Newborn Screening News

Volume 8, Issue 2 **April 2007**

IMPORTANT MESSAGE!
DON'T DAMAGE/COVER BARCODE

The barcodes on the NBS collection forms **are** used by DSHS laboratory testing equipment. Do **NOT** damage the barcodes in any way. This includes punching holes, applying labels, stickers or tape or writing in the barcode area. Damaging the barcode may severely delay the screening process.

Newborn Screening Report Cards

The DSHS Laboratory Services Section will soon be providing Newborn Screening Report Cards to all facilities, physicians, and health care providers who submit newborn screening specimens. Receiving a properly collected specimen in a timely manner has always been critical, but even more so with the expanded panel. Fourteen disorders can be life threatening within the first week of life, and unsatisfactory or delayed specimens could lead to serious consequences for an affected infant.

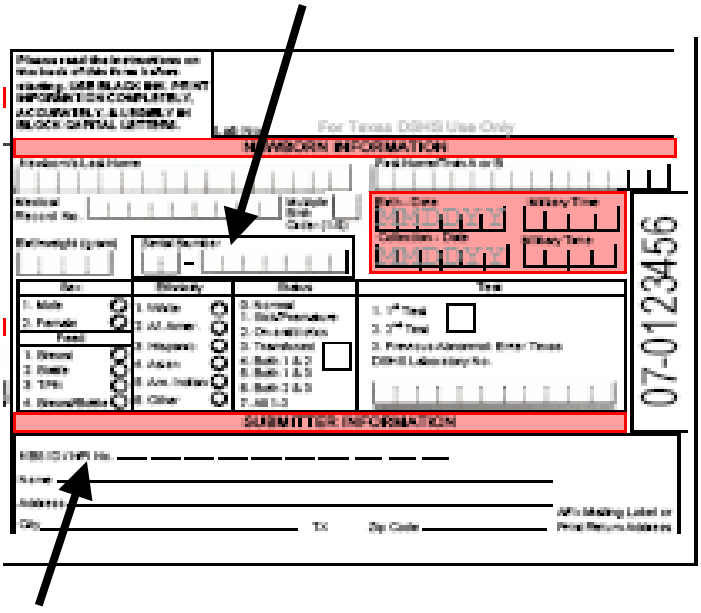
The report card will provide program statistics to submitters including: the number of newborn screens submitted, transit times from specimen collection to receipt in the DSHS laboratory, and the number of unsatisfactory specimens grouped by specific quality issues. Since each specimen card may have up to 3 unsatisfactory codes, a separate count is given with the total number of unsatisfactory codes. For program comparisons, report cards will also include statistics from other submitters with similar birth totals, and from all Texas submitters.

The initial report card will be for the period January – December 2006. Thereafter, reports will be sent on a quarterly basis. The report card is intended to be used as a tool to monitor and improve newborn screening procedures, such as specimen collection, demographic data entry, and processing. We hope that this will be a useful assessment tool for each facility. DSHS will use the data to identify sites that need assistance in order to provide educational tools or on-site training.

Specimen Collection: New Demographic Information

Serial Number

- The new specimen collection kits include a 'Parent Copy' as the first sheet, which contains the kit serial number. Birthing facilities should complete the information on this sheet and give it to the parent.
- Explain that the parent copy should be taken to the baby's next office visit, will help link the child's first and second screen results, and allows for quick notification of abnormal test results OR of a possible disorder.
- The physician performing the second screen should review the parent copy and write the initial kit's serial number in the designated area on the second screen kit.



NPI – National Provider Identification
The Administrative Simplification provisions of the *Health Insurance Portability and*

Accountability Act of 1996 (HIPAA) mandated the adoption of standard unique identifiers for health care providers, as well as the adoption of standard unique identifiers for health plans. The purpose of these provisions is to improve the efficiency and effectiveness of the electronic transmission of health information. The Centers for Medicare & Medicaid Services (CMS) has developed the **National Plan and Provider Enumeration System (NPPES)** to assign these unique identifiers.

If you are a **Health Care Provider**, the National Provider Identifier (NPI) is your standard unique identifier.

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Newborn Screening News
Newsletter of the Texas Newborn Screening Program

If you would like to receive additional copies of Newborn Screening News for your facility staff or satellite clinics, please send the following information to the Newborn Screening Program at newborn@dshs.state.tx.us or call 1-800-252-8023 ext. 2129 or fax to 512/458-7450.

Facility
Contact Person
Address
City, State, Zip Code

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Coming Soon
Genetic Testing for Galactosemia

The DSHS Laboratory Services Section is pleased to announce the DNA Analysis Laboratory will soon provide a molecular testing service to detect three common Galactosemia mutations, and a frequent variant in the galactose-1-phosphate uridyl transferase (GALT) gene. This service will be part of the Newborn Screening Program. It can also be used to determine carrier status of families with a diagnosed child.

Galactosemia is an inborn error of metabolism caused by a deficiency of the GALT enzyme. Each newborn in the State of Texas is screened for galactosemia by a fluorometric assay to detect GALT enzyme activity. Specimens with abnormally low GALT activities are reported to the Case Management program for follow-up. False positive results are often caused by environmental factors such as heat and humidity and the practice of batching during the specimen handling procedure. The Duarte-2 variant has a reduced GALT activity and may be detected by the screening test. The DNA testing for the Duarte variant will help to differentiate those babies that may not need immediate intervention.

Fee-for-service testing will also be available to provide genetic testing of the GALT gene in other individuals with Galactosemia or to determine carrier status in family members of the patients. Specimens will need to be submitted to the DSHS Laboratory Services by physician request.

The three Galactosemia mutations included in the testing panel are Q188R, S135L and K285N, accounting for about 68% of the Classical Galactosemia alleles in the United States (U.S.) population. The testing panel includes the Duarte-2 variant (the N314D mutation) that is present in 5% of the general U.S. population and reduces activity of the GALT enzyme by ~25%. The results will be reported back to the submitters as homozygous, heterozygous, or negative for each of the four mutations/variant tested and should be interpreted in the context of the patient's GALT biochemical phenotype.

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If you have any questions, please contact the DNA Analysis Laboratory at 512-458-7158.

TEXAS Department of State Health Services

To order free educational materials from the Newborn Screening Program for patients and specimen collection information for submitters, please call 1-800-252-8023, ext. 2129 or order online:
http://www.dshs.state.tx.us/newborn/pubs.shtm

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