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Newborn Screening a Success, but Testing Varies Widely by State: Push to Make Tests Universal Nationwide

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Deb Gould is dedicated to a disease that most people have never heard of. She hadn't even heard about it until a routine newborn screening test diagnosed her second child with the same condition that unknowingly had led to the death of her first.

In 1986, Gould's second child, Kevin, was born. As is the norm, a sample of blood was drawn from his heel to undergo a litany of newborn screening tests that can detect life-threatening conditions. He was diagnosed with a rare genetic disorder known as MCAD, which meant Kevin's body was unable to break down fats into energy — a condition that if left undiagnosed and untreated could lead to sudden death. Unfortunately, the newborn screening test that saved her son's life hadn't yet been developed when her first child, Kristen, was born. Gould's second child is now 20 years old, but the death of Kristen led Gould to become a forceful advocate for comprehensive newborn screening and to create a support network for families with similar experiences.

"We felt like we were the only ones in the world dealing with this," Gould told *The Nation's Health*. "We had to do something to help ourselves and help others."

Gould and her family founded the Fatty Oxidation Disorders Family Support Group in 1991. Starting with just 10 members, the group now has a mailing list of 1,100, including families and health professionals. While the disorder that started it all can now be detected and treated, not every state requires its newborn screening program to test for MCAD. Newborn screening programs are a state's responsibility to develop and implement, and so the state in which a baby is born determines the disorders she or he will be tested for. Gould wants a universal and comprehensive newborn screening program based on the recommended 29 screening tests, and she said it "sickens" her to meet families who have lived through the death of more than one child without a diagnosis.

"Expanded newborn screening is extremely vital," she said. "If you catch just one child, what a world of difference it makes."

Every year in hospitals across the United States, 4 million newborns are tested for a variety of disorders via their state's newborn screening program. While every state tests for some disorders, only a handful mandate testing for all of the 29 conditions recommended for screening by the American College of Medical Genetics. The recommendations, commissioned by the U.S. Health Resources and Services Administration, were released in a 2004 report from the college.

Newborn screening began in the 1960s with the invention of a test for phenylketonuria, commonly known as PKU, which prevents the body from processing a part of protein called phenylalanine. If left undetected, PKU causes brain damage and mental retardation. Fast forward to the 1990s and the application of tandem mass spectrometry technology — which is used to analyze blood at the molecular level — and now the scope of detectable disorders at birth is greatly expanded.

While only five states and the District of Columbia mandate newborn screening for all 29

recommended conditions, there has been great progress during the last few years, according to Jennifer Howse, MD, president of the March of Dimes. So far in 2006, two-thirds of all newborns have been screened for 20 or more conditions, twice the rate of 2005, she said, adding that she expects every state will implement the American College of Medical Genetics recommendations within the next couple of years. Barriers to expanding newborn screenings can include additional costs, a lengthy legislative process and coordination with hospitals and laboratories as well as training personnel.

"From our standpoint, (newborn screening) is one of the real success stories in public health because it combines a low-cost, population-based screening program with highly effective treatment that literally saves lives and prevents long-term chronic disability," Howse told *The Nation's Health*.

As technology continues to advance, opening the doors to detecting multitudes of disorders at birth, public health officials will face a tough question: What should newborns be tested for? Howse sat on the American College of Medical Genetics' Newborn Screening Steering Committee during the development of the 2004 guidelines, and she said a trio of "classic public health" criteria led to a recommendation to screen for 29 disorders: which conditions have an accurate test, which conditions can be detected in newborns and which conditions there are effective treatments for. The March of Dimes, Howse and many of her colleagues hope that eventually every state will screen newborns for the 29 disorders. And while medical literature often cites concerns about the parental anxiety caused by the possibility of false-negative or false-positive test results, Howse noted that such a large screening program will inevitably yield false results.

"In the states, their labs set the threshold for the detection of these anomalies low enough so that they don't miss anything," she said. "You cast a wide net, you control to the greatest extent possible, but you don't miss anybody because to miss one of these conditions...the consequences are horrible."

Usually, though, such consequences are successfully avoided. Every year, 200 U.S. children are saved from mental retardation because a newborn screening test detects PKU and allows treatment to begin, said Scott Grosse, PhD, senior health economist at the National Center for Birth Defects and Developmental Disabilities at the Centers for Disease Control and Prevention. Before newborn screening, about 2 percent of all people with severe retardation had PKU, but that is now prevented. More than 1,000 children annually test positive for sickle cell disease, but because newborn screenings translate into early treatment, the mortality rate for such children younger than age 3 is now no higher than it is for the general population. In other words, newborn screening has essentially eliminated the excess toddler mortality rate associated with sickle cell, Grosse told *The Nation's Health*.

"For those families, it's made a huge difference," he said. "We have some great success stories."

However, Grosse said that uniformity in newborn screening across the nation probably won't happen — not necessarily because some states won't adopt testing for the 29 recommended disorders, but because some states will always want to go beyond that. As states consider new newborn screening mandates, he said, decisions should be grounded in evidence-based review. While the American College of Medical Genetics guidelines are a consensus of expert recommendations, they did not go through a more traditional evidence-based process, he noted. States should consider what criteria is sufficient when legally mandating screening for a particular disorder, he said.

"I'm not saying (a screening) is not worth doing and I'm not criticizing the recommendations," Grosse said. "But there are going to be many more disorders coming down the road and people

will advocate screening, so where do you draw the line? There should be an objective, evidence-based process."

The question of whether newborns should be screened for conditions for which there is not an effective intervention has been raised, and as of yet, "no national consensus has been developed," according to a report published in the September issue of *Pediatrics*. Celia Kaye, MD, PhD, the report's author and senior genetics advisor for the National Newborn Screening and Genetics Resource Center at the University of Texas Health Science Center, said the ability to look at many chemicals at once is both the problem and the power of tandem mass spectrometry, as it provides many advantages, but raises many questions. The technology can pick up abnormalities that researchers don't know the significance of, however she said it is worth it to find conditions, even though an effective treatment is uncertain.

"It's a wonderful opportunity to find out about rare diseases," Kaye told *The Nation's Health*. "And once you've got the children in your hands, the desire for effective treatment is so compelling that those treatments will emerge."

However, screening is only the first step of a multi-part system that includes follow-up, diagnostic testing, disease management and evaluation, according to Kaye's *Pediatrics* report. She said while short-term follow-up in states is "very good" — praising the dedication of state public health workers — comprehensive, long-term follow-up is lacking. Julie Miller, program manager of the newborn screening and genetics program at the Nebraska Department of Health and Human Services, said she would "love" to do long-term tracking and data collection, but there isn't enough staff or funding to do so — and it's always been that way, she added.

About 26,000 newborns are screened each year in Nebraska, which expanded its newborn screening panel with the help of supportive state legislators in 2003. Testing for eight conditions is mandated for each newborn in Nebraska, however, testing for the remainder of the 29 recommended conditions is offered universally to every parent at no additional charge, Miller said, adding that about 97 percent of parents take advantage of the offer. If a disorder is detected, Miller and her colleagues notify the appropriate physician and give her or him the tools with which to best communicate with the family. It's only if there is no physician in the picture that the health department contacts a family directly.

"(Newborn screening) can mean the difference between life and death, it can mean the difference between normal development and normal health versus mental retardation, seizures, liver disease, cataracts," Miller said.

She noted that while the screening guidelines are starting to create uniformity between states, every state has different available resources — so if federal policy-makers do decide to standardize newborn screening, additional resources would have to flow down to the states.

"If there was equity in resources across states, I think that absolutely we should be screening for (conditions) recommended on the national level," she said.

For more on newborn screening programs, visit www.marchofdimes.com or <http://mchb.hrsa.gov/screening>. For more news from *The Nation's Health*, visit www.thenationshealth.org.