Newborn Screening News

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TEXAS NEWBORN SCREENING SYMPOSIUM

In March 2004, the Newborn Screening Case Management Program (NBS), NBS Lab, the March of Dimes and the National Laboratory Training Network co-sponsored three symposia on Texas Newborn Screening.

This program is appropriate for laboratory and nursing personnel involved with newborn screening specimen collection and testing.

Program Description
A key component to ensuring healthy babies is the collection and testing of neonatal specimens. Newborn screening testing performed at the Texas Department of Health helps to identify abnormalities in babies that, left untreated, would become catastrophic problems. Early diagnosis and medical intervention can prevent many or all of the serious clinical symptoms. The symposium was designed to help participants learn about accurate specimen collection and the impact it has on the testing process and, ultimately, the health of Texans.

Objectives
At the conclusion of the symposium, participants would be able to:

* Identify screening tests performed on infants born in Texas
* State the incidence of disease in the population of Texas
* Properly complete the information on the specimen form
* Summarize problems associated with incorrect or incomplete information that accompanies a screening specimen
* Explain the proper collection process for a neonatal screening sample
* Identify causes of unsatisfactory specimens and explain the error that caused the problem
* Describe steps taken by the Texas Department of Health to assure quality testing results
* Distinguish between a normal and abnormal screening test result
* Relate the follow-up process that occurs when an abnormal screening result is obtained
* Discuss the impact of untreated disease in children with genetic abnormalities

The Newborn Screening Symposium had 48 attendees in San Antonio, 25 in Dallas and 41 in Houston. Each participant was asked to complete an evaluation form of the program. Due to the positive comments we received, we hope to continue these symposia in other areas of the state if funding becomes available.

Literature on specimen collection can be ordered, free of charge, through the Newborn Screening Case Management Program by calling 1-800-422-2956 extension 2129.

Specimen Collection Materials

* Practitioner's Guide
* Specimen Collection Guide
* Neonatal Screening Specimen Collection and Handling Procedure (posters 8½ x 11 & 11 x 17)
* Simple Spot Check (posters 8½ x 11 & 11 x 17)
* Weight Conversion Chart
* Slide Presentation (CD-ROM)

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www.tdh.state.tx.us/newborn/newborn.htm

SPOTLIGHT ON PKU
(Phenylketonuria)

Phenylketonuria (fennel-key-ton-uria) is often called PKU for short. PKU is a condition some children have that affects their ability to properly use protein. The problem concerns one particular part of protein, the amino acid called phenylalanine ("phe" for short). When a person eats foods containing protein, enzymes break down the protein into separate amino acids as building blocks for body growth and repair. In PKU, one of the enzymes does not function properly, the one needed to convert "phe" into another amino acid, tyrosine. As a result, "phe" accumulates in the blood and other parts of the body. The excess "phe" prevents the brain from growing and developing normally. It also causes other problems such as skin rash, excessive restlessness, irritable behavior and a musty body odor.

HOW IS IT TREATED?
At the present time, a diet low in "phe" is the only treatment for PKU. If the diet is started early enough and closely followed, the child's development will be normal in almost all cases. Some "phe" is essential for growth. Too much "phe" is harmful. The diet must be carefully planned to allow enough "phe" for the child to grow normally, yet not enough to produce the harmful effects of excessive "phe". This balance between too much and too little "phe" is different for each child. A child's needs depend on the severity of the enzyme deficiency and the child's age, growth rate, and current state of health. The right amount of "phe" for the child is determined through blood tests that measure the amount of "phe" in the child's blood. The diet prescription is adjusted accordingly by the physician and nutritionist. Several special formula-milk substitute products are available. These special formulas make it possible to plan a diet that is low in "phe" but adequate in protein, calories, and essential vitamins and minerals. In addition to special formulas, the child with PKU can have foods that are low in protein and "phe" in measured amounts. This includes most fruits and vegetables, some cereals and candies and special breads, cookies and pastas. All high-protein foods such as milk, meat, eggs and cheese, which contain large amounts of "phe", must be completely eliminated from the diet. This diet record, with the exact kinds and amounts of food eaten, will need to be kept just before each monitoring blood test. The nutritionist will use this record to decide what changes, if any, need to be made in the diet prescription.

HOW DOES ONE GET PKU?
The child acquires PKU by inheriting it from his parents. Inherited characteristics such as eye color, hair color and PKU are carried on special parts of the cell called genes. Everyone carries four or five different abnormal genes without showing signs of any of the disorders. PKU occurs in a child who has two genes for PKU, one inherited from his father and the other from his mother. It is estimated that one in every 70 persons is a carrier for PKU and that the disorder affects one in every 15,000 to 20,000 infants born in the United States.

SUMMARY
The severe mental retardation and other problems associated with PKU can be prevented by early diagnosis and proper diet control. The success of diet control requires the cooperation of everyone who comes in contact with the child. These efforts are without question, well rewarded.

The Texas Department of Health provides, without charge to residents of Texas who are diagnosed with PKU, blood specimen analysis through the Texas Department of Health Laboratory in Austin.

NEW LABORATORY CUTOFFS FOR PKU
Following a year of reporting an increased rate of PKU presumptive positive results and no foreseeable change in the analytical methodology, the laboratory reviewed the analytical data to determine if a change in cut-off would be appropriate.

It is our goal to return to the 2001-2002 presumptive positive rate of 0.03%. This would be an average of approximately 20 specimens/month.

The cutoff for presumptive positive specimens was increased slightly on April 1, 2004. This should result in fewer false positives.

CLARIFICATIONS FROM THE NEWBORN SCREENING LABORATORY

Patient Demographic Information
On the 2004 NBS specimen collection card the new field of “Birth Order” must be limited to the current pregnancy. Therefore a singleton birth indicates a “1” in this field and a multiple birth indicates “1”, “2”, “3”, etc. as appropriate for the birth order for the multiple.

Number of Blood Circles to Fill
When collecting the blood into the specimen filter circles the recommended number of circles to fill is all five (5). It is most important to **completely** fill one circle at a time, ensuring that the blood has absorbed all the way through the filter to the other side. **Do not** apply blood to both sides of the filter. Five filled circles is not mandatory because three (3) well collected circles is better than five (5) poorly collected circles.