Alabama expands newborn screening

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The Alabama Newborn Screening Program began screening infants for an additional inheritable disorder on April 5 when it initiated screening for Biotinidase Deficiency.

State law requires every facility that delivers babies to screen all infants for specific metabolic and inheritable disorders. This is important because a baby with one of these rare illnesses may appear healthy at birth, but irreparable damage can occur by the time symptoms become visible. The Alabama Department of Public Health's Bureau of Clinical Laboratories conducts all screening tests for the approximately 60,000 infants born yearly in the state.

The Alabama Newborn Screening Program began in 1969 when phenylketonuria, or PKU, testing began. In the next quarter century, additional tests became available and were required to screen infants for Hypothyroidism, Hemoglobinopathies, Galactosemia and Congenital Adrenal Hyperplasia.

Later this year, the scope of the Alabama Newborn Screening Program will expand by utilizing tandem mass spectrometry. This technology allows screening for at least 30 different inborn errors of metabolism in a single process using the dried blood spot specimen routinely collected for newborn screening. Testing for these disorders will begin as soon as pilot studies are completed.

Dr. Tom Miller, director of the Bureau of Family Health Services, said, "One of the most phenomenal abilities of tandem mass spectrometry is that this technology will assist in detecting these diseases presymptomatically in children. Early diagnosis allows for early intervention. Many of these children would become profoundly disabled or suffer an early death if not diagnosed in the newborn period."

With the addition of Biotinidase, amino acid, organic acidemia, and fatty acid oxidation disorders, Alabama will become one of the few states that screen for all of the nine disorders, as well as hearing loss, that are recommended by the March of Dimes. Therapy for disorders detected through newborn screening usually continues throughout an individual's lifetime.

The initiation of timely treatment and the provision of adequate follow-up requires close coordination among the various components of the screening program. Partners in this process include the hospital, the Bureau of Clinical Laboratories, the infant's family and physicians, and specialists who oversee long-term treatment and monitoring. The University of Alabama at
Birmingham and the University of South Alabama are involved in diagnostic and long-term treatment efforts.

"Comprehensive newborn screening will prove to be a monumental benefit for the children of Alabama," State Health Officer Dr. Donald Williamson said. "The Health Department’s mission is to serve the people of Alabama by assuring conditions in which they can be healthy. What better way can we do that than by detecting problems in our children early?"

For more information, contact the Alabama Department of Public Health, Newborn Screening Program at (334) 206-2971 or (334) 206-5955.

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