Parents Prod States to Boost Newborn Testing

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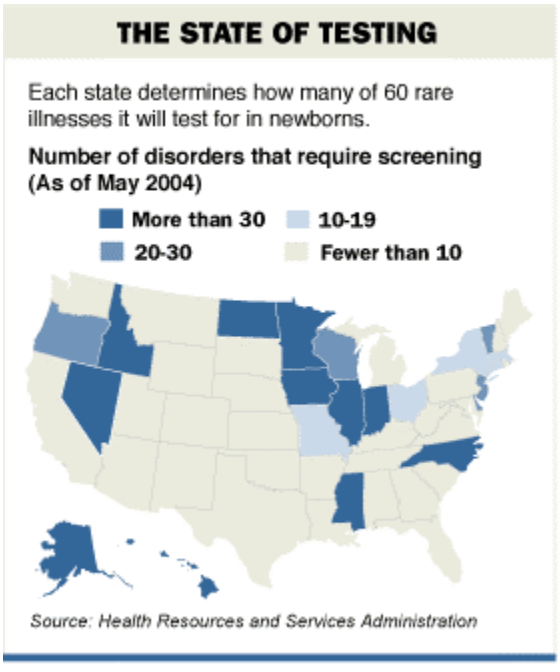
Reacting to angry parents who say their sick children could have been diagnosed and treated at birth, states across the country are significantly expanding the number of rare but devastating diseases for which they test newborns.

Health advocates say many states are failing to incorporate into their mandatory newborn screening programs relatively inexpensive tests for several dozen diseases, despite emerging evidence that the tests can save lives and prevent illnesses that can disable children for life.

Advocates point to numerous cases where children born with illnesses in one state aren't being detected and treated while children with similar illnesses in nearby states are.

Following several emotionally charged meetings with parent activists, New York state health department officials say the state has agreed to soon triple the number of diseases for which it tests newborns. Missouri officials last month announced they will expand the number of diseases screened at birth to 25 from five. California's legislature this week passed a law that increases the number of illnesses it will test to 41 from four, making it one of only two states screening for that many conditions.



The actions come as a growing number of parents, doctors and public-health officials are calling for states to conform to a proposed national standard for testing infants. Pressure to meet the standard is certain to intensify in late September with the public release of a long-awaited report on newborn screening from the American College of Medical Genetics.

The report will be presented to a special advisory committee to the secretary of the U.S. Department of Health and Human Services. Officials familiar with the report say it will urge that every state test for at least 30 genetic diseases, each of which can be detected from a blood sample taken immediately after birth. The ailments typically can be treated or even cured by relatively simple dietary changes.

The illnesses, with tongue-twisting names like short chain acyl-coa dehydrase deficiency, or SCADD, and congenital adenal hyperplasia, or CAH, are inherited disorders caused by the malfunctioning of genes that control the body's metabolism of proteins and enzymes. Common foods can produce toxins that damage the brain or muscles of affected children.

The fact that some states test for certain diseases and others don't "has become a national tragedy," says R. Rodney Howell, who directs the pediatrics department at the University of Miami School of Medicine and heads the federal advisory committee on newborn screening.

Members of the advisory committee say that at their next meeting in late September they expect to recommend that the federal government immediately begin providing states with funds to use a new disease-detection technology, to hire and train staff to conduct the tests, and to counsel parents of affected children, as well provide grants to help states educate doctors and prospective parents about the availability of the tests.

But news of the expected federal recommendations is already putting pressure on states to act. Mike Watson, who heads the American College of Medical Genetics' team compiling the new report, presented a preliminary version of the group's 30-disease recommendation to the HHS committee in early June, and at several other medical genetics meetings since late spring.

The circulated report "is getting the attention of everyone in the field" of newborn screening, says Paul Fernhoff, a professor at Emory University in Atlanta and chairman of Georgia's newborn screening advisory task force. Dr. Fernhoff says that as a result of the report he expects that Georgia's health department will begin testing for all 30 diseases being recommended. Georgia currently tests for only nine conditions.

Though each of the diseases in the new list is relatively rare, combined they occur in about one in 1,000 births, or about 4,000 of the four million or so children born each year in the U.S., according to estimates by the U.S. Centers for Disease Control and Prevention. Much of the damage can be avoided if a child's illness is detected soon after birth and his or her diet is altered or supplemented with certain vitamins.

Yet few couples are aware of these diseases. That is because they arise only if a child inherits two copies of a defective gene, one from each parent. Parents who carry just one defective copy are generally healthy and have no reason to suspect they may pass a disease to their children.

Beginning in the mid-1960s, every state began testing newborns for phenylketonuria, or PKU, by taking a spot of blood from an infant's heel. By the mid-1990s, most states tested for at least four diseases. Because the tests were mandatory and included in a hospital's delivery charge passed along to insurers, most parents generally were unaware of the tests unless they came back positive.

By 1996, several states and private laboratories began offering a multidisease test that employs a powerful new detection technology called tandem mass spectometry, or simply tandem mass. The tandem mass machines can detect about 40 genetic illnesses from a blood sample, and typically add about $60 to $80 to a hospital bill, although some states absorb the cost. Some states have been slow to employ tandem mass because doing so required new legislation to buy the machines, at about $300,000 to $500,000 each, and the funding to hire and train staff to conduct the tests and monitor treatment if a disease is detected.

Some state officials have argued that the diseases are so rare it didn't make economic sense to invest in the new technology. Advocates for mandatory use of tandem mass counter that the rarity of the diseases is exactly why each state should be looking for all the detectable disorders. "It's common for parents with sick, but undetected, kids to spend months, sometimes years, going from doctor to doctor, trying to figure out what's wrong," says Kelly Leight, a parent of an affected child who founded CARES Foundation, a Millburn, N.J., group that is lobbying for expanded state testing.

Jill and Peter Fisch, of Scarsdale, N.Y., didn't find out what was ailing their 3-year old son until last November, when doctors finally diagnosed him with the genetic enzyme deficiency SCADD. New York doesn't test for SCADD. Matthew is now on a diet that is expected to mute the disease's impact, but Ms. Fisch says he has muscle problems that limit his physical activity. Ms. Fisch has been pushing New York to expand its testing.

"We will never know what Matthew's full potential could have been because he suffered so many setbacks while we were looking for a diagnosis," she says.

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