

# Newborn Screening Quality Assurance Program

## PROFICIENCY TESTING

## Sickle Cell Disease and Other Hemoglobinopathies

Volume 22, No. 3

Panel 3

November 2012

### INTRODUCTION

On October 1, 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 74 panels were mailed by overnight FedEx mail. The packages went to 50 domestic laboratories and 24 foreign laboratories. This PT report is a compilation of data reports received from 71 of the participating laboratories by the designated deadline date. There were 3 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 January 7, 2013. ❖

### MEETINGS AND TRAINING

7th Annual Sickle Cell Disease Research & Educational Symposium and National Sickle Cell Disease, Miami, Florida on April 14-17, 2013. ❖

### SPOTLIGHT

(June 18, 2012) A Chicago woman is the first Midwest patient to receive a successful stem cell transplant to cure her sickle cell disease without chemotherapy. The transplant technique is rel-

atively uncommon and is a much more tolerable treatment for patients with aggressive sickle cell disease who often have underlying organ disease and other complications, says Dr. Damiano Rondelli, professor of medicine at UIC, who performed Thomas's transplant.

The procedure initially allows a patient's own bone marrow to coexist with that of the donor. Since the patient's bone marrow is not completely destroyed by chemotherapy or radiation prior to transplant, part of the immune defense survives, lessening the risk of infection. The goal is for the transplanted stem cells to gradually take over the bone marrow's role to produce normal healthy red blood cells.

ScienceDaily. Retrieved November 13, 2012, from <http://www.sciencedaily.com/releases/2012/06/120618194714.htm>. ❖

### 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©3 organization that accepts donated cord blood for clinical use. ❖

CDC/APHL

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

***Specimen and Lab Certification***

Year: 2012 Panel: 3

**Presumptive Clinical Phenotypes**

	<b>Specimen 312H1</b>	<b>Specimen 312H2</b>	<b>Specimen 312H3</b>	<b>Specimen 312H4</b>	<b>Specimen 312H5</b>
<b>Expected Presumptive Phenotype</b>	FA	FAS	FA	FA	FA
<b>Accepted Presumptive Phenotypes</b>	FA	FAS	FA	FA	FA

**Presumptive Clinical Assessments**

	<b>Specimen 312H1</b>	<b>Specimen 312H2</b>	<b>Specimen 312H3</b>	<b>Specimen 312H4</b>	<b>Specimen 312H5</b>
<b>Expected Presumptive Clinical Assessment</b>	01	02	01	01	01
<b>Accepted Presumptive Clinical Assessments</b>	01	02	01	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
- 20 Assessment not listed
- 21 Unsatisfactory specimen
- 22 Unidentified variant, fast or aging band  
Specimen not evaluated (NE)

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**Frequency Distributions**

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Phenotypes			Clinical Assessments		
Specimen Number	Hemoglobin Phenotypes	Frequency Distributions	Specimen Number	Presumptive Assessments	Frequency Distributions
<b>312H1</b>	FA	70	<b>312H1</b>	01 Normal	70
	AF	1			
<b>312H2</b>	FAS	68	<b>312H2</b>	02 Hemoglobin S carrier	69
	AFS	1			
	FAU	1			
	FA	1 *			
<b>312H3</b>	FA	70	<b>312H3</b>	01 Normal	70
	AF	1			
<b>312H4</b>	FA	70	<b>312H4</b>	01 Normal	70
	AF	1			
<b>312H5</b>	FA	70	<b>312H5</b>	01 Normal	70
	AF	1			

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

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