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ARIZONA BABIES TESTED FOR 28 POTENTIAL DISORDERS

By Jennifer Ryan, Tribune

As 6-year-old Ethan Smith struggles with balance, coordination and sensory motor skills, his mother constantly is reminded of his genetic disorder.

Had Smith received more extensive blood tests at birth, he could have been diagnosed and treated earlier, possibly avoiding developmental delays. But at the time, standard newborn screening did not test for Ethans disorder.

Starting next spring, however, state officials plan to expand the number of disorders every baby in Arizona is screened for from eight to at least 28, including Ethans condition. The effort is expected to identify many more disorders, opening the door to treatment that could prevent disabilities and save babies lives.

"Ill always wonder what would have happened if he had been screened at birth," said Ethans mother, Mary Beth Smith of Phoenix. "We would not want anyone to go through what we went through."

A grass roots effort that included the March of Dimes, state health officials and Tempe parent Deborah Houk helped pass a state law this year expanding newborn screening.

Their work is part of a national effort to require every state to test for 29 disorders, including metabolic conditions and hearing loss, as recommended in a report this year by the American College of Medical Genetics.

"With many disorders, the first (clue) is death," said Jan Kerrigan, newborn screening program manager for the Arizona Department of Health Services. "Its really huge in terms of the lives saved and the quality of lives saved."

Policymakers nationwide have questioned whether its worth the money to test millions of babies for disorders affecting relatively few children.

Of the estimated 90,000 newborns in Arizona screened for eight disorders last year, about 100 tested positive. With expanded screening, state officials expect 38 to 59 more babies will test positive.

"Each disease individually is rare, but as a collective group of disorders, theyre not that uncommon," said Dr. Randy Heidenreich, professor of pediatrics and a biochemical genetics specialist at the University of Arizona College of Medicine. Having an effective screening program is important because rare conditions could be missed by doctors who may never have seen a patient with one of the conditions, he said.

For parents of children with genetic disorders, newborn screening is about saving children, no matter what the number, Houk said. Her 19-month-old daughter, Lauren, died in 1989 of the same genetic disorder Ethan Smith has: Medium chain acyl-Co-A dehydrogenase deficiency, or MCADD.

The condition prevents the body from breaking down and using stored fats, which can turn toxic and cause sudden death. Children with the disorder are easily treated with medicine and the right diet.

The diagnosis came when Houk was pregnant with her third child, Austin, prompting additional testing of her son at birth. The screening came back positive. Early diagnosis and treatment has kept Austin, now 14 years old, alive, she said.

Houk told the story at the Legislature this year, attending every hearing of the newborn screening bill she

could. During a final read before the bill was passed in the House, a lawmaker asked Houk and about a dozen of her supporters to stand.

"I think my knees were about to give out," Houk said. "In my own heart I felt like it was (Laurens) law and all my efforts were in honor of her. Her short life has made a huge impact on future children."

Dr. Jennifer Caplan, a Scottsdale pediatrician, said she is excited to see expanded newborn screening coming to Arizona.

One of her patients died before tests could confirm that he had a genetic disorder.

"This could have been prevented if we had known earlier," she said. "That delay in diagnosis can mean the difference between life and death."

The expanded screening program in Arizona is expected to cost about $5.7 million next fiscal year, paid for primarily by health insurers. Each baby will continue to be tested twice once after birth in the hospital and once after a well-child visit to a pediatrician using five drops of blood from a heel prick.

While 23 states have expanded their screening programs to include more than 20 of the disorders, the rest screen for fewer, according to the March of Dimes. Fifteen states and the District of Columbia screen for fewer than 10 disorders. Only Mississippi currently screens for all 29.

The disparities mean that about 80 percent of babies born in the U.S. receive screening for fewer than 20 disorders.

"Its really a shame that in this day and age that what state your baby is born in makes a difference," Smith said. "You would think there would be a standard for this type of thing, and the answer has always been money."

Expanding newborn screening requires states to spend thousands of dollars on new technology that can detect many more genetic disorders.

The state health department paid $834,722 for three tandem mass spectrometry machines, which will allow Arizona to screen newborns for 28 disorders, Kerrigan said.

"This is a big effort. This is a big technological shift for newborn screening," Heidenreich said.

Testing for the 29th disorder, cystic fibrosis, likely will involve more debate because the state will have to spend about $120,000 on additional technology and staff, Kerrigan said.

The bigger cost is missing an early diagnosis, Smith said. Because of developmental delays that may be tied to his delayed diagnosis, Ethan has required speech, occupational and physical therapy.

"You can either pay now or pay later," she said.

REPORTER CONTACT INFO:
Contact Jennifer Ryan by email at jryan@aztrib.com, or by phone at (480) 898-6535.