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New Jersey Makes Strides In Newborn Screening

By MARALIESE BEVERIDGE

Recent changes in New Jersey law broaden the requirements of the state's newborn screening law, with the goal of saving lives and preventing serious medical conditions.

Every newborn in the United States is required by law to be screened for a variety of genetic and metabolic disorders that, if left untreated, could be life-threatening or cause serious medical problems. The number and types of mandated tests vary widely from state to state — creating controversy in some areas — but a recent change in New Jersey law broadens the program and gives parents expanded options in newborn screening.

"Although screens for more than 75 disorders have been developed, each state has its own requirement for how many tests will be performed at birth," explains Allen Bergum, program chief at the state Department of Health and Senior Services' Inborn Errors of Metabolism Laboratory. Bergum was one of many state and national speakers who discussed newborn screening at a recent NJHA conference.

The number of tests required by each state varies from as few as three to a maximum of 40. New Jersey law specifies 20 disorders that newborns must be screened for, but the state laboratory routinely tests for a total of 33 conditions. In addition, New Jersey healthcare providers are required by law to inform parents of the option of supplemental screening to cover additional disorders.

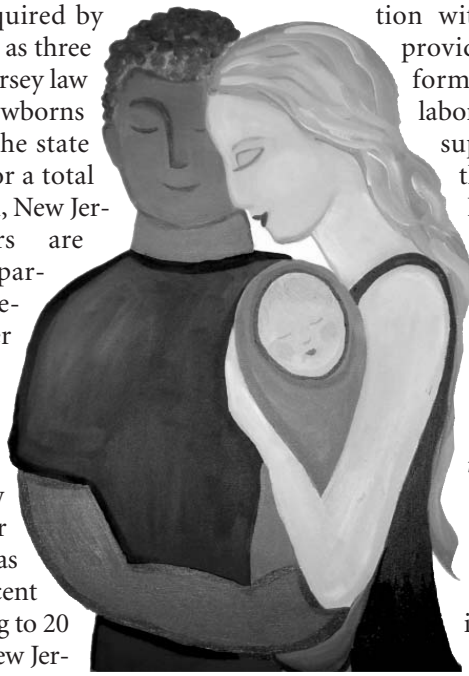

ILLUSTRATION BY PAMELA BROWN-VILLARUZ

Recent Changes, Historical Roots

As recently as 2001, New Jersey law required just four screens. That number has grown significantly in recent years, with the state expanding to 20 by 2003. At the same time, New Jersey also began screening all newborns for hearing loss within the first month of life.

Recent technological advancements, including DNA-based tests and tandem mass spectrometry, have made more tests available in a shorter period of time. New Jersey's mandated 20 screens — plus the 13 additional screens routinely run in the state lab — places it among the frontrunner states for newborn screening nationally.

Conditions covered under New Jersey's screening mandate include congenital hypothyroidism, cystic fibrosis, maple syrup urine disease, galactosemia and hemoglobinopathies such as sickle cell disease.

Another advancement in newborn screening in New Jersey came in March, when the state Legislature passed a bill that requires healthcare providers to educate parents on the availability of supplemental tests prior to birth. A request for supplemental testing must be made in coordination with a physician or healthcare provider and testing must be performed through a qualified private laboratory. The cost of this optional supplemental testing is borne by the family. In response, NJHA's Health Research and Educational Trust has undertaken a statewide campaign to distribute this important information to both healthcare providers and the general public through a variety of printed and electronic materials. *(For more, see Page 4 Interview.)*

Bio-chemical testing of newborns dates back to the early 20th century and primarily consisted of visual data and inspections and relatively simple chemistries. This was essential-

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ly the only testing model in place until the early 1960s when these basic standards not only became mandatory but were also revolutionized by the development of a simple dried blood spot screening test that could identify possible risk pre-cursers for certain disorders. This test was initially referred to as the PKU test, which eventually stood as an umbrella label for a handful of other tests that developed over the next decade.

"Today, we are able to perform many screens from one blood spot sample."

— Gloria M. Rodriguez

The development of additional tests was slow going until the advent of MS/MS technology, which can test for 14 bio-elements from one blood sample. This groundbreaking technology has paved the way for expanded screening possibilities.

Patient and Provider Roles

Screens are not diagnostic tests, but rather risk assessments that indicate the need for testing. The earlier a disorder is detected, the earlier the intervention and the greater the assurance that treatment will be received. It is the responsibility of each hospital to collect a baby's blood sample on a special filter paper using a heel stick, following a state-mandated protocol, and mailing it to the state laboratory. The state lab sends the results to the hospital's physician of record or the physician who ordered the test. Follow-up procedures are crucial, especially in the event of abnormal results that require the parents to bring their baby back for additional tests. In such cases the lab also notifies the state program, which contacts the physician for repeat screenings or diagnostic tests and connects the infant's family with a specialist to start immediate treatments.

"Today, we are able to perform many screens from one simple blood spot sample taken from the newborn's heel," says Gloria Rodriguez, director of DHSS's Family Health Services. "Right now, while 33 tests are automatically handled through our lab, parents need to know that supplemental testing is available and that it must be handled through a private lab."

If parents are considering supplemental testing, it is essential that they be educated well in advance about these screens since testing kits must be obtained several months prior to the birth. It also is important for parents to realize that their health insurers may not cover the full range of testing.

Challenges and Controversies

All states are facing the challenge of managing expanded newborn screenings at the rate tests are becoming available and are at different stages in approaching this challenge. While parents' groups and other advocates push for greater screening mandates, the number of screens that are mandatory varies among the states for many reasons, including individual population needs, funding for technology and political and economical demographics.

"One controversial issue being examined is the lack of standardized definitions or processes for the states to determine which additional disorders to screen and how to count these screens," says Firoozeh Vali, PhD, assistant vice president of research for HRET. "For example, one state might count a screen that is performed to detect four versions of a disorder under its broad category as one disorder, while another state might consider the same screen as four disorders. The need for a standard way to count these screens is necessary before we can compare state programs and the number of disorders they screen."

Another issue: States with relatively low birth rates might not be able to justify spending scarce health dollars for cost-prohibitive technology that screens for disorders that are relatively rare.

"One controversial issue is the lack of standardized definitions or processes."

— Firoozeh Vali

A national task force was formed in 1999 to meet the challenges of standardizing newborn screening. While its report provided a good general assessment of newborn screening programs on a national level, it did not address which or how many diseases should be included in states' newborn screening programs.

New Jersey's response to this sensitive issue was to form the state's Newborn Screening Advisory Panel in April 2000. It focused on reviewing current newborn screening in New Jersey and the many additional disorders for which screening existed. Ultimately, the panel made recommendations that led to the expansion of New Jersey's screening program in 2001 and 2003. When adding disorders to be screened, experts consider how prevalent the disorder is, whether it is treatable with early intervention, the morbidity risk if the disorder is left untreated and whether an accurate screening test is available. The panel also made recommendations that prompted HRET to launch its initial public education campaign on the enhanced newborn blood and hearing screening programs in 2003. That work continues today, with HRET continuing to develop and distribute a variety of resources for parents and healthcare professionals.

Newborn screening has long been wrought with controversy, and the debate is likely to grow as technology continues to develop and expand the possibilities in newborn screening.

While there is no easy answer to the question "how much testing is enough," there is consensus on one point: Newborn screening can improve — and even save — lives through quick detection and early intervention.

Perspective

GARY S. CARTER, FACHE – President

Anybody who's visited my office lately and seen the family photos on my desk know that I'm the proud grandfather of four beautiful granddaughters. My family and I count our blessings that those little girls are healthy.

Sadly, not all families are as fortunate. Millions of infants each year are born with a seemingly endless array of genetic, metabolic and infectious disorders. These conditions can lead to severe health problems, mental retardation, even death. The good news is that many of these conditions — things like cystic fibrosis, sickle cell disease and congenital hypothyroidism — can be managed with quick detection and early treatment.

That's why newborn screening is such an important — and emotionally charged — topic. All states require some level of mandated newborn screening to detect these conditions. Some states, quite frankly, do a better job of it than others. New Jersey has made major strides in recent years to screen for a greater number of conditions, with state health officials and healthcare providers working together to create a screening program that is effective and efficient. NJHA has played an instrumental role in that work, serving on the state's advisory panel on newborn screening and now, through our Health Research and Educational Trust, developing educational resources for both our hospital members and new and prospective parents.

That's an important part of our healthcare mission and one that I can embrace, wholeheartedly, on behalf of our member hospitals, the families of New Jersey and the four little girls whose smiles grace my desktop.

Joshua's Story: Mother Fights to Expand Newborn Screening

By KERRY McKEAN KELLY

For Sandi Hammer and her son Joshua, life has been changed by a matter of 12 days.

Joshua, age 5, was born with maple urine syrup disease, a rare inherited metabolic disorder that, if left untreated, can lead to mental retardation, physical disabilities and death. Today, state law requires that babies born in New Jersey be tested for the condition in the first hours of life. Caught early, the disease can be managed and the most severe implications prevented with a special daily diet and close medical supervision.

Unfortunately, Joshua was born before his disease was added to the list of screenings required by state law. His condition wasn't detected until he was 12 days old — and was already in a coma, suffering seizures and breathing through a respirator in the intensive care unit.

"Had he been picked up earlier, he wouldn't have gone into crisis," said Hammer, who lives in Springfield.

Today, Joshua is a happy, active kindergartener. And although he's progressing well, he still confronts his condition on daily basis, facing myriad blood tests, doctors appointments, severe diet restrictions and other struggles to keep it under control.

Two years after Joshua was born, Sandi Hammer gave birth to a second son, Matthew. Matthew also was born with MSUD, but this time, the family was prepared through early screening and treatment that began the day he was born. Matthew was spared the early complications that his older brother endured, and Sandi Hammer believes that his condition is easier to manage still today, thanks to the early treatment he received.

"Every baby deserves a chance… and these screenings give every baby and every family that chance for a good quality of life," she says.

The Hammer family's experience has prompted Sandi to speak out for expanded newborn screening. She spoke to healthcare providers recently at an NJHA education session on recent changes to the state's newborn screening law. In fact, she was among a group of parents who lobbied state lawmakers to adopt the changes.

"I will talk to whoever will listen," she says. "If we can help one family from going through this, it is worth every trip to Trenton."

"Every baby deserves a chance."

— Sandi Hammer

Sandi Hammer says she knows there many complicating factors in expanding newborn screening — the expense, the potential for false positives, the ethical questions. But she urges anyone who will listen to keep focused on the children and the families who live with these conditions.

"There is a parent and a child at the other end of this discussion, and I want to make sure people understand this," she said.

And she applauds the state for expanding the number of mandated screenings, and she praised healthcare providers at NJHA's recent conference for educating themselves and their patients about newborn screening.

"We need to do whatever we can to save these babies," she said.

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Interview: Firoozeh Vali, PhD

By DEBBIE FURCHAK

HRET, with the support of grants from the March of Dimes — New Jersey Chapter, the N.J. Department of Human Services — Office of Prevention of Mental Retardation and Developmental Disabilities and the N.J. Department of Health and Senior Services, recently launched a statewide campaign to educate parents about newborn screening. Firoozeh Vali, PhD, HRET's assistant vice president of research, discusses the project.

Q What is the major goal of the newborn screening project?

Between 2001 and 2003, New Jersey expanded its biochemical screening program and increased its mandated screens from four to 20. The state's hearing screening program was also expanded during this period. This project's major goals are twofold: to improve parents' knowledge about enhanced newborn blood and hearing screenings and improve their compliance with follow-up instructions; and to improve clinicians' knowledge about new screenings and protocols.

Q What new resources are now available to hospitals and parents?

HRET has developed a variety of educational resources for prospective parents and healthcare professionals. They include a New Jersey-specific videotape available in English and Spanish, two brochures in English and Spanish – one short and simple brochure, the other longer with more extensive information, and an accompanying poster available in five languages (English, Spanish, French Creole, Chinese and Arabic).

Currently, HRET is widely distributing copies of these resources to all New Jersey hospitals with maternity services and birthing classes, all appropriate provider offices, mainly OB/GYNs, family practitioners and pediatricians, as well as state and community-based agencies involved in providing prenatal care or other types of services to pregnant women.

Q How will HRET measure the effectiveness of this campaign?

HRET will measure the effectiveness of the educational resources through a comprehensive evaluation plan using a three-pronged approach: feedback from clinicians, feedback from prospective parents recruited from birthing classes around the state and analysis of data obtained from NJDHSS on parents' compliance behavior from 2002 to 2005 and if they promptly bring their baby back when repeated confirmatory screenings or additional tests are needed.

Q How can healthcare providers obtain copies of these resources?

Order forms are available on NJHA's Web site at *www.njha.com.* Providers can also contact HRET at 609-275-4145.