



# Newborn Screening News

Volume 4, Issue 4

December 2003

## HISTORY OF TEXAS NEWBORN SCREENING

Dr. Robert Guthrie (1916-1995) developed the test for screening newborns for phenylketonuria (PKU). His research was inspired by his son and his niece. The son had non-PKU mental retardation and his niece, born in 1958, was diagnosed with PKU at the age of 17 months. Dr. Guthrie developed the test in the early 1960s and lobbied for mandatory screening of PKU. The Guthrie test, used throughout the U.S., opened the door to screening for and prevention of mental retardation in other disorders.

The Texas Department of Health (TDH) began screening newborns for PKU in 1965. Galactosemia was added in 1979 followed by hypothyroidism in 1980, hemoglobinopathies in 1983 and congenital adrenal hyperplasia (CAH) in 1989. The two inborn errors of metabolism, phenylketonuria (PKU) and galactosemia, are treated by diet. The endocrine disorders, congenital hypothyroidism and congenital adrenal hyperplasia (CAH), are treated with medication (hormone replacement therapy). Sickle cell disease complications may be prevented through a program of medical supervision and antibiotics administered at an early age.

Texas law [TAC §37.55(a)] states the physician or non-physician attending a newborn has the primary responsibility for causing the screening tests to be performed - and that a satisfactory and valid blood specimen be submitted to TDH on a properly completed filter paper collection form. The TDH Laboratory performs the newborn screening test series on specimens received from more than 4,000 locations around the state. **Approximately 3,500 specimens arrive in the mail each day and each specimen is visually inspected to determine if it is suitable to test.**

## INVALID SPECIMENS

The leading cause of specimen rejections in Texas is that the specimens are too old upon receipt by the Newborn Screening Laboratory in Austin. Prompt submission of newborn screening specimens is important for rapid detection, diagnosis, and treatment of affected infants.

Other leading causes of invalid specimens include:

- \* **Specimen quantity insufficient for testing.**
- \* **Specimen appears scratched or abraded.**
- \* **Specimen not dry before mailing.**
- \* **Specimen appears supersaturated.**
- \* **Specimen appears diluted, discolored or contaminated.**
- \* **Specimen exhibits serum rings.**
- \* **Specimen appears clotted or layered.**

The Texas Newborn Screening Laboratory is available to answer any questions you may have regarding specimen collection. Please feel free to call us at 1-800-422-2956 ext. 7333. You may also request specimen collection posters and collection guides at ext. 2129.

## It's Not Just PKU

There has been much confusion among doctors, hospitals and parents when newborn screening tests are called "PKU tests" or "PKU's".

Parents are upset unnecessarily for fear their child has PKU, when in fact, a routine repeat filter for another disorder has been requested.

**Please make every effort to call the test a "newborn screening test" rather than a "PKU".**

**Texas Department of Health  
Newborn Screening Program  
Publications Order Form**

Please enter quantity desired in box beside each item.

**Newborn Screening General Information for Parents and Consumers**

	Newborn Screening: For Your Baby's Health (Brochure)
	Screening para los recién nacidos: Para la salud de su bebé (Brochure)
	Newborn Screening Poster Size=16"x20" (English/Spanish- Front/Back)
	Second Screen Reminder (Refrigerator Magnet)
	Los bebés necesitan el segundo examen (1 man para el refrigerador )

**Specimen Collection for Health Professionals**

	Newborn Screening Specimen Collection Guide (English Only)
	Newborn Screening Program Practitioner's Guide (English Only)
	Simple Spot Check - - (English) 8? x 11 poster
	Simple Spot Check - - (English) 11 x 17 poster
	Prueba Selectiva Sencilla - - (Spanish) 8? x 11 poster
	Neonatal Screening: Blood Specimen Collection and Handling Procedure -- (English) 8? x 11 poster
	Neonatal Screening: Blood Specimen Collection and Handling Procedure -- (English) 11 x 17 poster
	Procedimiento de toma y manipulacion de muestras de sangre para pruebas en recién -- (Spanish) 8? x 11
	Weight Conversion Chart - - Pounds and Ounces to Grams

**Please complete:**

Facility:
Street Address:
City/State/Zip:
Attn:
Phone:

**Fax your order to: 512/ 458-7450      Attn: Outreach Education**

**Mail to:**            Newborn Screening Program  
                          Texas Department of Health  
                          1100 West 49<sup>th</sup> Street  
                          Austin, Texas 78756

If you wish to order online, please go to <https://www.tdh.state.tx.us/newborn/pubs.htm>  
 Or call us toll-free at 1-800-422-2956 extension 2129.