Newborn Screening News

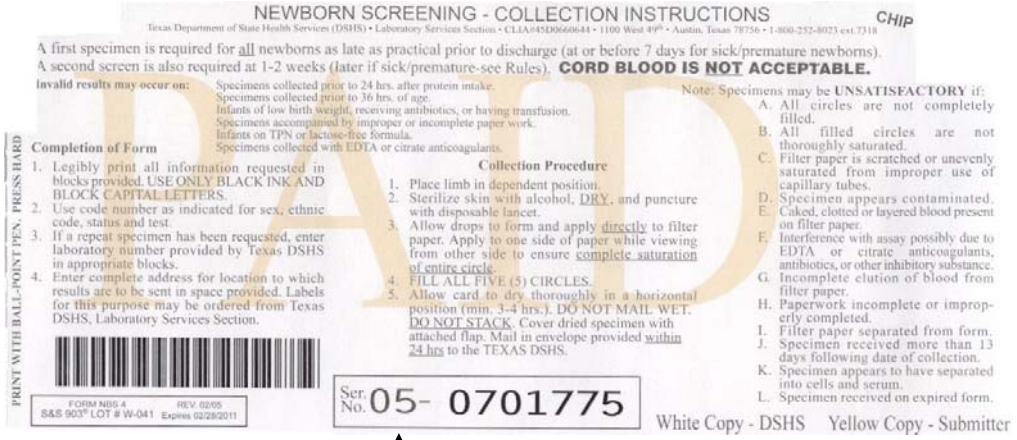
Volume 7, Issue 2 April 2006

REMINDER: ROTATE YOUR FILTER PAPER STOCK

USE NEWBORN SCREENING FORMS THAT BEGIN WITH 05- SERIAL NUMBERS BEFORE FORMS BEGINNING WITH 06- SERIAL NUMBERS.

The total number of unsatisfactory specimens received for January 2006 was 1,938 (2.9% of 66,616). The increase was due to the submission of specimens on expired 2004 forms. The number of expired forms rejected so far in 2006 is as follows: Jan – 1039; Feb – 241; Mar – 87.

It is very important to rotate stock when replenishing your supply of newborn screening kits. CLIA regulations require DSHS to discard any specimens received that have exceeded the expiration date. Submitting invalid specimens results in retesting and delays the screening of a newborn, placing a newborn at risk for delayed diagnosis of a screened condition.


NEWBORN SCREENING - COLLECTION INSTRUCTIONS

CHIP

Texas Department of State Health Services (DSHS) • Laboratory Services Section • CLIA#45D0660644 • 1100 West 49th • Austin, Texas 78756 • 1-800-252-8023 ext.7318

A first specimen is required for all newborns as late as practical prior to discharge (at or before 7 days for sick/premature newborns).
A second screen is also required at 1-2 weeks (later if sick/premature-see Rules). **CORD BLOOD IS NOT ACCEPTABLE.**

Invalid results may occur on:
Specimens collected prior to 24 hrs. after protein intake.
Specimens collected prior to 36 hrs. of age.
Infants of low birth weight, receiving antibiotics, or having transfusion.
Specimens accompanied by improper or incomplete paper work.
Infants on TPN or lactose-free formula.
Specimens collected with EDTA or citrate anticoagulants.

Note: Specimens may be **UNSATISFACTORY** if:
A. All circles are not completely filled.
B. All filled circles are not thoroughly saturated.
C. Filter paper is scratched or unevenly saturated from improper use of capillary tubes.
D. Specimen appears contaminated.
E. Caked, clotted or layered blood present on filter paper.
F. Interference with assay possibly due to EDTA or citrate anticoagulants, antibiotics, or other inhibitory substance.
G. Incomplete elution of blood from filter paper.
H. Paperwork incomplete or improperly completed.
I. Filter paper separated from form.
J. Specimen received more than 13 days following date of collection.
K. Specimen appears to have separated into cells and serum.
L. Specimen received on expired form.

PRINT WITH BALL-POINT PEN. PRESS HARD

Completion of Form
1. Legibly print all information requested in blocks provided. USE ONLY BLACK INK AND BLOCK CAPITAL LETTERS.
2. Use code number as indicated for sex, ethnic code, status and test.
3. If a repeat specimen has been requested, enter laboratory number provided by Texas DSHS in appropriate blocks.
4. Enter complete address for location to which results are to be sent in space provided. Labels for this purpose may be ordered from Texas DSHS, Laboratory Services Section.

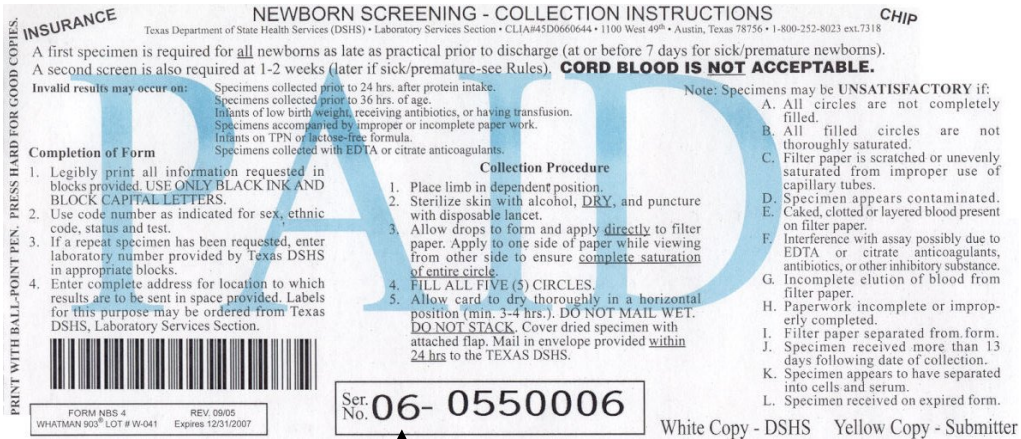
Collection Procedure
1. Place limb in dependent position.
2. Sterilize skin with alcohol, DRY, and puncture with disposable lancet.
3. Allow drops to form and apply directly to filter paper. Apply to one side of paper while viewing from other side to ensure complete saturation of entire circle.
4. FILL ALL FIVE (5) CIRCLES.
5. Allow card to dry thoroughly in a horizontal position (min. 3-4 hrs.). DO NOT MAIL WET. DO NOT STACK. Cover dried specimen with attached flap. Mail in envelope provided within 24 hrs to the TEXAS DSHS.

FORM NBS 4 REV. 02/05
S&S 903® LOT # W-041 Expires 02/28/2011

Ser. No. 05- 0701775

White Copy - DSHS Yellow Copy - Submitter

2005


INSURANCE

NEWBORN SCREENING - COLLECTION INSTRUCTIONS

CHIP

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FORM NBS 4 REV. 09/05
WHATMAN 903® LOT # W-041 Expires 12/31/2007

Ser. No. 06- 0550006

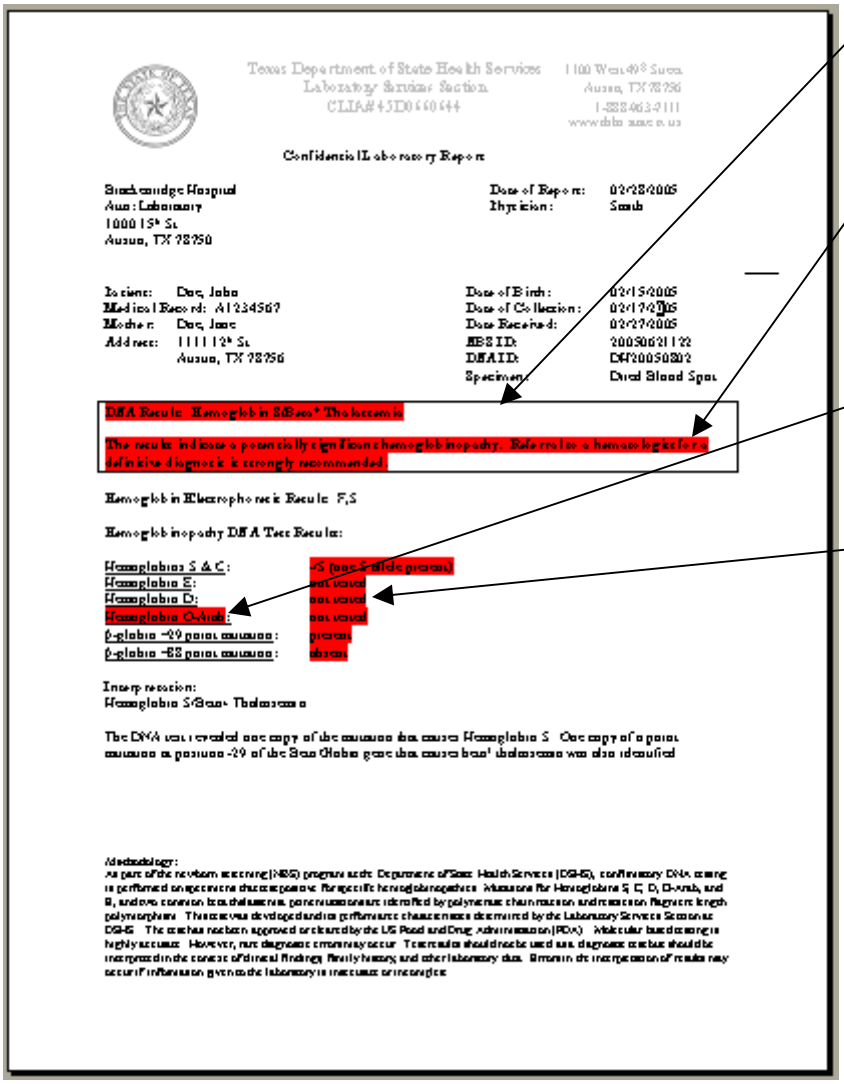
White Copy - DSHS Yellow Copy - Submitter

2006

Notice

DNA Report Format Change

The format of the Hemoglobinopathy DNA report has been modified to make it easier to read and understand. Please see below for highlights of the changes.



Texas Department of State Health Services
Laboratory Services Section
CLIA#45D0660644

Confidential Laboratory Report

Stockcandge Hospital
Attn: Laboratory
1000 15th St
Austin, TX 78750

Date of Report: 02/28/2005
Physician: Smith

Patient: Doe, John
Medical Record: A1234567
Mother: Doe, Jane
Address: 1111 12th St
Austin, TX 78756

Date of Birth: 02/15/2005
Date of Collection: 02/17/2005
Date Received: 02/27/2005
NBS ID: 20050621122
DNA ID: D#20050202
Specimen: Dried Blood Spot

DNA Result: Hemoglobin S/Beta+ Thalassemia

The results indicate a potentially significant hemoglobinopathy. Referral to a hematologist for a definitive diagnostic is strongly recommended.

Hemoglobin Electrophoresis Result: F,S

Hemoglobinopathy DNA Test Results:

Hemoglobins S & C:	+/S (one S allele present)
Hemoglobin E:	not tested
Hemoglobin D:	not tested
Hemoglobin O-Arab:	not tested
β-globin -29 point mutation:	present
β-globin -88 point mutation:	absent

Interpretation:
Hemoglobin S/Beta+ Thalassemia

The DNA test revealed one copy of the mutation that causes Hemoglobin S. One copy of a point mutation at position -29 of the Beta Globin gene that causes beta+ thalassemia was also identified.

Methodology:
As part of the newborn screening (NBS) program at the Department of State Health Services (DSHS), confirmatory DNA testing is performed on specimens that are positive for specific hemoglobinopathies. Mutations for Hemoglobins S, C, D, O-Arab, and E, and two common beta-thalassemias, point mutations are identified by polymerase chain reaction and restriction fragment length polymorphism. These assays developed under performance characteristics determined by the Laboratory Services Section at DSHS. The newborn screening test cleared by the US Food and Drug Administration (FDA). Molecular based testing is highly accurate. However, rare diagnostic errors may occur. Test results should not be used as a diagnosis and should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in the interpretation of results may occur if information given on the laboratory is inaccurate or incomplete.

- Easier to locate the DNA result.
- A statement of recommendation is included for any clinically significant result.
- Addition of Hemoglobin O-Arab variant in the mutation panel.
- Change of result descriptions. (see details below)

Result Descriptions		
Test	**Old**	**New**
S&C	+/+	-/- (no S or C alleles present)
	+/S	-/S (one S allele present)
	+/C	-/C (one C allele present)
	SS	SS (two S alleles present)
	SC	SC (one S allele, one C allele present)
	CC	CC (two C alleles present)
	not tested	not tested
E	+/+	-/- (no E allele present)
	+/E	-/E (one E allele present)
	EE	EE (two E alleles present)
	not tested	not tested
D	+/+	-/- (no D allele present)
	+/D	-/D (one D allele present)
	DD	DD (two D alleles present)
	not tested	not tested
O-Arab		-/- (no O allele present)
		-/O (one O allele present)
		OO (two O alleles present)
		not tested
-29	positive	present
	negative	absent
	not tested	not tested
-88	positive	present
	negative	absent
	not tested	not tested

Any questions? Contact the Newborn Screening State Laboratory 1-800-252-8023 ext. 7158