



# Newborn Screening Quality Assurance Program

## PROFICIENCY TESTING

## Sickle Cell Disease and Other Hemoglobinopathies

Volume 20, No. 2

Panel 2

June 2010

### INTRODUCTION

On May 3, 2010, 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 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 72 of the participating laboratories by the designated deadline date. There were 5 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. Consensus among the domestic laboratories was less than 80%, therefore, Specimen 20H4 was deemed not evaluated. Please refer to the frequency distribution report. This specimen was certified in our CDC laboratory as FAD by IEF, HPLC, and restriction fragment length polymorphism (RFLP-DNA based testing).

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 October 4, 2010. ❖

### MEETINGS

**July 20-23, 2010**, 1st Global Congress on Sickle Cell Disease, 1910-2010: 100 Years of Science, Still Seeking a Global Solutions, Accra International Conference Centre, Accra, Ghana

**September 21-24, 2010**, SCDA 38th Annual Convention, Gaylord National Hotel & Conference Center Washington D.C. [www.sicklecelldisease.org](http://www.sicklecelldisease.org)

**November 22-27, 2010**, Fourth International Congress 2010 Sickle Cell Disease International Organization sponsored by The Sickle Cell Disease International Organization in collaboration with Centre for Genetic Diseases & Molecular Biology Department of Biochemistry, Pt. J.N.M. Medical College, Raipur (C.G.) INDIA.

### 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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Direct inquiries to:  
Centers for Disease Control and Prevention (CDC)  
4770 Buford Highway, NE, MS/F43  
Atlanta, GA 30341-3724

Phone: 770-488-7897  
FAX: 770-488-4255  
E-mail: [NMeredith@cdc.gov](mailto:NMeredith@cdc.gov)

Editor: Nancy Meredith  
Production: Connie Singleton  
Sherri Zobel  
Sarah Brown



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

***Specimen Certification***

Year: 2010 Panel: 2

**Presumptive Clinical Phenotypes**

	<b>Specimen 20H1</b>	<b>Specimen 20H2</b>	<b>Specimen 20H3</b>	<b>Specimen 20H4</b>	<b>Specimen 20H5</b>
<b>Expected Presumptive Phenotype</b>	FAS	FA	FAC	Not Evaluated FAD	FA
<b>Accepted Presumptive Phenotypes</b>					

**Presumptive Clinical Assessments**

	<b>Specimen 20H1</b>	<b>Specimen 20H2</b>	<b>Specimen 20H3</b>	<b>Specimen 20H4</b>	<b>Specimen 20H5</b>
<b>Expected Presumptive Clinical Assessment</b>	02	01	03	Not Evaluated 08	01
<b>Accepted Presumptive Clinical Assessments</b>					

- 01 Normal Hemoglobin
- 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-not evaluated (NE)
- 22 Unidentified variant, fast, or aging band

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**Frequency Distributions**

Year: 2010      Panel: 2

Phenotypes			Clinical Assessments		
Specimen Number	Hemoglobin Phenotypes	Frequency Distributions	Specimen Number	Presumptive Assessments	Frequency Distributions
20H1	FAS	72	20H1	02 Hemoglobin S carrier	72
20H2	FA FAV	70 2*	20H2	01 Normal Hemoglobin 22 Unidentified variant, fast, or aging band	70 2*
20H3	FAC FCA	71 1	20H3	03 Hemoglobin C carrier	72
20H4	FAD FDA FAD/G FAV FADV FADB FAU FD FA F NE	33 1 2 9 1 1 1 2* 10* 1* 11	20H4	08 Hemoglobin D carrier  22 Unidentified variant, fast, or aging band  20 Assessment not listed 06 Hemoglobin SD disease 01 Normal Hemoglobin 04 Hemoglobin SS disease 21 Unsatisfactory specimen Not evaluated (NE)	37  11  1* 2* 10* 1* 10
20H5	FA FAV	70 2*	20H5	01 Normal Hemoglobin 22 Unidentified variant, fast, or aging band	70 2*

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**.

**CENTERS FOR DISEASE CONTROL AND PREVENTION (CDC)**  
ATLANTA, GA 30341

**Director**

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**Newborn Screening and Molecular Biology Branch**

Carla Cuthbert, Ph.D.

**Chief Emeritus**

**Newborn Screening and Molecular Biology Branch**

W. Harry Hannon, Ph.D.



**Contributors:** Barbara W. Adam  
Carol Bell  
Dana Chafin  
Paul Dantonio  
Victor R. De Jesus, Ph.D.  
Marie C. Earley, Ph.D.  
Elizabeth M. Hall  
L. Omar Henderson, Ph.D.  
Sharon Kerr  
Francis Lee, Ph.D.  
Lixia Li, Ph.D.  
Timothy Lim, Ph.D.  
Zuzheng (Roy) Luo  
Joanne Mei, Ph.D.  
Nancy Meredith  
Hien Nguyen  
Shannon O'Brien  
David Simms  
Sherri Stevens  
Robert Vogt, Ph.D.  
Golriz Yazdanpanah  
Hui Zhou, Ph.D.

**Production:** Sarah Brown  
Felicia Manning  
Teresa Moore  
Connie Singleton



**ASSOCIATION OF PUBLIC HEALTH LABORATORIES**  
SILVER SPRING, MD 20910

**President**

Susan U. Neill, Ph.D., M.B.A.

**Chairman, Newborn Screening and Genetics in Public Health Committee**

Cheryl Hermerath, M.B.A., DLM(ASCP), RM(NRCM)

**Chairman, Newborn Screening Quality Assurance Quality Control Subcommittee**

Gary Hoffman, B.S.

**INQUIRIES TO:**

*Nancy Meredith, Editor • Centers for Disease Control and Prevention (CDC)*  
*Newborn Screening Quality Assurance Program • Mailstop F-43*  
*4770 Buford Highway, N.E. • Atlanta, GA 30341-3724*  
*Phone (770) 488-4582 • FAX (770) 488-4255 • E-mail: NMeredith@cdc.gov*