Newborn Screening News

Volume 7, Issue 3 July 2006

Specimen Collection Form Expiration Date
Important Information:

Specimens received on "05-" cards that are collected after 12/31/2006 will be **REJECTED**.
The 2005 forms were printed with the **wrong Date of Expiration**. Any form with a **serial number beginning with "05-" will expire on Dec. 31, 2006.** Use these forms first.

The **first two digits** of the **form serial number** are the **Year of Manufacture**.

Specimen Collection Forms Expire Two Years following manufacture.

Year of Manufacture	Date of Expiration
2005	12/31/2006
2006	12/31/2007

Specimens collected on Expired Forms are **REJECTED**.

Newborn Screening Expansion

House Bill (HB) 790 of the 79th Legislature required the Department of State Health Services (DSHS) to expand the number of newborn screening (NBS) tests. It also required DSHS to conduct a study to determine whether it is more cost-effective to contract with a private vendor to conduct the newborn screening tests or to conduct them at the state laboratory.

A six-member team evaluated the two proposals submitted by vendors in response to the Request for Proposal issued by DSHS. The evaluation team, which included two national (NBS) experts and staff from the Texas Health and Human Services Commission, concluded unanimously that it is more cost-effective for the state laboratory to conduct the newborn screening tests.

DSHS will increase the current screening panel to 28 (including hearing) of the tests recommended by the American College of Medical Genetics and the March of Dimes.

Quick Reference To Proposed Panel of Newborn Screening Disorders

Biotinidase Deficiency (BIO) BIO is an enzyme deficiency that occurs in about 1 in 60,000 newborns and can result in seizures, hearing loss, and death in severe cases. Treatment is simple and involves daily doses of biotin. (1)

Congenital Adrenal Hyperplasia (CAH) CAH is caused by decreased or absent production of certain adrenal hormones. The most prevalent type is detected by newborn screening in about 1 in 15,000 newborns. Early detection can prevent death in boys and girls and sex misassignment in girls. Treatment involves lifelong hormone replacement therapy. (1)

Congenital Hypothyroidism (CH) Inadequate or absent production of thyroid hormone results in CH and is present in about 1 in 3,500 newborns. Thyroid hormone replacement therapy begun by 1 month of age can prevent mental and growth retardation. (1)

Galactosemia (GAL) Failure to metabolize the milk sugar galactose results in GAL and occurs in about 1 in 50,000 newborns. The classical form detected by newborn screening can lead to cataracts, liver cirrhosis, mental retardation and/or death. Treatment is elimination of galactose from the diet usually by substituting soy for milk products. (1)

Homocystinuria (HCY) HCY is caused by an enzyme deficiency that blocks the metabolism of an amino acid that can lead to mental retardation, osteoporosis and other problems if left undetected and untreated. The incidence is approximately 1 in 350,000 U.S. newborns. Treatment may involve dietary restrictions and supplemental medicines. (1)

Maple Syrup Urine Disease (MSUD) MSUD is a defect in the way that the body metabolizes certain amino acids and is present in about 1 in 200,000 U.S. newborns. Early detection and treatment with dietary restrictions can prevent death and severe mental retardation. There is an increased risk in Mennonites. (1)

TEXAS Department of State Health Services

Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency The most common disorder in the way the body metabolizes fatty acids is called MCAD deficiency. Undetected, it can cause sudden death. Treatment is simple and includes ensuring regular food intake. The incidence from newborn screening is not yet known, but is thought to be approximately 1 in 15,000 newborns. (1)

Phenylketonuria (PKU) An enzyme defect that prevents metabolism of phenylalanine, an amino acid essential to brain development, is known as PKU and occurs in approximately 1 in every 19,000 U.S. newborns. Undetected and untreated with a special diet, PKU leads to irreversible mental retardation. Persons of European descent are at increased risk. (1)

Sickle Cell Disease (SCD) Sickle cell anemia (Hb-SS-Disease) is the most prevalent SCD and causes clogged blood vessels resulting in severe pain and other severe health problems. Other common SCDs include Hb-SC-Disease and various thalassemias. Newborn screening detects about 1 in 2,500 newborns with SCD annually. Persons of African or Mediterranean descent are at an increased risk. (3)

Tyrosinemia (TYR1) People with tyrosinemia have problems breaking down an amino acid called tyrosine from the food they eat. If not treated, the condition causes severe liver disease and other serious health problems. Treatment consists of medication and a diet low in tyrosine. The estimated incidence is 1 case in every 100,000 live births. (1)

Other Fatty Acid Oxidation (FAO) Disorders include Carnitine Uptake Defect (CUD), Long-Chain Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD), Trifunctional Protein Deficiency (TFP) and Very-Long-Chain Acyl-Co A Dehydrogenase Deficiency (VLCAD) Besides MCAD deficiency, other FAO disorders may be detected through newborn screening. They are usually described in categories based on the length of the fatty acid involved. Undetected and untreated they can cause seizures, coma, and even death. The incidences of various FAO disorders are not known since it is only recently that early detection through newborn screening has occurred. (4)

Organic Acid (OA) Disorders include 3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC), Beta-Ketothiolase Deficiency (BKD), Glutaric Acidemia Type I (GAI), Hydroymethylglutaric Aciduria (HMG), Isovaleric Acidemia (IVA) Methylmalonic Acidemia(MMA) (Cbl A and Cbl B forms) (Cbl A,B), Methylmalonic Acidemia (mutase deficiency form) (MUT), Multiple Carboxylase Deficiency (MCD) and Propionic Acidemia (PROP) Organic acidemias are a group of metabolic disorders that lead to accumulation of organic acids in the blood and urine and may be detected in newborn screening through analysis of acylcarnitine profiles. Symptoms can be diminished by restricting protein in the diet and supplementation with vitamins and/or carnitine. Because newborn screening for these disorders is relatively new, the degree of occurrence in newborns is not yet known. (9)

Urea Cycle Disorders (UCD) include Argininosuccinic Acidemia (ASA) and Citrullinemia (CIT) A UCD is a genetic disorder caused by a deficiency of one of the enzymes responsible for removing ammonia from the blood stream. Some UCDs may be detected as a part of newborn screening. They are characterized by seizures, poor muscle tone, respiratory distress, and coma, and result in death if left undetected and untreated. Because newborn screening for these disorders is relatively new, the degree of occurrence in newborns is not yet known. (2)

NEWBORN SCREENING PROPOSED RULES

Proposed rules for the Newborn Screening Program are available for a 30-day public comment period beginning July 8, 2006. The rules are located at the *Texas Register* web site:

http://www.sos.state.tx.us/texreg/issues.shtml

NEW DESIGN OF SPECIMEN COLLECTION FILTER PAPER

The newborn screening program plans to add an additional page to the specimen collection form. This first page of the collection kit will be a “parent copy” that states the basic purpose of newborn screening and contains directions for the parent and submitter. The directions will explain that the “parent copy” of the first newborn screen is to be taken by the parent to the baby’s doctor. The barcode label from it will be placed on the second NBS demographic information page when the 2nd specimen is collected. This is to allow the 1st and 2nd specimens to be linked in the laboratory.

The new form will also include a few new demographic information fields, such as:

- Time of Birth
- Time of Specimen Collection
- Primary Care Physician Name
- Primary Care Physician Address
- Primary National Provider Identification Number

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Questions can be addressed to newborn@dshs.state.tx.us or by calling 1-800-252-8023.

Publication Number 05-10897
www.dshs.state.tx.us/newborn/