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Costly Newborn Test Fuels Debate on Value

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BODY:

Jill Wood of Fairfax knows firsthand about the body chemistry disorders that strike thousands of newborns each year.

Two years ago Wood's 3-day-old daughter Hayley died of a rare metabolic disorder called carnitine palmitoyltransferase, or CPT II. At the time Wood, like many new parents, had no idea such disorders existed — or that they can often be treated if detected soon after birth.

Across the country, standards for newborn testing vary greatly: Several states test for as few as four disorders that affect up to one in 4,000 babies. Connecticut tests for 30, according to recent data presented last month at a Senate subcommittee hearing on newborn screening. But many parents say they don't always know which tests are administered and which aren't.

Now the March of Dimes, the leading U.S. birth defects prevention advocacy group, has called for all states to test for a minimum of nine disorders. Many, including Virginia and Maryland, already test for eight of these nine; the District tests for seven. In Virginia, testing is mandatory unless parents sign a waiver citing a religious objection. In the District and Maryland, testing is routine but voluntary; parents sign an informed consent form, which they usually receive on arrival at the hospital or at the first visit to a pediatrician.

The nature of the ninth test, however — or rather, the technology required to perform it — has brought state testing policies under new scrutiny by Congress, the General Accounting Office and some medical establishment heavies — including the Department of Health and Human Services (HHS), the American College of Medical Genetics and the American Academy of Pediatrics.

The test, now performed by only four states — Connecticut, North Carolina, Ohio and Wisconsin — is for a condition called medium-chain coA dehydrogenase deficiency, or MCAD. It strikes the one baby in 15,000 who lacks an enzyme needed to convert food fat to energy. If the condition is not treated through diet beginning immediately after birth, infants can develop seizures, respiratory failure or cardiac arrest, or die. Not only can the blood analysis required to check for MCAD find that disorder; it can detect perhaps another 30 disorders, including the one that killed Hayley Wood. But unlike the first eight tests on the March of Dimes list, which labs can do easily for an average $35 for the set, MCAD requires a sophisticated and expensive analysis device, called a tandem mass spectrometer (TMS). Each one costs $400,000; training and paying an operator and a lab analyst add to the expense.

These costs have raised critical questions about infant screening: Should testing standards continue to vary according to individual states' commitments to infant health and ability to pay? Or should the federal government mandate national standards? And in either case, is the benefit high enough to justify the cost?

Screening for newborn body chemistry disorders began in the early 1960s with a test for phenylketonuria, or PKU. An enzyme deficiency that is present in one in 12,000 babies, PKU can result in mental retardation unless protein intake is sharply reduced for babies starting very soon after birth. Recognition that the cost of testing was far less than the cost of caring for a mentally retarded person helped spread the adoption of PKU testing by state after state. States gradually

expanded the number of required tests; generally these screened for disorders that met World Health Organization standards as "important health problems" for which an accepted treatment existed.Until, that is, the expensive screening for MCAD came along. TMS screening, the technology used to detect MCAD, "has created a complex question for newborn screening programs," says Brad Therell, director of the National Newborn Screening and Resource Center in Austin. In addition to high cost and the need for skillful interpretation of the results, he says, "availability of appropriate medical expertise for follow-up of the [TMS] screening results is sparse . . . and treatment for some of the detectable disorders may not result in a cure."

Others are also asking just how much public money should be spent to detect illnesses, only some of curable. "In an era in which there are limited dollars for health care, especially for children's health care, it's not enough to ask whether newborn screening is beneficial," says Jeffrey Botkin, chairman of the American Academy of Pediatrics bioethics committee and professor of pediatrics at the University of Utah Medical School. "We also have to ask whether it's as beneficial as other uses of those dollars, such as mental health benefits and increased support for immunizations. . . . We ought to be spending the dollars for all of those things, but since there's not enough to go around, hard questions have to be asked before the allocations are made."

The problems, Botkin told Senate subcommittee members last month, "highlight the need for uniform national policy on the selection of newborn screening tests."

There's also an issue of emotional and psychological costs for parents whose infants receive false positive results. Across all tests, Botkin said, only a surprisingly low 1 percent to 5 percent of babies who initially test positive turn out to have the disorder. Botkin said even after parents receive confirmation that there's nothing wrong with their children, many continue to worry. TMS screening, he said, could improve the accuracy of the screening for many of the disorders.

Parent advocacy groups endorse the adoption of national standards to eliminate inequities and reduce delays in sending, processing and returning lab tests that they say put children at unnecessary risk.

The Winston, Ga.-based Save Babies Through Screening, was founded by Tera Mize in memory of her baby Tyler, who died of galactosemia, an enzyme disorder, nine days after birth and several days before his parents received his positive test results. "To know that where you live can mean the difference between whether or not your child lives or dies is just unbelievable," says Mize. "I have yet to meet a parent who learns of the differences among states and isn't outraged about the discrepancy. We wouldn't accept different state standards for Pap, breast or cholesterol screening."

But even as the questions are being asked, there is momentum on several fronts to improve several aspects of newborn screening. A bipartisan group of senators recently asked that Congress authorize a $25 million appropriation that would go, in part, toward creation of an expert panel to advise the HHS secretary about newborn screening tests. In addition, the American College of Medical Genetics, a group of physicians and researchers, is midway through a two-year assessment of criteria for deciding which newborn conditions warrant testing.

In the meantime, states are moving ahead with their own agendas. Maryland, with 70,000 births each year, has purchased two TMS devices. Within a year or so, the state plans to decide which disorders it will screen for using the new technology, says **Susan Panny,** head of Maryland's newborn screening program.

Virginia plans to begin using the new testing system by 2004. The District, through a contract with a Pennsylvania company, Neo Gen Screening, uses TMS screening but tests only for seven disorders on the March of Dimes list. The District is discussing whether to expand the list, according to Joyce Brooks, a newborn screening expert with the D.C. Department of Health.

There is also a push for better public education about testing for parents and health care professionals. Sen. Christopher J. Dodd (D-Conn.), who is chairman of the Health, Education, Labor and Pensions Committee's subcommittee on children and family, plans to introduce legislation that would allocate $15 million for this purpose as well as technology training and follow-up care.

Such education is pivotal, says Marina Weiss, March of Dimes senior vice president for public policy and government affairs. "Many parents have absolutely no idea about screening unless a child is diagnosed with a disorder. Obstetricians often don't review the screening process, and most parents are unlikely to see a pediatrician until after the blood sample has been taken."

The American College of Obstetricians and Gynecologists has not taken an official position on educating parents

about newborn screening, but Laura Riley, head of the group's committee on obstetrical practice, says some obstetricians do talk with their patients about the disorders, generally during the third trimester.

For now, says Tera Mize, the best parents can do is learn what tests their state conducts and consult their pediatrician about whether to have a private lab augment the list (see "Getting Ahead on Baby Screening").Jill Wood regards the answer as obvious. She says she now gives out lab test kits at baby showers. "We recently visited our daughter's grave instead of holding her second birthday party," says Wood. "I think newborn screening should be as well known as amniocentesis."

Francesca Lunzer Kritz and Sharon Mazel are Washington area health care writers.