

SCREENING NEWBORNS:

Just the Beginning

Newborn screening and tracking diagnosed children into adulthood varies among states.



BY ALISSA JOHNSON

Stephen Monaco's life was forever changed on May 30, 2001. That morning the normally energetic 3-year-old who loved Clifford the Big Red Dog was found unresponsive. Today Stephen sits in a wheelchair, brain-damaged and fed through a G-tube. Next to him, his sister Caroline is a vibrant, healthy 5-year-old. One would never guess that these siblings share the same condition: isovaleric acidemia. For Caroline, the disease was caught early, and she was treated immediately at birth.

Stephen and Caroline's mother, Jana Monaco, is quick to point out that Stephen finds joy in his life expressed through laughter and smiles, but her hopes and dreams for her once healthy son were changed overnight with the sudden onset of illness—a bitter pill to swallow with the knowledge that newborn screening could have detected his condition, allowing for treatment and thus preventing the disorder's devastating consequences.

Today, newborns are screened for Stephen's condition in his home state of Virginia as a result of legislation sponsored by Senator Linda "Toddy" Puller. It consoles Stephen and Caroline's mother that babies with their disorder will be identified in the future

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and have Caroline's quality of life rather than Stephen's.

"Newborn screening at the current time is by far the most common form of genetic testing being done," says Dr. Rodney Howell, chair of the federal Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children. "Almost 90 percent of the babies in the country are getting about 30 tests. That's over a hundred million genetic tests being done a year."

TESTING AND FOLLOW-UP VARIES

All states have well-established newborn screening programs and many have expanded them in recent years. In 2005, the American College of Medical Genetics released a report recommending screens for 29 core conditions and identifying 25 other conditions that could be detected in the same screening. According to the National Newborn Screening and Genetics Resource Center, as of February 2008, 37 states require screening for at least 30 disor-

ders with some doing as many as 50, but only 12 states actually screen for the core 29.

In addition, what states do to track children who have been identified with a disorder as they grow into adults varies from state to state.

"There is no systematic way currently in the country where any of these children are followed in an organized fashion," Howell says. "That obviously creates an enormous problem for two reasons: You do not know what has been the outcome for these children and there is no standard way of treating them."

Howell says the federal advisory committee is looking for ways to follow the identified children to see what happens to them, to find out what interventions are done and to test new therapies. The committee hopes to clarify what kinds of long-term follow-up are most helpful. Since states are charged with carrying out newborn screening programs, they will clearly have a role in making it happen.

State laws appear to do a good job addressing short-term follow-up, but long-term follow-up is scarce. Timothy Hoff, associate professor of Health Policy and Management at the University at Albany School of Public Health, conducted a survey in 2005 that focused on how state newborn screening programs defined long-term follow-up, whether and how it was conducted, and whether staffing and resources mattered. Hoff learned



SENATOR

LINDA "TODDY"

PULLER

VIRGINIA



Getting the Needed Care

As the effort to develop new treatments continues, frustrations over the ability to get existing therapies persist for some families.

Jana Monaco, mother of two children with isovaleric acidemia, says one medication her children need is covered by insurance, but the other is not. Far worse, she says, is that some families don't have coverage for the therapeutic formula these babies need. "We have been fortunate," Monaco says, "but many families do not have coverage for formulas specifically designed for their metabolic disorder. You could fall under the poverty level and maybe qualify that way, but what about middle class society?"

While she considers herself lucky, Monaco's husband recently declined an appealing job offer when he learned that the insurance policy available through the employer included a clause excluding coverage for the expensive dietary formula her children need.

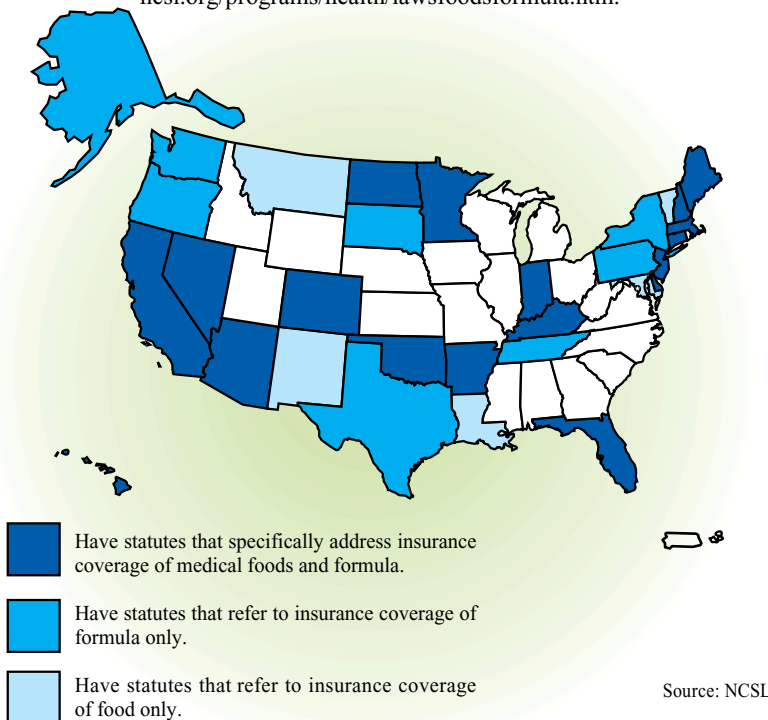
The Connecticut General Assembly passed a bill last year that requires health insurers to cover special food products and formulas for children under age 12 with inherited metabolic diseases. Representative Brian O'Connor, chair of the Insurance and Real Estate Committee, says that although he is hesitant about mandates, proponents of the bill were convincing. "They demonstrated that the cost of the product was much more expensive than regular baby formula and was putting a strain on households." At least 35 other state legislatures require insurance coverage of medically necessary foods and formulas to treat disorders identified through newborn screening. Of those states, 17 laws specifically mention medical foods and formulas.

Five of the 36 state laws provide for access to medical foods or formula through various public programs. Oklahoma adopted a resolution "encouraging" coverage of medical foods and formula. The District of Columbia also provides the indigent with treatment for disorders identified through newborn screening.

"I have had several patients who have been forced to go without formula, especially the older patients," says Rani Singh, director of the Metabolic Nutrition Program at Emory University. "The biggest challenge we face is that the women with PKU [phenylketonuria] can have a normal pregnancy if they are on a strict diet before getting pregnant. However, even Medicaid patients cannot get approval for formula until they become pregnant. By the time they go through the approval process, it is already too late, and they are at risk of having a child with mental retardation and other problems."

State Insurance Coverage Requirements for Medical Foods and Formula

State requirements for coverage may have caps or age limits. For more information, please see NCSL's website on coverage of medically necessary foods and formula to treat disorders identified through newborn screening at www.ncsl.org/programs/health/lawsfoodsformula.htm.



SCREENING WILL SERVE RESEARCH

New treatments will become more available as the Newborn Screening Translational Research Network proposed by the Department of Health and Human Services National Institute for Child Health and Human Development moves forward. The institute is in the process of establishing an organization to serve as a coordinating center for the network that will help develop new screening methods, clinical trials for new medicines and support research to study the long-term health of children identified through newborn screening. States have the potential to play a critical role in the network.

“Most states can be key in the research network with the material and information they collect as part of their screening program,” says Dr. Rodney Howell, chair of the federal Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children. “Some of these research projects will fit in with the state mandates and permit the states to participate without doing anymore screening than they already are. I think that there is a great opportunity for states to be very important members of a research team.”

that all states have well-functioning short-term follow-up programs, but most do little or nothing long term. He believes states are missing out on a valuable opportunity. By tracking these children, states would better understand the costs and benefits of testing.

Senator Puller says she recognized the need to ensure follow-up in her newborn screening bill, but had concerns about becoming too specific and hampering the health department. “It makes more sense to have the health depart-

ment creating regulations for follow-up. The program needs to have flexibility and having that much detail in the code is burdensome.”

The Health Resources and Services Administration Maternal and Child Health Bureau has funded seven Genetics and Newborn Screening Regional Collaborative Groups to improve states’ screening capacity and equalize distribution of resources within regions.

Dr. Cary Harding, a consultant to the Northwest Regional Newborn Screening

Program, deals with Alaska, Hawaii, Idaho, Nevada and Oregon, some military bases in California, New Mexico and Washington, and birthing centers scattered across the Pacific. Harding must coordinate any initial treatment for infants with positive screens.

“Once a diagnosis is made, who ends up taking care of the children is different depending on the state,” he says. In Harding’s experience, access to specialty services varies, partly because there are not enough genetic and metabolic centers in some areas. He notes that providing services through telemedicine can help, but there are often economic and licensing barriers to consulting in another state. Harding has also found that every state collects information differently, if at all, about how infants fare in the long run.

KEEPING TRACK OF PATIENTS

The regional collaborative groups are moving toward the goal of following children at least through childhood. For example, Dr. Susan Berry, professor of pediatrics at the University of Minnesota, is working on a project to develop treatments for metabolism disorders

detected through newborn screening. “Because these disorders are rare, you need collaboration across a wide geographic area to accumulate enough patients for meaningful evaluation,” she says. “The problem was that we did not have a real way to keep track of the patients. We have focused on finding a way to capture their clinical history and progress.”

Berry and her colleagues have developed an online, secure information system that allows centers in the region to enter information about patients. The information system currently is focused only on children with one particular disorder, but the region plans to add more conditions over time. Through the system, Berry hopes to improve the care children with these disorders receive.

“If we could compare practices from institution to institution and look at the differences, we might be able to tease out whether one treatment was more useful than another. This would give us a foundation to undertake a more specific, planned clinical trial,” Berry says.

Jana Monaco who, in addition to being a parent, serves as president of the Organic Acidemia Association, agrees that an important part of long-term follow-up is the ability to conduct research to benefit patients. “Personally, researchers were able to identify the exact mutation that my children have. It doesn’t change anything for them, but it does help physicians to better understand why a child like Stephen made it to 3½ before he went into severe crisis versus a newborn who might go through the same thing in the first week of life. By researching and identifying the various mutations, one can understand how aggressively you need to treat a child.”

The value of being able to find patients is now particularly apparent on a larger scale since a new drug to treat phenylketonuria (PKU), the first disorder screened for in newborns, was recently approved by the Food and Drug Administration. There’s no way to track down all of the patients identified with PKU since the 1960s, so it is impossible to ensure that they all get the new drug, which is both easier to take and less costly than the special formula traditionally used.


FEDERAL GRANTS MAY HELP

New federal legislation, still pending in the House, may help newborn screening programs understand how children are faring as they grow up. The bi-partisan Newborn Screening Saves Lives Act of 2007 passed the

Senate in December. It would provide grants to improve education and outreach on newborn screening and follow-up care. The act would also create a clearinghouse of newborn screening information.

The number of children diagnosed in the first few days of life continues to grow with each new baby screened by the states. Jana Monaco acknowledges that the magnitude of the commitment to these children may be greater than initially anticipated as lives are prolonged by

new therapies. But she says it is important to remember that these conditions are very rare: “We are not talking about thousands and thousands of babies in each state, but every life does count. These children are becoming adults who are going to need life-long care.”

 **CHECK OUT** our 50 state information on newborn screening at *State Legislatures* online www.ncsl.org/magazine.