Newborn Screening Quality Assurance Program

Quarterly Report
Volume 27, No.3

Sickle Cell and Other Hemoglobinopathies Proficiency Testing Program (HbPT)

Issued: November 27, 2017

Introduction

This report is the summary of HbPT data reported within the specified period for Quarter 4, 2017. 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 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*
417H1	FAS		02 Hemoglobin S trait	
417H2	FA		01 Normal - no abnormal Hb found	
417H3	FAS		02 Hemoglobin S trait	
417H4	FA		01 Normal - no abnormal Hb found	
417H5	FS	FSU	04 Hemoglobin SS disease	23 Hemoglobin S with an uncommon variant

***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 October 2, 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 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. Laboratories were asked to report one presumptive phenotype derived from results of all 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 reported presumptive clinical assessments and the frequency of misclassifications.

Table 2. Frequency Distribution of Reported Phenotypes

Specimen	Presumptive Clinical Phenotype	Frequency	#Correctly Classified	#Misclassified	Phenotype Not Reported
417H1	FAS	66	66	0	1
	Other	7	1	5	
417H2	FA	71	71	0	1
	Other	2	1	0	
417H3	FAS	63	63	0	1
	FSA	5	0	5	
	Other	5	2	2	
417H4	FA	71	71	0	1
	Other	2	1	0	
417H5	FAS	3	0	3	1
	FS	43	43	0	
	FSA	3	3	0	
	FSU	4	4	0	
	FSV	10	10	0	
	Other	10	7	2	

Table 3. Frequency Distribution of Presumptive Clinical Assessments

Specimen	Presumptive Clinical Assessment	Frequency	#Correctly Classified	#Misclassified	Clinical Assessment Not Reported
417H1	Hemoglobin S trait	69	69	0	0
	Other	4	0	4	
417H2	Normal - no abnormal Hb found	73	73	0	0
417H3	Hemoglobin S trait	71	71	0	0
	Other	2	0	2	
417H4	Normal - no abnormal Hb found	73	73	0	0
417H5	Hemoglobin S trait	5	0	5	0
	Hemoglobin SS disease	48	48	0	
	Assessment not listed	4	3	1	
	Hemoglobin S with an uncommon variant	12	12	0	
	Other	4	2	2	

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

.

♦ Methods Reported

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 — Cellulose Acetate	5	3	3
	04	Isoelectric Focusing	125	9	6
	10	Bio-Rad Screening HPLC	190	4	3
	12	Other*	15	1	1
	14	Prima Ulta^2 HPLC	30	0	0
2	01	Electrophoresis— Cellulose Acetate	5	0	0
	04	Isoelectric Focusing	80	3	2
	10	Bio-Rad Screening HPLC	34	3	3
	11	Extended Gradient HPLC	6	0	0
	12	Other*	13	0	0
	14	Prima Ulta^2 HPLC	12	0	0
3	02	Electrophoresis— Citrate Agar	5	0	0
	04	Isoelectric Focusing	2	0	0
	12	Other*	3	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 17 Presumptive Phenotype misclassifications and 14 Presumptive Clinical Assessment misclassifications.

Future Shipments

The Newborn Screening Quality Assurance Program will ship next panel of specimens for HbPT in January 2018.

Acknowledgements

The specimens for this program were prepared from umbilical cord blood samples supplied by Cleveland Cord Blood Center, Cleveland, Ohio; 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

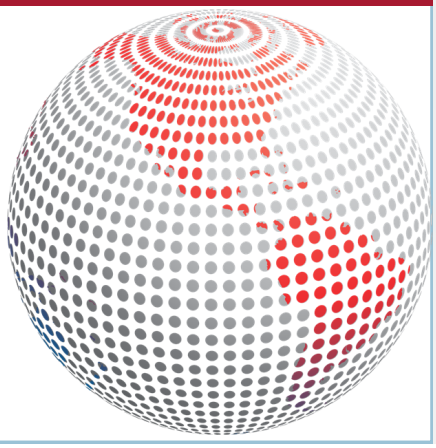
This program is co-sponsored by the Centers for Disease Control and Prevention (CDC) and The Association of Public Health Laboratories (APHL)

NEWBORN SCREENING QUALITY ASSURANCE PROGRAM

Direct inquiries to:

Centers for Disease Control and Prevention
4770 Buford Highway NE, MS/F19
Atlanta, GA 30341-3724
Email: jvm0@cdc.gov

Editors
Joanne Mei
Irene Williams



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.

INQUIRIES TO:

Irene Williams, Editor • Centers for Disease Control and Prevention (CDC) • Newborn Screening Quality Assurance Program
Mailstop F-24 • 4770 Buford Highway, N.E. • Atlanta, GA 30341-3724
Phone (770) 488-4582 • NSQAPDMT@cdc.gov
E-mail: IWilliams1@cdc.gov