



Newborn Screening Quality Assurance Program

PROFICIENCY TESTING

Sickle Cell Disease and Other Hemoglobinopathies

Volume 22, No. 2

Panel 2

June 2012

INTRODUCTION

On April 30, 2012, we distributed five dried-blood-spot (DBS) specimens prepared from umbilical cord bloods to all active participants for the Panel 2 Sickle Cell Disease and Hemoglobinopathies Proficiency Testing (PT) event. A total of 75 panels were mailed by overnight FedEx mail. The packages went to 51 domestic laboratories and 24 foreign laboratories. This PT report is a compilation of data reports received from 67 of the participating laboratories by the designated deadline date. There were 8 laboratories that did not report this quarter. We distribute this quarterly 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 and clinical assessments appears on page 3.

The individual data verification for each laboratory with evaluation comments appears on page 4.

We will continue to ship three PT panels this year for Hemoglobinopathies. The next shipment of materials from the Sickle Cell and Hemoglobinopathies PT program will be on October 1, 2012. ❖

MEETINGS AND TRAINING

Research led by Howard Hughes Medical Institute (HHMI) investigator Stuart H. Orkin of Children's Hospital Boston, Dana Farber Cancer Institute, and Harvard Medical School shows that silencing a protein known as BCL11A can reactivate fetal hemoglobin production in adult mice and effectively reverses sickle cell disease. The findings, reported October 13, 2011, in *Science Express*, reveals that BCL11A is one of the primary factors involved

in turning off fetal hemoglobin production. The paper's first author, Jian Xu said, "It has been hypothesized for three decades that fetal hemoglobin could be turned on once we understood the mechanism of hemoglobin switching, and this is the first evidence of a target to do that."

ACKNOWLEDGMENTS

The specimens for this survey were prepared from umbilical cord blood samples supplied by Cleveland Cord Blood Center, Cleveland, Ohio. They are an independent not-for-profit 501(c)(3) organization that accepts donated cord blood for clinical use. ❖

CDC/APHL

This program is cosponsored by the Centers for Disease Control and Prevention (CDC) and the Association of Public Health Laboratories (APHL).

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**Newborn Screening Quality Assurance Program
Sickle Cell Disease and Other Hemoglobinopathies**

Specimen and Lab Certification

Year: 2012 Panel: 2

Presumptive Clinical Phenotypes

	Specimen 212H1	Specimen 212H2	Specimen 212H3	Specimen 212H4	Specimen 212H5
Expected Presumptive Phenotype	FA	FAC	FAS	FA	FA
Accepted Presumptive Phenotypes	FA	FAC	FAS	FA	FA

Presumptive Clinical Assessments

	Specimen 212H1	Specimen1 212H2	Specimen 212H3	Specimen 212H4	Specimen 212H5
Expected Presumptive Clinical Assessment	01	03	02	01	01
Accepted Presumptive Clinical Assessments	01	03	02, 22	01	01

- 01 Normal--no abnormal Hb found
- 02 Hemoglobin S carrier
- 03 Hemoglobin C carrier
- 04 Hemoglobin SS disease (Sickle cell anemia)
- 05 Hemoglobin SC disease
- 06 Hemoglobin SD disease
- 08 Hemoglobin D carrier
- 09 Hemoglobin E carrier
- 12 Hemoglobin SE disease
- 16 Alpha-thalassemia (Bart's Hb)
- 18 Hemoglobin EE disease
- 21 Unsatisfactory sample. Specimen not evaluated (NE)
- 22 Unidentified variant, fast or aging band

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Sickle Cell Disease and Other Hemoglobinopathies**

Frequency Distributions

Year: 2012 Panel:2

Phenotypes			Clinical Assessments		
Specimen Number	Hemoglobin Phenotypes	Frequency Distributions	Specimen Number	Presumptive Assessments	Frequency Distributions
212H1	FA	66	212H1	01 Normal	66
	FAS	1*		02 Hemoglobin S carrier	1*
212H2	FAC	60	212H2	03 Hemoglobin C carrier	66
	FCA	6		01 Normal	1*
	FA	1*			
212H3	FAS	64	212H3	02 Hemoglobin S carrier	64
	FAD	1*		08 Hemoglobin D carrier	1*
	FS	1*		04 Hemoglobin SS disease	1*
	FA	1*		01 Normal	1*
212H4	FA	65	212H4	01 Normal	65
	FAC	1*		02 Hemoglobin S carrier	1*
	FAS	1*		03 Hemoglobin C carrier	1*
212H5	FA	66	212H5	01 Normal	66
	FAS	1*		02 Hemoglobin S carrier	1*

Note: An astrick (*) denotes a missed phenotype and or assessment.

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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