

*EP (Exceptional Parent) rarely endorses the positions expressed in a guest editorial but in this case, we not only concur with Mr. Hehmeyer's position on Universal Newborn Screening but we wholeheartedly endorse it. Moreover, we wish to be clear on this point: **EP supports and endorses Universal Newborn Screening. Universal means all: all newborns should receive all available tests.** Some states have adopted comprehensive newborn screening or expanded newborn screening. We find these descriptions misleading because they are neither comprehensive nor expanded. Each state and territory needs to screen newborns for hearing and for all 30 + inborn errors of metabolism for which testing can currently be done and therapy exists. We need to protect the lives of the single greatest asset we have as a nation and as a people—our children. **Universal Newborn Screening should be a national imperative. In our opinion, the time for debate has ended and the time for action has arrived.***

This article, along with a copy of EP's monograph on Universal Newborn Screening, has been sent to all US Senators and each state's director of public health in hopes that they will champion this issue. EP also encourages readers to reprint or cite this article (giving proper credit) for circulation in other forums. —EP

Editorial Commentary ~ The Case for Universal Newborn Screening Charles P. Hehmeyer

Did you know that right now, the United States allows 1,000 of its own children to be killed or injured each year? Not intentionally or with malice, but it is done nonetheless.

Every year approximately 4,000,000 babies are born in the U.S. Of those 4,000,000 babies, at least 1 in 4,000 is born with a treatable metabolic disorder that is likely to kill or harm the child before diagnosis. That's 1,000 kids—an average of 20 per state—every year. Measured against the backdrop of 4 million births each year, 1,000 is "statistically" very low. But try telling that to new parents whose child may have had one or more of these disorders and who could have been saved if the disorder had been detected at birth.

Since early 1993, a method of screening babies at birth for treatable metabolic disorders has been available. That method is called "tandem mass spectrometry" (MS/MS). If blood from a baby with a metabolic disorder is screened by MS/MS approximately 24 hours after birth, chances are very high that, with treatment, the child will not die or be injured. On the other hand, if a baby with a metabolic disorder is not screened, chances are very high that the child will die or will be permanently and severely injured. It's that simple. But, more than eight years later, by and large we don't use MS/MS to screen newborns in the US. The interesting question is, Why?

What is a metabolic disorder?

Metabolic disorders are inherited genetic errors in which a person is unable to process a specific enzyme. People with metabolic disorders cannot break down their food

properly. The cells in their bodies will not be able to turn food into the fuel that bodies need to run efficiently. There are dozens of metabolic disorders; they are identified either by the substance which cannot be metabolized properly or by the place in the metabolic pathway where the problem occurs.

The treatment is to reduce or eliminate intake of the food which cannot be metabolized and/or to supplement the diet with the missing products that would have been produced if the enzyme worked properly. Treatment also may include supplements that help defective enzymes work better.

PKU—where it all began

The best-known metabolic disorder is phenylketonuria (PKU), which occurs in about 1 in 15,000 babies. PKU results in a block in the breakdown pathway of phenylalanine, an amino acid. Amino acids are the building blocks of protein. All protein that humans eat is made up of some combination of about 20 amino acids. If a child with PKU eats too much normal protein, unmetabolized phenylalanine builds up in the blood and damages the brain. Prolonged elevations of phenylalanine cause profound brain damage...mental retardation. Children with undiagnosed PKU once ended up with severe mental retardation, often literally pulling their hair out and injuring themselves. In the 1940s and 1950s, doctors began to understand PKU and how to treat it with diet. In 1961, Dr. Robert Guthrie of the University of Buffalo developed a test to identify PKU in newborns. Dr. Guthrie's test requires several drops of a baby's blood, retrieved at birth and placed on a small filter paper. Typically the blood is taken from the baby's heel by pricking it with a lancet. The test is itself performed by "growing out" bacteria on the baby's blood spots on the filter paper. If the blood spots contain a lot of phenylalanine, indicative of PKU, the bacteria quickly grow much larger than if there is little phenylalanine.

Dr. Guthrie realized that this test was an effective screen that could prevent mental retardation in children with PKU, and urged public health officials and professional medical associations to assure that all babies be screened at birth for PKU. Health officials ignored Guthrie and the medical community questioned both the screening method and the benefit of early diagnosis of PKU. The medical community and government officials also disputed the cost-effectiveness of screening for such a rare disorder. So Guthrie, many parents of children with PKU, and other advocates for people with mental retardation, went directly to state legislatures to ask for laws requiring PKU screening. In 1963, Massachusetts became the first state to mandate screening for PKU. In time, all other state legislatures followed.

Today, laws require that every baby born in the US and its possessions be screened for PKU. The good news: Over the years, literally thousands of children have been saved a lifetime of disability from PKU. The bad news: When states passed mandatory PKU screening laws, their legislatures entrusted screening responsibility to the states' departments of health. Importantly, that responsibility includes authority to decide which, if any, additional metabolic disorders will be screened by law.

To date, screening for only one other disorder, congenital hypothyroidism (CH), has been mandated in all 50 states. (CH is an endocrine disorder caused by inadequate production of the thyroid hormone thyroxine.) No other metabolic disorder is included in the mandatory screening of all 50 states. In fact, most states screen for only four to six disorders; several screen for only three.

Thus, a baby born in Idaho or Illinois is screened for galactosemia and, even if the disease condition is present, the child is likely to live and develop normally—but not in Louisiana or Washington state, where there is no galactosemia screening. On the other hand, a baby born in Idaho or Illinois with maple syrup urine disease (MSUD) will not be screened for that disorder and is very likely to suffer brain damage or die—but not in Georgia or Pennsylvania, which do screen for MSUD.

Shortly after his PKU screen, Dr. Guthrie developed similar screens using the same heelstick for other metabolic disorders, such as MSUD. He called this "piggybacking." A relatively uncommon disorder like MSUD itself might not justify the cost of screening all babies. Because all babies already are being screened for PKU, the incremental cost to identify kids with MSUD is small enough to merit piggyback screening. Each state made its own decisions about piggyback screening and are, for the most part, inconsistent from state to state.

Tandem mass spectrometry

In 1991-1992, scientists at Duke University completely changed the newborn screening landscape. They adapted an existing technology called mass spectrometry to newborn screening and called it MS/MS, which stands for tandem mass spectrometry. Using the same newborn blood spots obtained for PKU screening, a machine—in one test—can identify dozens of metabolic disorders. MS/MS is much faster than Dr. Guthrie's test and much more sensitive and specific (with far fewer false positive and false negative results). MS/MS took the concept of piggybacking to a remarkable new level. It is important to understand that we are not talking about an additional invasive procedure or an additional test; rather, we are talking here about the same blood spot on the same piece of filter paper!

By 1993, based on techniques developed at Duke, a laboratory at Pittsburgh's Magee-Womens Hospital first began offering newborn screening using MS/MS technology to states and hospitals. This laboratory's complete newborn screening package cost \$15.00 and screened for more than 30 disorders, using both MS/MS technology and other screening methods. The opportunity to save thousands of babies' lives had arrived. But what happened?

State departments of health—part 1

Currently, each state health department in the US decides which disorders to screen. Since 1993, several labs have emerged that are capable of screening large volumes of newborn blood samples using MS/MS. NeoGen Screening in Pittsburgh is the corporate

successor to the Magee-Womens Hospital program that began offering MS/MS in 1993. NeoGen offers comprehensive newborn screening for more than 30 metabolic disorders. Baylor University Health Center in Dallas has for several years offered supplemental newborn screening using some MS/MS to the patients of its physicians and to the community at large. The New England Newborn Screening Program also has provided some MS/MS screening to the New England states for the past several years. Recently the Cord Blood Registry began offering MS/MS newborn screening.

The question is why so few states have elected to use these resources when doing so would protect thousands of children. The answer is that state health departments have a vested interest in protecting their turf, their labs, their jobs and their influence. They are, in short, bureaucracies. And screening in outside facilities by MS/MS presents, in their minds, a threat to them. Though there are no cures for any of the inborn errors of metabolism that are identified by tandem mass spectrometry, all are treatable! Children and their families who are affected by screening decisions made by state health departments are not just statistics. I'd like to introduce you to a few kids.

State departments of health—part 2

I will tell you why I believe Andrew died, and why Bailey was injured; and Michael and Tangi; and thousands of other little boys and girls, and why countless others will continue to die or be injured: The self interest of state health department decision-makers. If MS/MS screening technology has been available since at least 1993, why weren't Michael, Tangi and Bailey screened?

- The answer is not because these disorders are too rare. Screenable, treatable metabolic disorders occur in the aggregate in at least 1 in 4,000 US births. More than 25 years ago, we implemented an entire national newborn screening program for PKU, a single disorder, which occurs in 1 in 15,000 births. It is simple arithmetic. If it is good for PKU why isn't it good for other inborn errors of metabolism?

- The answer is not because these disorders cannot be treated. Every disorder currently detectable by MS/MS is treatable, usually through diet and monitoring. Admittedly treatment is not always successful. But usually the results are remarkably good—so long as treatment begins before injury occurs. No metabolic disorder is perfectly treatable, including PKU, which is screened for universally. Cancer is not perfectly treatable. Neither is diabetes. That doesn't mean screening and treatment should be withheld.

- The answer is not because screening is too costly. NeoGen has offered many states the opportunity to screen all babies comprehensively using MS/MS for the same price (and sometimes less) than the state charges hospitals to screen for only a handful of disorders. Ohio, New York, New Jersey, Massachusetts and other states have rejected these offers. However, Massachusetts has begun a pilot program for extended newborn screening in which 10 screens are routine and the rest are optional. New Jersey recently announced plans to phase in several new screens before the end of 2002. One

reason for the slow introduction in New Jersey is the state's decision to have testing performed by its own state health department. I wonder how many lives will be altered in the meantime.

Health departments almost uniformly have opposed competition by private labs, insisting on a monopoly for the state lab. The reason is simple: If private labs are allowed to compete, state labs will lose that contest immediately and state jobs may be lost, budgets might be cut and, ultimately, the state labs may be downsized or eliminated. Most state labs cannot afford their own MS/MS equipment, and even if they could, they have no one qualified to run the equipment and interpret the results. Rather than give in and farm out the screening work to a more qualified and better-equipped private lab, nothing happens. Well, something does happen: Babies get screened for only a few disorders; some die; some are permanently and severely injured. These are our children: the single greatest treasure we have as a nation and as a people. If you are troubled by this, good. You should be. I am.

Given the statistical prevalence of treatable metabolic disorders, not using MS/MS today to screen babies comprehensively is no different from shooting a gun into a crowded stadium. You cannot know whom you will injure, but it is a safe bet that you will injure someone. Here's what one of the leading experts on newborn screening said (more diplomatically) more than three years ago in the flagship journal of the profession of chemistry:

"If MS/MS technology is such an improvement over presently used methods, with fewer false-positive results and less expense over time because of greater efficiency, why are newborn screening programs not rushing to adopt it? The wrong answer is that the technology is too new or unknown. It has been discussed widely in newborn screening circles and within the academic metabolic community....The correct answer requires an understanding of how newborn screening is conducted in the United States....[T]hose who have knowledge of and expertise in these disorders have no authority in newborn screening, and those who have this authority are usually insufficiently informed and resistant to change – especially when they could be impacted personally by this change....With few exceptions, health department screening programs do not have and are unlikely to be able to acquire and retain the expertise required to adequately perform MS/MS analysis and interpret the results in a timely manner."

—Levy, H., "Clinical Chemistry," 44, No. 12, 1988, pp. 2401-02 (Editorial) (emphasis added).

I submit to you that most state health departments turned their backs on Michael, Tangi and Bailey. And they have turned their backs on your children, your grandchildren and any child you care about. They protected their bureaucracies rather than our kids.

The Sin of Omission

Is there any reason why most parents in this country are never even told that comprehensive MS/MS screening is available? Robert Burke, the father of Bryce Burke, a Texas boy who became brain damaged from MCAD, put it as well as I have ever heard:

"Somebody out there today has a child being born and they're sitting there holding their son or daughter and thinking it's a miracle, so thankful that they have a healthy child, when what they really have is a time bomb waiting to go off. Somebody should have told them...there is a way to defuse it." In Mississippi, the law was changed this spring to require healthcare providers to inform parents that MS/MS comprehensive newborn screening is available for approximately \$20. This law is the result of lobbying by Robin Haygood, the mother of Ben Haygood, a Mississippi 2-year-old who died from MCAD because he was not screened.

In most other states, however, parents do not have to be told of MS/MS screening. Louisiana, Alabama, Texas, and Florida do not offer MS/MS newborn screening; they do not inform parents that it is available. History teaches us that many Ben Haygoods must die and the hearts of many Robin Haygoods must be shattered before the people charged with protecting children in those states will do their jobs.

Operation World Record

I am often asked to speak publicly about newborn screening. Typically, I ask the audience what they think would happen if a foreign government declared its intention to kill or injure 1,000 US babies each year. No one doubts that such a threat would prompt a military response that would cost billions of US tax dollars. I wonder why some people perceive screening any differently, particularly when—putting aside the simple humanity of saving children's lives—screening would save our country money. Children with metabolic disorders who are not screened—and live—cost taxpayers millions in Medicaid and Social Security dollars, far more than the cost to screen all 4,000,000 babies born each year.

The answer, of course, is that those in government typically support only those things that pump money into their bureaucracy and nothing that takes money away—no matter who is hurt. The problem is that the enemy is within. On May 18, yet another family who had lost a child to an unscreened metabolic disorder visited my office. While I looked at these devastated parents, I began to think about the bedtime stories they must have read to their little boy; about his first words; about how these parents must have wanted to protect their little baby; and how they must feel that they failed in that. Even though any failure wasn't theirs.

After a while, I looked down, and on the floor I saw the front page of the Wall Street Journal that I had read that morning. And suddenly it hit me. There was an article about

the "great" state of Texas, the very state in which so many children whose families I knew had died or been injured for lack of screening.

From the article, I learned that the Texas government was quite unhappy that the world-record largemouth bass had been plucked on June 2, 1932 from Montgomery Lake in Georgia. It seems that officials in Texas want the world-record bass to be caught in Texas. Thus, for the past 14 years Texas has spent millions of dollars (with additional millions in private support) to breed massive bass and stock them in the streams and lakes of Texas, virtually assuring that a world-record bass would soon be pulled from Texas water. The government program is code-named "Operation World Record."

Josias Wilson

Joey Wilson was born in late November 1998 in Garland, Texas. Like Michael Metil, Joey was born with GA-1. Although Baylor Hospital in Dallas is one of the hospitals offering MS/MS screening—and was in 1998—Baylor Hospital in Garland never told Joey's parents about the test. Like Michael, Joey now has profound disabilities. His parents cannot afford to get him the care he needs.

I no longer have any answers for parents like the Wilsons, who ask why their little boy was injured because he didn't get a \$20 test that was being offered in a nearby city in a hospital with the same name. I no longer have any response when they ask why they were never even told such a test exists; or why virtually no parent of a newborn child anywhere in the United States is ever told that we have the capability to screen for 30+ inborn errors of metabolism. I cannot explain why gynecologists and obstetricians do not inform couples planning to have children of this capability and what it means. I have no answers to why the media don't spread the word with the same fervor and enthusiasm that they publish the most recent homicide or rape or theft or athletic accomplishment.

But I now know that Texas is soon going to have another world record, and I guess that after that feat, everything will be okay.

•••**Charles P. Hehmeyer** is a medical malpractice lawyer in Philadelphia who represents families of children with metabolic disorders across the country. He is a 1986 graduate of the University of Pennsylvania Law School and a frequent speaker to national metabolic support groups and has appeared in the media advocating expanded newborn screening.

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'The Case for Universal Newborn Screening'

<http://www.eparent.com/healthcare/unbs.htm>

(Please note: If you can, try to get an actual copy of this EP article because the real-life family stories and pictures that we could not reproduce here really bring home the reality and ramifications of NOT being screened at birth! DLG)