Newborn Screening News

Volume 2, Issue 4 **October 2001**

What's New?

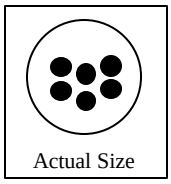
New Laboratory Methodology Implemented for newborn screening

Specimens Involved: The PekinElmer™ technology will be used only for the Newborn Screening (NBS) collection card specimens (including 1st test - under 7 days, 1st test - 7 days and older, 2nd test, and requested repeats). All confirmatory and monitoring specimens received with the G-127 and G-128 forms will continue to be tested as they have in the past.

Changes You Will See: Reporting ranges for Phenylalanine (PKU) will be: Normal, Slightly Elevated, Moderately Elevated, and Very Elevated.

What YOU Can Do To Help: Completely fill out one circle at a time on the NBS collection card

Why?: to enable the lab to get 6 dots punched from one circle (see diagram)



Reason for extra circles: to provide specimen for repeat testing when abnormal results are found prior to release of the final results.

Number of Circles Needed: 5 circles completely filled to ensure that we can conduct all required testing.

Methodology Descriptions For the Technically Oriented Submitter

(excerpts taken from the PerkinElmer™ Life Sciences assay kit insert procedures)

Phenylalanine: The Wallac Phenylalanine Test is a modification of the fluorometric procedure published by McCaman and Robins in 1962. The assay is based on the enhancement of the flourescence of a phenylalanine-ninhydrin reaction product by the dipeptide, L-leucyl-L-alanine. This method measures phenylalanine quantitatively in the presence of other amino acids.

Galactose: The Wallac Neonatal Total Galactose Test is fluorometric assay that simultaneously measures galactose and galctose-1-phosphate.

Thyroxine *(T_4):* The AutoDELFIA Neonatal T_4 assay is a solid phase, time-resolved fluoroimmunoassay based on the competitive reaction between europium-labeled T_4 and sample T_4 for a limited amount of binding sites on T_4 specific monoclonal antibodies.

TSH*:* The AutoDEFLIA Neonatal hTSH assay is a solid phase assay based on the direct sandwich technique in which two monoclonal antibodies are directed against two separate antigenic determinants on the hTSH molecule.

17-A-OH-progesterone: The AutoDEFLIA Neonatal 17-A-OH-progesterone (17-OHP) assay is a solid phase, time-resolved fluoroimmunoassay based on the competitive reaction between europium-labeled 17-OHP and sample 17-OHP for a limited amount of binding sites on 17-OHP specific polyclonal antibodies.

TDH Stock # 05-10897

The Story on How This All Happened

Ten years ago, the Newborn Screening (NBS) Laboratory began dreaming, researching, and planning to update the technology used to analyze the battery of NBS diseases. On July 2, 2001 two 53-foot semi-tracter trailer trucks arrived. The first truck arrived at 8:00 am and was loaded with instrumentation from PerkinElmer™ Life Sciences. The instrumentation included 5 AutoDEFLIAs (2 more arrived later in July), 2 Victors, 2 Isoteks, some incubators, shakers, 6 multi-punch punching machines (2 more arrived in August), and lots of computer equipment, including CPU's, monitors, and printers. Additional equipment continued to arrive throughout the months of July and August. At 1:00 pm the second truck arrived loaded with refrigerated reagents to be used for the analysis of phenylalanine, galactose, T_4, TSH, 17-" OHP, and GALT. Due to the size of the trucks all the equipment and reagents had to be delivered to the main warehouse and then transported across the parking lot to the laboratory. On a 100° day in Austin there was great urgency to keep all refrigerated reagents cold. All available lab technicians helped transport, unpack, date, and store these critical reagents in a minimal amount of time. What a sight! What a staff!

During July and August the PerkinElmer™ staff was on-site to install equipment, verify functionality, and to train the Texas Department of Health (TDH) NBS staff. Once trained the NBS staff worked to validate the instruments and methodologies. In addition, criteria for repeat testing, abnormal cutoffs, and reporting were determined. Although the methodology for testing hemoglobinopathies will continue to be Isoelectric Focusing (IEF), the use of microtiter plates for all analytes has required some adjustments in the specimen to gel application and gel reading procedures. As with any new test or piece of equipment, problems were encountered, some minor and some major. The TDH NBS staff and PerkinElmer™ staff addressed all problems to find workable solutions.

Testing with the new technology began with specimens which were processed by laboratory specimen acquisition on Julian Date 235 (August 23, 2001).

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Contact Information: The NBS Case Management Staff will continue to contact submitters with instructions on how to follow-up abnormal screening results. As we become more familiar with the new testing equipment and methodologies, we will continue to fine-tune the analysis procedures. As you, the submitters, hospitals, doctors, and clinics, begin to receive results from the new technology please feel free to contact the TDH NBS Laboratory with comments and questions at the following telephone numbers:

1-800-422-2956
press 1 for English
press 3 for the Newborn Screening Program
press 3 for the Newborn Screening Laboratory Services
press 5 for Technical Information
press 1 for Newborn Screening

or **1-512-458-7318** (Texas Department of Health Laboratory)
press 1 if you know your party's extension
press 7333 for the Newborn Screening Laboratory

TDH

For free literature about the Texas Newborn Screening Program, call **1-800-422-2956**.
Or order online at **www.tdh.state.tx.us/newborn/pubs.htm**