standard phenotypic nomenclature when reporting the phenotype.
- Not insert symbols or blank spaces into the presumptive phenotype nomenclature.
- Report presumptive clinical assessments not listed in the drop-down menu under the comment section in order to receive points for your overall score.

Table 2 shows the frequency distribution of participant reported presumptive clinical phenotypes along the frequency of misclassifications. Table 3 shows the frequency distribution of reported presumptive clinical assessments and the frequency of misclassifications.

Table 2. Frequency Distribution of Reported Phenotypes

Specimen	Presumptive Clinical Phenotype	Frequency	#Correctly Classified	Misclassifications*
118H1	FAS	68	68	0
	Other	5	0	3
118H2	FA	71	71	0
	Other	2	0	0
118H3	FAC	70	70	0
	Other	3	0	1
118H4	FA	46	46	0
	FAB	7	7	0
	FA BARTS	16	16	0
	Other	4	1	1
118H5	FA	71	71	0
	Other	2	0	0

*Clerical errors are not reflected in the misclassification frequency shown.

Table 3. Frequency Distribution of Presumptive Clinical Assessments

Specimen	Presumptive Clinical Assessment	Frequency	#Correctly Classified	# Misclassifications
118H1	Hemoglobin S trait	71	71	0
	Other	2	0	2
118H2	Normal - no abnormal Hb found	72	72	0
	Other	1	0	1
118H3	Hemoglobin C trait	72	72	0
	Other	1	0	1
118H4	Normal - no abnormal Hb found	40	40	0
	Alpha thalassemia	27	27	0
	Assessment not listed	5	4	1
	Hemoglobin C trait	1	0	1
118H5	Normal - no abnormal Hb found	71	71	0
	Other	1	0	1

♦ Total Specimen Error Frequency by Testing Algorithm

Table 4 shows the frequency of errors per testing algorithm for all specimens. Algorithms reported by less than two participants are not shown.

Table 4. Frequency of Errors Per Testing Algorithm

Primary	Secondary	Tertiary	Total Specimens	Presumptive Phenotype Errors	Presumptive Clinical Assessment Errors*
Electrophoresis - Cellulose Acetate			5	0	0
Isoelectric focusing			54	2	1
Isoelectric focusing	Isoelectric focusing		4	0	0
Isoelectric focusing	Isoelectric focusing	Bio-Rad Screen-ing HPLC	5	0	0
Isoelectric focusing	Isoelectric focusing	Other Methods	3	0	0
Isoelectric focusing	Bio-Rad Screening HPLC		26	0	0
Isoelectric focusing	Primus Ulta 2 HPLC		19	0	0
Isoelectric focusing	Electrophoresis - Cellulose Acetate	Electrophoresis - Citrate Agar	5	0	0
Bio-Rad Screening HPLC			126	0	0
Bio-Rad Screening HPLC	Isoelectric focusing		51	2	5
Bio-Rad Screening HPLC	Bio-Rad Screening HPLC		7	0	0
Bio-Rad Screening HPLC	Other Methods		10	1	0
Other Methods			5	0	0
Other Methods	Isoelectric focusing		5	0	0
Other Methods	Other Methods		5	0	0
Primus Ulta 2 HPLC			20	0	0
Primus Ulta 2 HPLC	Isoelectric focusing		10	0	0

*Algorithms reported by less than two participants are not shown.

*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

♦ Specimen 118H4

Specimen 118H4 was prepared from an umbilical cord blood unit containing Bart's Hb (alpha-thalassemia). We sorted the frequency of presumptive phenotypes and presumptive clinical assessments by testing algorithm in Table 5. Phenotypes are listed as reported on the participant data report form. Algorithms that used electrophoresis alone or in combination with another method were more likely to detect the low percentage of Bart's Hb present in the specimen.

Table 5. Phenotypes and Clinical Assessments for Specimen 118H4 Sorted by Testing Algorithm

Algorithm	Total Participants	Presumptive Phenotype	Presumptive Phenotype-Frequency	Presumptive Clincial Assessment	Presumptive Clinical Assessment Frequency
Cellulose Acetate	1	FA	1	Normal	1
IEF	8	FAB	1	Alpha thalassemia (Bart's Hb)	7
		FA Bart's	3		
		Bart's	2		
		FA	1	Normal	1
		FA + Bart's	1		
IEF + IEF	2	FA#	1	Alpha thalassemia (Bart's Hb)	2
		FA	1		
IEF + HPLC	11	FA	4	Normal	5
		FA + Bart's	2	Alpha thalassemia (Bart's Hb)	6
		FAb	1		
		FAB	1		
		FAB2	1		
		Brt+	1		
		FA Barts	1		
IEF + Cellulose Acetate + Citrate Agar	1	FA + Bart's	1	Alpha thalassemia (Bart's Hb)	1
IEF + IEF + HPLC	2	FAB	2	Alpha thalassemia (Bart's Hb)	2
HPLC	30	FA	28	Normal	28
		FA fast other	1	Alpha thalassemia (Bart's Hb)	1
				Assessment not listed	1
HPLC + IEF	12	FA	2	Normal	2
		FA+BARTS	1	Alpha thalassemia (Bart's Hb)	7
		FA+B	1	Assessment not listed	2
		FAB	1		
		FAB'S	1		
		FAC	1	Hemoglobin C trait	1
		FABart's	1		
HPLC + HPLC	1	FA	1	Normal	1
HPLC + Capillary Electrophoresis	2	FAX	1	Assessment not listed	2
Capillary Electrophoresis	1	F>A>Barts	1	Alpha thalassemia (Bart's Hb)	1
IEC– HPLC + MS/MS	1	FA	1	Normal	1
MS/MS + IEF	1	FA	1	Normal	1

Evaluations

Overall, participants reported five Presumptive Phenotype misclassifications and seven Presumptive Clinical Assessment misclassifications. Presumptive phenotype and presumptive clinical assessment scoring deductions were issued where reporting instructions were not followed.

Future Shipments

The Newborn Screening Quality Assurance Program will ship next panel of specimens for HbPT on April 3, 2018.

Acknowledgements

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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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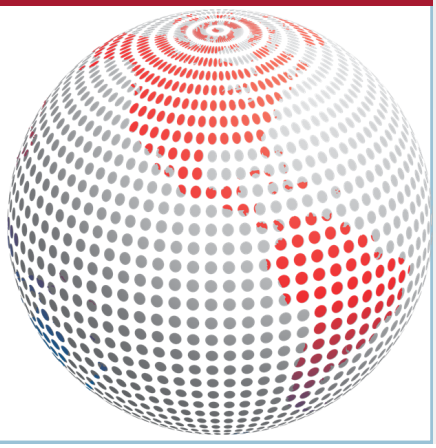

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