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Health, Education, Labor and Pensions Subcommittee on Children and Families Hearing
Newborn Screening Saves Lives: The Past, Present and Future of the Newborn Screening System

Thursday, September 26, 2013

Good morning Chairwoman Hagan, Ranking Member Enzi, and members of the Subcommittee on Children and Families. My name is Dr. Jennifer Howse, and I'm proud to serve as president of the March of Dimes Foundation, a unique partnership of scientists, clinicians, parents, members of the business community and other volunteers affiliated with 52 chapters and over 200 divisions in every state, the District of Columbia and Puerto Rico. I appreciate this opportunity to testify today on newborn screening, one of the great public health victories of the 20th century, and one which continues to save infants' lives every day.

The March of Dimes is a national voluntary health agency founded in 1938 by President Franklin D. Roosevelt to support research and services related to polio. Today, the Foundation works to improve the health of women, infants and children by preventing birth defects, premature birth and infant mortality through research, community services, education and advocacy. In 1998, the March of Dimes established its Global Programs division to extend its mission overseas through partnerships with countries to deliver interventions directed at reducing birth defects and preterm birth.

Background

Newborn screening is a critically important and highly effective public health program for testing every newborn for certain genetic, metabolic, hormonal and functional conditions not otherwise apparent at birth. Approximately 1 in every 300 newborns has a condition that can be detected through screening. Newborn screening detects conditions that, if left untreated, can cause disabilities, developmental delays, illnesses or even death. If diagnosed early, many of these disorders can be managed successfully, which not only reduces the physical burden of disease but can also help to reduce the associated economic burden on families, communities, and government.

Since the mid-1960s, the success of newborn screening programs has led to routine testing for the over four million infants born in the United States each year. The Centers for Disease Control and Prevention (CDC) estimates that each year over 6,000 newborns are diagnosed as having a treatable metabolic condition and another 12,000 are found to have hearing impairment that requires follow up. The majority of newborn screen tests are performed using a single sample of a few drops of blood from the newborn's heel, usually taken in the hospital 24 to 48 hours after birth. Hearing screening and screening for critical congenital heart disease (CCHD) are performed with non-invasive devices; hearing screening utilizes a handheld device held near the infant's ear, while pulse oximetry is used to test for CCHD by way of a small probe that clips onto a newborn's hand or foot.

History of Newborn Screening

This year, our nation is celebrating the 50th anniversary of newborn screening; however, the program's origins reach back much earlier. In 1959, after the March of Dimes had led our nation to the successful development of the Salk and Sabin polio vaccines and refocused our mission on birth defects prevention,

we initiated discussions about newborn screening on a large scale as a means to detect and prevent the catastrophic consequences of metabolic conditions such as phenylketonuria (PKU). This led to a grant to Dr. Robert Guthrie to support his development of a simple and effective population-based screening test for PKU. Dr. Guthrie's work demonstrated conclusively that identifying infants with PKU and immediately beginning a low-protein diet could completely avert the otherwise devastating developmental disabilities PKU causes. These results were so dramatic that the state of Massachusetts mandated PKU screening for all infants in 1968, beginning the modern era of newborn screening.

Subsequently, the March of Dimes funded research into tests for other genetic and metabolic diseases in newborns as we promoted newborn screening as a central component of newborn medical care. The Foundation is deeply proud of our decades-long history of funding research that has led or contributed to the development of numerous newborn screening tests, including those for congenital adrenal hyperplasia, biotinidase deficiency, and others. Together, these tests have allowed us to preserve the health and wellbeing of thousands of children.

As more tests became available, however, a patchwork developed in which some states screened for numerous disorders and other very few. In 2000, the March of Dimes led the way in proposing a national standard for newborn screening which included a core list of 9 disorders, with provisions for expanding the list as science and technology evolved. At the same time, the March of Dimes and others in the policy community began working with Congress to bring new attention and focus to this rapidly developing field. We worked to identify policy changes that would allow the federal government to assist states in evaluating new tests and determining whether to include them in their screening panels. The landmark Children's Health Act of 2000 (P.L. 106-310) included two vital provisions that advanced newborn screening policies. The law created the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children to provide expert evaluations of new tests and consideration of challenges in the field. It also established federal grants to enhance and evaluate state newborn screening programs, allowing them to develop and implement best practices.

In August 2004, the American College of Medical Genetics (ACMG) submitted a report requested by the Health Resources and Services Administration (HRSA) setting out proposed nationwide standards for state newborn screening programs. The report listed 29 core treatable disorders that should be targeted directly and an additional 25 secondary conditions for which test results should be reported. These secondary disorders were not directly targeted by newborn screening because they did not yet have documented treatments or because there was limited knowledge of their natural history. Their presence would be revealed, however, in the course of screening for the core conditions. The ACMG recommendation to screen all newborns for 29 core conditions was endorsed by the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children as well as the March of Dimes in 2005.

The federal Recommended Uniform Screening Panel (RUSP) gave advocates a powerful tool to press state legislatures to adopt this consistent set of tests. The March of Dimes led a grassroots advocacy campaign to secure adoption of the recommended uniform panel in every state, issuing annual report cards to document progress. And it was spectacularly effective: in 2004, only 21 states screened for at least nine of the recommended conditions, but just four years later all but two states were screening for at least 21.

Since 2010, the Advisory Committee, with the Secretary's approval, has added two new conditions to the Recommended Uniform Screening Panel: severe combined immunodeficiency (SCID) and critical congenital heart disease (CCHD). A third condition, Pompe Disease, is currently awaiting a decision by the Secretary. This year alone, the March of Dimes and allies like the American Heart Association have advocated successfully for 24 states to add CCHD to their newborn screening panels. This system of review and recommendations by the expert Advisory Committee, approval and dissemination by the HHS Secretary, and adoption by the states continues to work effectively to ensure that tests are evaluated appropriately and then adopted in a timely fashion to protect the health of our nation's infants.

The Newborn Screening Saves Lives Act

The remarkable progress of newborn screening over the past two decades persuaded Congress to pass the Newborn Screening Saves Lives Act in 2008. The law renewed and updated various programs that underpin states' newborn screening efforts as well as the Secretary's Advisory Committee. Most notably, it codified the authority of the Secretary of Health and Human Services to establish the Recommended Uniform Screening Panel and to accept or reject the Advisory Committee's recommendations to add conditions to the RUSP. The law is now due for its regular five-year renewal.

The March of Dimes is deeply grateful to Subcommittee Chairwoman Kay Hagan and Senator Orrin Hatch and Representatives Lucille Roybal-Allard and Mike Simpson for introducing S. 1417 and H.R. 1281, the Newborn Screening Saves Lives Reauthorization Act. Reauthorization is critical to ensuring we continue to provide the most accurate and comprehensive screening available to our nation's children.

Passage of the Newborn Screening Saves Lives Reauthorization Act is essential to the continued success of newborn screening programs across our nation. Most importantly, reauthorization will ensure the uninterrupted continuation of the Secretary's Advisory Committee on Heritable Disorders and its work. The Advisory Committee's charter expired in April of this year, and it was only through the timely action of Health and Human Services Secretary Kathleen Sebelius that it was extended on a discretionary basis for up to an additional two years. Maintaining and updating the Recommended Uniform Screening Panel that states use to adopt and implement new conditions is vital, and ongoing and planned evidence reviews should not be delayed.

The Newborn Screening Saves Lives Reauthorization Act also extends important grant programs at the Health Resources and Services

Administration, Centers for Disease Control and Prevention and National Institutes of Health, including:

- Seven Genetics and Newborn Screening Regional Collaborative Groups (RCs) and a National Coordinating Center (NCC) funded by HRSA, which strengthen and support the genetics and newborn screening capacity of states using a regional approach to addressing mal-distribution of genetic services and resources. Special emphasis is given to underserved populations and those families and providers in rural areas. The RCs include all states, U.S. Territories and the District of Columbia.
- The Critical Congenital Heart Disease (CCHD) Newborn Screening Demonstration Program, a three-year HRSA grant designed to support the development, dissemination and validation of screening protocols and newborn screening infrastructure for point of care screening specific to CCHD. CCHD presents special challenges to implementation since it is not tested with the blood spot.
- *Baby's First Test*, a national educational resource center for newborn screening presently operated by Genetic Alliance under a HRSA grant. *Baby's First Test* informs and empowers families and healthcare providers throughout the newborn screening experience.
- The Newborn Screening Technical Assistance and Evaluation Program (NewSTEPs) funded by HRSA, which serves as a technical assistance program for state newborn screening systems.
- Newborn Screening Quality Assurance Program (NSQAP), a comprehensive CDC program devoted to ensuring the accuracy of newborn screening. NSQAP is the only comprehensive program in the world devoted to ensuring the accuracy of newborn tests. In 2012, the program guaranteed the quality of newborn testing in more than 550 laboratories worldwide, and assured identification of between five and six thousand infants with treatable diseases who might have otherwise died or become severely disabled.
- The Hunter Kelly Research Program, which supports numerous grants and contracts to develop and improve technologies related to newborn screening. Through the Hunter Kelly Newborn Screening Research Program, the Eunice Kennedy Shriver National Institute of Child Health and Human Development also funds the Newborn Screening Translational Research Network, a resource for investigators engaged in newborn screening related research.

Conclusion

Today, 42 states and the District of Columbia require screening of at least 29 of the 31 treatable core conditions. Millions of babies have been screened for dozens of disorders, and in thousands of cases, the health and wellbeing of those children has been preserved. Newborn screening represents a model federal-state public health partnership that has produced extraordinary improvements in child health.

We must not allow this vital public health effort to falter. Our most immediate challenge is to preserve and renew the Newborn Screening Saves Lives Act. On behalf of over 3 million March of Dimes volunteers and countless other organizations and

families, I urge Senators to cosponsor and support S. 1417 and the Committee to report the legislation. We look forward to working closely with the committee and chamber leadership to ensure it can be passed as soon as possible in both the Senate and the House. Furthermore, although beyond the jurisdiction of this Committee, I urge Congress and the Administration to agree on a balanced approach to deficit reduction that protects investments in programs such as newborn screening. Authorization bills are only effective insofar as funding is appropriated to implement their provisions.

Newborn screening has improved and saved the lives of countless thousands of affected children. Thank you for your attention to this vitally important child health issue. The March of Dimes stands ready to assist you in ensuring that newborn screening programs will continue to preserve the health and wellbeing of newborns for many years to come.