



# Newborn Screening Quality Assurance Program

## PROFICIENCY TESTING

## Sickle Cell Disease and Other Hemoglobinopathies

Volume 20, No. 1

Panel 1

February 2010

### INTRODUCTION

On January 11, 2010, we distributed five dried-blood-spot (DBS) specimens prepared from umbilical cord bloods to all active participants for the Panel 1 Sickle Cell Disease and Hemoglobinopathy Proficiency Testing (PT) event. A total of 77 panels were mailed by overnight FedEx mail. The packages went to 50 domestic laboratories and 27 foreign laboratories. This PT report is a compilation of data reports received from 74 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 (Hbs) 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, therefore, the next shipment for the Hemoglobinopathy PT program will be on May 3, 2010. ❖

### MEETINGS

The National Conference on Blood Disorders in Public Health will convene March 9-12, 2010 at the Crowne Plaza Ravinia in Atlanta, GA. ❖

### SPOTLIGHT

New research at St. Jude Children's Research Hospital in Memphis, Tenn., has found that cholesterol drugs known as statins hold promise as a treatment for sickle cell disease. The findings, published February 1, 2010 in *J Clin Invest.* 2010;120(2):627-635, found that among mice with a type of sickle

cell disease, those treated with statins lived much longer after being infected with pneumococcal bacteria than those who didn't get the treatment. The cholesterol drugs appeared to stop cells from being killed by bacterial toxins and reduced the risk that germs would invade the blood system, according to the report. Children with sickle cell disease face a serious threat from pneumococcal bacteria, and the researchers think preventive treatment with statins may reduce their risk of infection. ❖

### 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: 2010 Panel: 1

**Presumptive Clinical Phenotypes**

	<b>Specimen 10H1</b>	<b>Specimen 10H2</b>	<b>Specimen 10H3</b>	<b>Specimen 10H4</b>	<b>Specimen 10H5</b>
<b>Expected Presumptive Phenotype</b>	FA	FAS	FAS	FAC	FA
<b>Accepted Presumptive Phenotypes</b>	FAV FA Fast	FAS	FAS	FAC	FAV FA Fast

**Presumptive Clinical Assessments**

	<b>Specimen 10H1</b>	<b>Specimen 10H2</b>	<b>Specimen 10H3</b>	<b>Specimen 10H4</b>	<b>Specimen 10H5</b>
<b>Expected Presumptive Clinical Assessment</b>	01	02	02	03	01
<b>Accepted Presumptive Clinical Assessments</b>	22	02	02	03	22

- 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
- 22 Unidentified variant, fast or aging band

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**Frequency Distributions**

Year: 2010      Panel: 1

**Phenotypes**

<b>Specimen Number</b>	<b>Hemoglobin Phenotypes</b>	<b>Frequency Distributions</b>	<b>Specimen Number</b>	<b>Presumptive Assessments</b>	<b>Frequency Distributions</b>
<b>10H1</b>	FA	68	<b>10H1</b>	01 Normal Hemoglobin	68
	FAV	4		22 Unidentified Variant	5
	FA Fast	1		02 Hemoglobin S carrier*	1*
	FAS*	1*			
<b>10H2</b>	FAS	73	<b>10H2</b>	02 Hemoglobin S carrier	73
	FS*	1*		04 Hemoglobin SS disease*	1*
<b>10H3</b>	FAS	73	<b>10H3</b>	02 Hemoglobin S carrier	73
	FS*	1*		04 Hemoglobin SS disease*	1*
<b>10H4</b>	FAC	72	<b>10H4</b>	03 Hemoglobin C carrier	73
	FAC Bart's	1		05 Hemoglobin SC disease*	1*
	FSC*	1*			
<b>10H5</b>	FA	68	<b>10H5</b>	01 Normal Hemoglobin	68
	FAV	4		22 Unidentified Variant	5
	FA Fast	1		02 Hemoglobin S carrier*	1*
	FAS*	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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