



Newborn Screening News

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URGENT NOTICE:

Specimen are UNACCEPTABLE and will be REJECTED when:

- * received WITHOUT a date of collection
- * received on EXPIRED collection forms

These guidelines must be followed to ensure the quality of test results and compliance with the Clinical Laboratory Improvement Amendments (CLIA) of 1988.

Unacceptable Specimen Quality:

The Newborn Screening Laboratory receives some blood spot specimens in a condition unacceptable for testing. Collection of a satisfactory specimen is the responsibility of the person attending the birth. Certain types of specimens are known to give invalid results including old specimens, those with incompletely filled, abraded, discolored, diluted or clotted spots, those showing serum "rings" and those collected with EDTA or sodium citrate anticoagulant. In these cases the newborn screening report will state "UNSATISFACTORY - PLEASE RESUBMIT". Submitting invalid specimens results in the inconvenience of retesting and delays the screening of the newborn, placing the newborn at risk for delayed diagnosis of a screened condition. **IN CASES OF UNSATISFACTORY SPECIMEN REPORTS THE INFANT MUST BE RE- SCREENED AS SOON AS POSSIBLE (even if only one test is reported unsatisfactory).**


General submission guidelines for all specimen types submitted for testing can be found at our website:
http://www.dshs.state.tx.us/lab/MRS_intro.shtm.
 If we can be of any assistance to you in interpreting these guidelines, please contact us at 1-888-963-7111, ext. 7333.

A descriptive poster illustrating invalid specimens is available free of charge by contacting the Newborn Screening Program at 1-800-252-8023, extension 2129.



SUMMER HEAT

Did you know that the hot weather during our Texas summers can damage newborn screening specimens? Exposure to heat and/or humidity can affect the specimens two ways: (a) the blood may "bake" onto the filter paper making it difficult to dissolve blood off the filter paper for testing and (b) the heat may cause degradation of substances in the blood, such as GALT, and adversely affect test values. Specimens that have been affected by heat or humidity may erroneously test as abnormal. When heat/humidity exposure occurs during mail transport, there is little that a submitter can do to prevent the situation. However, your facility should review your own specimen handling practices to be certain that specimens are not exposed to a hot environment prior to mailing. Mail newborn screening specimens within 24 hours of collection. Specimens should never be left in a hot car before delivering to a post office or placed in direct sunlight near a hot window. **DO NOT** place specimens in an outdoor metal mailbox. By eliminating risks for heat exposure, you may be able to prevent incorrect abnormal specimen results and avoid the need for specimen re-collection.



To order free education 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.tdh.state.tx.us/newborn/pubs.htm>

PHENYLKETONURIA (PKU)

Laboratory Tests

The DSHS screening test for PKU is a discrete fluorometric assay that measures the level of phenylalanine. Abnormal results are reported using the following qualitative statements: slightly elevated, moderately elevated, and very elevated. Confirmatory serum phenylalanine may be obtained from the DSHS NBS laboratory. Tests to monitor phenylalanine levels on diagnosed PKU individuals are available on serum or whole blood collected on special filter paper cards. Confirmatory serum phenylalanine and monitoring phenylalanine results are reported as concentrations in mg/dL.

In the past, confirmatory serum phenylalanine and its metabolic product, tyrosine, could be obtained from the DSHS laboratory. Confirmatory serum phenylalanine will continue to be provided by DSHS however, **tyrosine levels will no longer be offered**. Additionally, phenylalanine confirmatory and diet monitoring results are now electronically printed for mailing.

Purpose of Phenylalanine Confirmatory and Diet Monitoring Program


Patients with PKU obtain their health care, including immunizations and routine health problems, from their local physician. After the initial diagnosis of PKU has been made, the metabolic center staff is responsible for the ongoing care for the patient's PKU. The metabolic clinic staff educates the family on PKU, provides counseling including the genetics of PKU and dietary management. The physician and nutritionist set up a schedule for periodic blood tests to monitor the patient's serum phenylalanine level and diet intake throughout life.


Even though the patient is seen frequently at the metabolic center, it will also be necessary for the patient to be seen by the primary care physician or designee more frequently for blood tests. In the first year of life, it is usually necessary for a child with PKU to be seen every week for a blood draw and growth charting. After the first birthday, it is usually necessary for the child to be seen monthly until age 5. Since the care of every child with PKU is individualized the metabolic center will coordinate care with the


primary care physician to determine the appropriate management for the child.


Services Available for PKU Patients


The following services are available upon request of a physician for any resident of Texas who is diagnosed through the Newborn Screening Program of the Health Screening Branch, Texas Department of State Health Services (DSHS).


 **Medical consultation is available by referral.** The Metabolic Centers in Texas specialize in metabolic disorders and accept referrals of PKU patients for metabolic evaluation and follow-up care. Because of the complicated nature of controlling a PKU diet, DSHS strongly recommends referral in all cases.

 **Direct nutritional care** for the patient is usually provided by the metabolic center. This service includes regular monitoring of the diet prescription and conferences with parents.

 **Biochemical monitoring of serum phenylalanine** is available through the DSHS Newborn Screening Laboratory.

 **PKU blood collection kits** can be provided by DSHS, upon request, which include mailing containers and laboratory forms. Call 1-888-963-7111 ext. 7661 for more information.

 **PKU formula assistance** may be available if the person with PKU is a resident of the State of Texas and not eligible for private insurance, CSHCN, Medicaid, WIC, or CHIP.

 **Educational materials** are available for the physician, parents, and family of a child with PKU.

If you would like to request any of the above services for your patient please contact:

Newborn Screening Program M-555
Health Screening Branch
Texas Department of State Health Services
1100 West 49th Street
Austin, Texas 78756
MC 1918
1-800-252-8023