Comprehensive Newborn Screening: *The Lifesaving Test You May Never Be Told About*

By Taryn Paladiy, Cate Walsh Vockley MS,CGC, and Jill Levy-Fisch

THE BIRTH OF A CHILD is among the most joyous events we as human beings ever experience. After months of anticipation and preparation, a precious little one joins a family. Parents dream of the things they will do with this new arrival, the birthdays, the family vacations, the continuing of family traditions. Above all, parents hope to provide the best possible life for their new child. Often they've spent their pregnancy trying to create the most healthy, optimal conditions for their child to thrive. The mother often has taken prenatal vitamins and watched her diet. The parents have read about how to best care for their baby, how to raise the child so that he or she will be happy and successful in life. The mother likely went through at least one screening procedure during her pregnancy, if not more.

Imagine, after the child has been born and celebrated, brought home from the hospital, and brought into the bosom of the family, something just doesn't seem right. Perhaps it starts with the baby crying constantly, or not having interest in eating. In what seems like hours, a baby that seemed perfectly healthy is suddenly being rushed to the Emergency Room. Imagine learning that although they did everything right as parents, their baby has suffered irreparable brain damage and will be severely disabled for life. Or worse, imagine that the baby does not survive the medical crisis.

Imagine learning that an incredibly simple and inexpensive screening test done

within 24-48 hours of birth could have prevented it all. Finally, imagine you as expectant parents were never once told of this test which is administered easily to newborns by taking a few drops of blood from the baby's heel and running it through a laboratory test system known as tandem mass spectrometry (MS/MS).

Sadly, parents in the United States experience just such events every year in the United States. The simple test? Comprehensive Newborn Screening. Newborn Screening is a procedure which saves precious lives every year, and yet, it is still widely unknown, and certainly poorly understood. Physicians, specifically OB/GYN's, rarely discuss comprehensive newborn screening with expectant parents. Neither do pediatricians, midwives, or other birthing caregivers. After the birth of a child, hospital staffs rarely explain to new parents the importance or value of Newborn Screening. There is basically a widespread lack of knowledge about the importance of Comprehensive Newborn Screening. And this lack of knowledge is costing lives unnecessarily.

What are Newborn Screening and Comprehensive Newborn Screening?

Newborn Screening is a simple procedure performed in the first 24-48 hours of a baby's life, which tests for TREATABLE inherited metabolic disorders. Through a quick heel prick on your baby's foot, caregivers are able to capture a few spots of blood on a paper card, which is then screened in a state laboratory for a panel of disorders. If babies screen positive for any of these disorders, it raises a red flag. It doesn't necessarily mean your baby has one of these disorders. It allows a diagnosis to be made through further testing. Management and treatment can be started right away, before any adverse effects have had a chance to take place, even while additional testing takes place to confirm the diagnosis. Treatment is often through diet, either through eliminating certain nutrients from the child's diet, or through adding nutritional supplements. Without treatment, babies may seem perfectly healthy at first, but by the time damage is visible, it is often too late, and devastating consequences follow, such as developmental delays, mental retardation, severe disabilities, and even death.

Newborn Screening is not new. Since 1964, babies in the US have been screened for inherited metabolic disorders, starting

with Phenylketonuria, known as PKU. (Because PKU was the first disorder screened for, medical staff may still refer incorrectly to the newborn screening procedure as the "PKU" test.) The current technology for newborn screening, tandem mass spectrometry (MS/MS), is proven, efficient, and effective. With this technology, and due to the advocacy of parents and professionals, the United State leads the world in newborn screening. Thousands of children whose disorders have been identified through newborn screening have gone on to live healthy, productive, "normal" lives because their treatment and maintenance was started soon after birth. Many more have escaped the truly devastating effects of their disorders, and, while they may still have some disabilities, they lead far better lives. As an example, consider a baby born with a biotinidase deficiency. Biotin is an essential vitamin, and if lacking, the baby will cause severe mental retardation. If the deficiency is detected at birth, the simple treatment is to give lifelong biotin supplements orally, much like taking any daily vitamin, to prevent the damage caused by this otherwise devastating disorder.

Unfortunately, however, where you live in the United States matters. Every state currently screens for 29 disorders mandated through the Newborn Screening Saves Lives Act of 2009. These disorders are part of the core uniform screening panel recommended by the Secretary of Health and Human Services' Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC). With the recent addition of SCID by the SACHDNC, the core number of disorders to be screened for is now 30, although most states have not implemented SCID screening yet. Some states go well beyond the original core 29, reporting the SACHDNC's secondary panel of disorders, as well. Currently, several states like Massachusetts and Mississippi screen for more than 50 treatable disorders at birth, while some screen for less than 39. The rest screen for something in between. As many as 10,000-12,000 babies are at risk every year, due to incomplete screening.

It is important for expecting military families to note that the policy of the U.S. Military is to provide for newborn screening in accordance with the state mandates where the family is located. Families who have been moved frequently and have more than one child may learn that their children have been screened for different disorders, depending on the state in which the child was born. It is crucial for military families to learn about newborn screening in the state where their child will be born, and to plan for Comprehensive Newborn Screening when necessary.

Comprehensive Newborn Screening is screening that parents can request in states where not all disorders are included in the state screening program. In some states, parents can simply request Comprehensive Newborn Screening (CNBS) in the hospital when their child is born, while in other states, CNBS can only happen if parents prepare carefully for it, discussing it with their prenatal and birthing caregivers, contacting the hospital prior to birth to ensure that the screening will be performed, negotiating with their insurance companies to ensure that the laboratory work will be covered.

"It is a crime that babies are falling through the cracks, not being screened for all treatable disorders," says Jill Levy-Fisch, President of the Save Babies Through Screening Foundation, the only non-profit in the US devoted exclusively to advocating for universal newborn screening. "The technology for screening all babies exists. It is inexpensive, especially when you compare it to the costs of caring for a child whose treatment is delayed due to lack of screening. How can such a simple, inexpensive screening not be performed on all babies born in this country?"

Lack of Knowledge, and Misinformation: More Babies at Risk

As many as 12,000 babies are at risk every year, and yet, where is the outcry? While statistically, this may seem insignificant, no parent looks at their newborn baby as a statistic. Why are so few parents, parents who took advantage of every screening possible during their pregnancy, silent

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About Save Babies Through Screening Foundation

As the only national non-profit organization devoted exclusively to newborn screening advocacy, the mission of the Save Babies Through Screening Foundation is to improve the lives of children by working to prevent death and disabilities resulting from disorders detectable through newborn screening tests. The Foundation is a leader in the national grassroots advocacy movement and media awareness and actively participates at the local, state, and federal levels to improve newborn screening. The Foundation's goal is to see that every child born is screened successfully, effectively and comprehensively.

Save Babies Through Screening Foundation, Inc.

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What To Do if You Are Expecting:

- Find out how many disorders are on your state screening panel (visit www.savebabies.org for an up-to-date listing.)
- Discuss Comprehensive Newborn Screening with your prenatal caregiver and birth hospital labor and delivery staff.
- Talk about it with grandparents and relatives. They can help with affordability and perhaps "Give a Gift of Life" by paying for a Comprehensive Newborn Screening Kit that can be used in any hospital or birthing clinic in the United States.
- Plan with your caregiver and hospital how Comprehensive Newborn Screening will be administered at the time of your child's birth, if needed, and appoint someone to ensure that the plan is followed (husband, a family member, a birthing assistant, etc.)
- Follow-up! Ask your caregiver about the results of your baby's newborn screening. If anything seems out of the ordinary, ask about it. Do not stop asking questions until you fully understand the results.

when it comes to advocating for better screening at the state level once their child is born? Why do so few parents request Comprehensive Newborn Screening? The answer is: they do not know about it nor have they been taught about the significance. Can you possibly conceive of a parent taking their baby home from the hospital ready to tackle the challenges of parenthood not knowing that he or she had biotinidase deficiency; then finding out four or six months later that their son or daughter was destined for a life with severe intellectual disabilities? Then learning that this could have been easily avoided? The thought is both offensive and discouraging.

First, there is a widespread lack of knowledge about Newborn Screening. When expecting parents hear the word "inherited", many assume that newborn screening does not apply to their baby, since they have no known family history of any of the disorders. What they do not realize is that very often there is no family history. Parents also incorrectly assume that if they have already had one or more children, that their new baby is not at risk, but this is also not true. It is not uncommon for parents to have several healthy children before having a child born with a disorder which can be identified through newborn screening. The Larks family from Colorado has lived this experience; after having 5 healthy children their 6th child, Damian, was identified through newborn screening as having 3MCC.

Unfortunately, while prenatal (obstetricians, midwives, etc.) and post-natal caregivers (neonatologists, nurses, pediatricians, etc.) are aware of newborn screening and may think they communicate to parents about it, many expectant parents never even know that newborn screening will take place. A brochure about NBS given in a packet of prenatal materials is just not enough! Even fewer expectant parents know about the option and importance of CNBS, for the same reasons. Staffs in hospitals often do not offer any information on the availability or importance of additional newborn screening in states were Newborn Screening is incomplete. Even caregivers performing the test sometimes do not fully understand its value, and, as such, screening samples can be mishandled, positive screens misinterpreted.

As if lack of education were not enough, there is also a small, but highly vocal group of individuals who have hijacked the newborn screening system in several states, turning the focus away from the real issue of saving babies' lives, and focusing on privacy issues. Such groups are suggesting to parents that it is more important to protect the genetic privacy of their babies than to protect the same babies from the potentially devastating consequences of lack of screening. Sadly, the tactics of such groups often misdirect and confuse expectant parents at a time when it is essential for them to understand the importance of newborn screening for their newborn baby. While the authors do not presume to judge the validity of privacy concerns, we decry any effort to change the Newborn Screening System which will cost the lives of babies.

Nicholas's Story

Nicholas was born on June 4th, 2010. After a very "normal" pregnancy, his family was not surprised to welcome another healthy and perfect little baby into the world. His parents were elated to be blessed with two healthy children, their 20 month old daughter Lia, and their new baby boy, Nicholas.

What should have been one of the happiest days of their lives, the day they were to leave the hospital with their son and welcome him to his home, turned into the beginning of a nightmare. Just as his parents were packing him into his car seat, a doctor came into the room and told them that he had some bad news. Nicholas had an extremely low T-cell count, and the doc-

tor said he most likely had Severe Combined Immune Deficiency (SCID). Babies born with SCID are born with no immune system. The mildest infection is devastating and often fatal. For those who survive a few infections, often by the time of diagnosis, damage to major organs and bodily functions is so extreme that even with diagnosis and late treatment, children rarely live long.

Nicholas was sent immediately to a leading children's hospital, where he underwent confirmatory diagnostic testing. His family was allowed to take him home, but he had to be completely isolated.

Several weeks later, the family learned that Nicholas did in fact have SCID. The only known cure for him was a bone marrow transplant, and the doctors were already working on finding him a suitable donor. His parents were absolutely terrified. Their baby, who on the outside looked as healthy as could be, was unable to live without needing a bone marrow transplant.

The best possible outcome for Nicholas was for him to undergo his transplant before he was 3 months of age, and while he was infection free. Fortunately, when he was just one month old, his doctors found a suitable donor through the National Bone Marrow Registry. At 7 weeks of age, Nicholas received his new bone marrow through an infusion. After a grueling 3-month hospital stay his family was allowed to bring him home. He had to be secluded from the outside world for 8 months, but every week during visits to his doctors, his family was given excellent news about his new immune system, which was growing and working wonderfully! The transplant was working. Instead of no immune system at all, Nicholas was developing an immune system that would enable him to lead a normal life!

Nicholas is now a 14 month old healthy, thriving, precious little boy. He has a fully functioning immune system, just as any other healthy child!

Unfortunately, Nicholas's outcome is rare for babies born with SCID. Because most states do not mandate newborn screening for SCID, most babies die from infection before they are diagnosed. In January 2010, the SACHDNC recommended the addition of SCID to the uniform newborn screening panel. The recommendation was made after significant research and piloting of SCID screening which showed the overwhelming effectiveness of screening in creating positive outcomes for children born with SCID. However, many states have not yet adopted the SACHDNC's recommendation, and do not yet screen for SCID. Nicholas was one of the first babies, to be diagnosed with SCID through Newborn Screening in Massachusetts, a state which started screening for SCID on a trial basis in 2009. It was because of his prompt diagnosis and organized medical care that he is the healthy child he is now, and he serves as proof that newborn screening is absolutely necessary.

Cassidy's Story

Cassidy was born on September 18, 2001. Her family was overjoyed that the beautiful girl they had so carefully prepared for had finally arrived. Her first year was a delight. She developed as expected, sitting at the age of 6 months, beginning to talk at 8 months, walking, even running, by 12 months. She was a happy toddler who loved to play and explore her world. Unfortunately, at the age of 17 months, Cassidy's life changed forever.

It started with a stomach flu. Cassidy was very sick and could not keep any fluids or foods down. Her parents took her to the emergency room for help. They were sent home when she seemed to revive, but a few days later they were back in the hospital with a child who was barely recognizable and no answers as to what was wrong. Test after test, and Cassidy's parents watched their daughter change, lost under wires and monitors, now unable to sit up, unable to suck her pacifier, unable to blink. Eventually, they learned that their daughter had glutaric academia, type 1 (GA1).

If Cassidy had been screened for GA1 at birth, she could have been placed on a protein restricted diet, fed special supplements, and hospitalized for fluids during illnesses such as the stomach flu. Instead, her family needed to learn how to care for her and try to help her recover from the brain damage she had suffered. Cassidy now has a feeding tube, can no longer sit up, walk, talk, or even hold her own head up. Cassidy's family loves her dearly and fills her days with as much smiles and happiness as they can give her. But they will always wish that she had the chance to live the life she was leading. Thankfully, GA1 is now on the screening panel for most states. Research and development of new tests like MS/MS (noted above) have improved the NBS system and led to identification of more affected children before devastation from an undiagnosed serious disorder occurs.

What is being done?

"Education is key. Awareness is vital. If more physicians, parents, and nurses were educated about newborn screening, there would be much less chance of needless, preventable devastation," said Anita Bailey. "Our daughter Cassidy lost her chance at a "normal" life, because we did not know

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there were other metabolic diseases out there she could, should have been screened for. We would have opted for more tests even if they were not paid for by the state. I opted to have the "triple screen" and multiple ultrasounds during my pregnancy. But I did not know that we were missing so much vital information!"

It is only through the advocacy of parents and dedicated professionals that the country's Newborn Screening program has developed as widely as it has today. The Save Babies Through Screening Foundation (SBTS) was formed 13 years ago, and today is the only non-profit organization in the U.S. dedicated exclusively to advocating for universal Newborn Screening. Formed by passionate parents who have been affected both negatively and positively by Newborn Screening, the organization is at the forefront of many advocacy efforts throughout the country. SBTS was intimately involved in the writing and passage of the Newborn Screening Saves Lives Act of 2007 that provided for education for parents, outreach to health care providers, and appropriate follow-up for newborn screening.

SBTS has also created countless educational materials, including, most recently, the awareness film "One Foot at a Time," which will be an educational tool available and utilized across the country. SBTS wants each parent and expectant parent to be an active part of their baby's newborn screening test. To do this, parents need to understand a lot about the screening process—the benefits and potential risks of screening, how and when the screening is done, how to learn your baby's results, and what it means and what to do if the screen is positive. SBTS has developed the video to help parents understand all of this and more, and to share the experiences of parents who have gone through the newborn screening and follow up process. You can view a 2-minute preview of the SBTS video by clicking the box below to activate the video. The entire 15-minute video is available for you to see on the Save Babies Through Screening Foundation website at www.savebabies.org/eduvidurl.

Click here to see a 2-minute preview of the SBTS video mentioned above.

SBTS provides a crucial service to parents looking for information on how to get Comprehensive Newborn Screening in their state. Their website provides information on which disorders are screened for in each state, and they also provide free Comprehensive Newborn Screening test kits to families in need. (See their website, www.savebabies.org, for more information.)

The SACHDNC continues to review the validity and effectiveness of screening for additional disorders, working closely with medical professionals, researchers, and family advocacy groups to ensure that no stone is left unturned in providing the best chance to every baby born in the United States. State Newborn Screening Committees are refining their screening protocols constantly, responding to existing and developing needs.

But still more needs to be done.

As a nation, we need to be more demanding of those who are entrusted to provide our care. They are supposed to provide explanations to us in response to our questions, but we in turn have the burden of asking the proper questions, which means we need to become more knowledgeable about comprehensive newborn screening. Knowledge must be spread throughout the country, from obstetricians to midwives, from Lamaze instructors to labor nurses, from neonatologists to NICU nurses. The more people are aware of the importance of newborn screening, the more we will be able to ask the right questions at the right time, and the more newborn screening will become part of the public consciousness.

While the United States leads the world in Newborn Screening, it is unacceptable that the number of conditions screened for varies by state, allowing thousands of babies to be brought home from the hospital at risk every year. We must demand comprehensive screening for every child in every state, advocating state-level decision-makers and stakeholders. Together, we must work to ensure that no child suffers or dies from a condition that could have been treated and managed if identified at birth. Together, we can strive towards a day when parents will not have to look back and say, "If I only had only known." •

Taryn Paladiy is a writer and development consultant who has served as Secretary of the Board of the Save Babies Through Screening Foundation since 2009. She is the mother of a child whose health was severely compromised due to mishandling of her newborn screening. Thankfully, her child has overcome the consequences of late diagnosis, but this experience has led Ms. Paladiy to advocate for universal newborn screening.

Cate Walsh Vockley, MA, CGC, is a certified genetic counselor working at Children's Hospital of Pittsburgh. She is a member of the SACHDNC Education and Training Subcommittee and is involved in research on the natural history of disorders diagnosable by NBS.

Jill Levy-Fisch is President of the Board of the Save Babies Through Screening Foundation. She has been an active advocate for Comprehensive Newborn Screening at the state and federal levels for 8 years, serving on various committees, including a subcommittee of the SACHDNC. She is the mother of three children, two of whom have a disorder detectable through newborn screening.

Resources:

Save Babies Through Screening Foundation
www.savebabies.org

Secretary's Advisory Committee on Heritable Disorders in Newborns and Children
www.Hrsa.gov/heritabledisorderscommittee/

National Newborn Screening and Genetics Resource Center
http://genes-r-us.uthscsa.edu/

American College of Medical Genetics
www.acmg.net

National Human Genome Research Institute
www.genome.gov

Organic Acidemia Resources: Bailey Baio Angel Foundation
www.baileybaioangelfoundation.com

SCID Resources:

Immune Deficiency Foundation
www.primaryimmune.org