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Subcommittee on Children and Families

Subcommittee Hearing-Newborn Screening Saves Lives: The Past, Present and Future of the Newborn Screening System

Testimony of

R.Rodney Howell, M. D.
Professor of Pediatrics
Member, Hussman Institute for Human Genomics
Miller School of Medicine, University of Miami
Miami, Florida

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Chairwoman Hagan, Senator Enzi and members of this subcommittee thank you very much for inviting me to testify today on Newborn Screening Saves Lives: The Past, Present and Future of the Newborn Screening System. I am a pediatrician who specializes in genetic disorders that produce serious biochemical abnormalities in children and was beginning my career at Johns Hopkins when newborn screening was beginning in Maryland the 1960s. I have had the opportunity to see first-hand for over nearly 50 years the remarkable accomplishments of our newborn screening programs in the United States.

Children with an inherited condition known as phenylketonuria, or PKU, if untreated have profound developmental delay with an average IQ of less than 20. This means that such untreated children, who have a normal lifespan, are unable to speak or care for even simple needs, and require full-time care. They are robbed of many of life's opportunities. Over 50 years ago, it was shown that babies with PKU identified at birth and treated with a very special diet could grow into adults with normal abilities. Dr. Robert Guthrie at the State University of New York in Buffalo solved a key problem, and developed a reliable, inexpensive test that could be done on all babies born in this country. This led to the beginning of newborn screening, which is carried out in every state under the leadership of the individual state health departments.

The use of the Guthrie test, or the PKU test, fairly quickly spread throughout the United States. And this week, we are celebrating the 50th anniversary of our Newborn Screening program. Since the beginning, newborn screening has been carried out under the aegis of the State Health Departments and has always been among the most successful preventive health programs in this country. And today, we have thousands of adults, treated for PKU from infancy functioning well in all the walks of life.

Since the benefit of the early diagnosis and treatment of PKU was so very dramatic, individual states, which are responsible for newborn screening, began to add tests for other conditions, using the same blood sample, to their newborn screening programs. Such conditions as congenital hypothyroidism were among the more common additions since early diagnosis and treatment of this condition also can prevent substantial developmental delay. But since each state has its own advisory panels, there developed considerable variation among the states. This variation was not only in the specific conditions being tested, but also the numbers of conditions included in the screening panel. In other words, whether your child would be identified to have a serious medical condition and receive the necessary life-saving medical intervention simply depended on the state in which your baby was born. This became a big problem for at-risk families who moved to another state between pregnancies. It was a lottery that the public health system never intended and consistency between the states needed be established.

Early efforts at harmonization of screening panels between states began when the Maternal and Child Health Bureau/HRSA charged the American College of Medical Genetics to evaluate the scientific and medical information related to screening for specific conditions, and to make recommendations based on this evidence. They convened an expert group which produced a report which recommended a uniform screening panel and system.

Then Title XXVI of the Children's Health Act of 2000 enacted sections of the Public Health Service Act which established the Advisory Committee on Heritable Disorders in Newborns and Children (Committee), which held its first meeting in 2004.

The Advisory Committee on Heritable Disorders in Newborns and Children was established to provide advice to the Secretary of Health and Human Services on newborn screening. It was my privilege to serve as the Founding Chairman of this Committee and continue in this role for the Committee's first eight years. When the Committee first began its work, there was extraordinary variation among the states in screening programs. In the year 2000, 35% of the states were testing for fewer than 5 conditions, and 65% were testing for 5-10 conditions—none were testing above this number. Early in its work, the Committee after careful review and study accepted the report of the American College of Medical Genetics and recommended that the more than 4,000,000 babies born each year in the United States be tested for 29 specific disorders including certain metabolic, and hearing deficiencies in early 2005.

It has been most gratifying to see how the various states have responded to recommendations from the Advisory Committee. Although states are responsible for their own screening programs, and virtually every state has an advisory committee that oversees decisions for that individual state, it is extremely difficult (even for large states) to have the extensive expertise required in the evaluation of these individually rare inherited conditions. The Advisory Committee membership contains or has access to all the required expertise. The legislation under which the Advisory Committee works also requires that all recommendations for inclusion in the newborn screening panel be evidence based. As the Committee has made recommendations, the states have been extremely responsive in reviewing these recommendations in light of their own needs, and in virtually every situation has adopted the recommendations of the Advisory Committee.

The Committee has established a program for the recommendation of other conditions to be added to the recommended uniform screening panel, or the RUSP. It is felt that any individual, group, or organization should be able to submit a nomination to the Committee for a condition to be added to the recommended RUSP. In order to accomplish this the Committee (*http://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders*) has developed a form outlining the exact information needed and directions for presenting such a nomination. To date, 10 completed nominations for new conditions to be considered for addition have been submitted to the Committee. After careful review by the Committee, and evidence review that would be necessary for consideration for newborn screening, three additional conditions have been recommended by the Committee for addition to the RUSP. The Secretary of HHS has approved two of these (severe combined immune deficiency and critical congenital heart disease) and is currently considering the recommendation of the third, Pompe Disease.

It is important to emphasize that the conditions that are included on the newborn screening panel all result in serious medical complications (e.g. developmental delay) and/or death if not recognized early. All children with these conditions benefit from early diagnosis and treatment.

Since the passage of Public Law 110-204 in 2008 (Newborn Screening Saves Lives Act of 2008) there has been great harmonization among the states, and at the end of 2010, 100% of US births were screened for over 30 conditions. And as a result of these expanded screening programs lives have clearly been saved.

The current implementation by the states of the core panel of conditions (not including severe combined immune deficiency and critical congenital heart disease both of which are in the process of being implemented across the country) will identify 5,064 children with hearing loss, 2,156 with primary congenital hypothyroidism, 1,775 children with sickle cell disease, 1,248 children with cystic fibrosis, and 239 children with medium-chain acyl-CoA dehydrogenase deficiency, and other important conditions for a total of 12,500 children yearly whose lives will be either profoundly altered or saved due to newborn screening.

The Secretary's Advisory Committee has worked tirelessly to meet the nation's public health needs and the needs of our children. I am particularly proud of the rigor that it has applied to the evidence review of conditions that have been nominated for consideration to the Committee. As a physician and a geneticist, I am equally encouraged by the therapeutic pipelines in development that represent great promise of new science and hold potential that we many help many more families and children. Certain of the mucopolysaccharide storage diseases are well-positioned, with new approved therapies, to be considered for addition to the newborn screening panels.

There are many new opportunities on the horizon but two come to mind. Two examples of how advances in science will impact newborn screening in coming years are Duchenne Muscular Dystrophy and spinal muscular atrophy. Both of these disorders result in profound and devastating health consequences for the affected children. In both these conditions, drug therapies are currently under development which will likely be of the greatest benefit if administered, presymptomatically, which will be very soon after birth. The availability of newborn screening programs for these disorders will be essential to benefit maximally from any new treatments.

The NIH Hunter Kelly component of the Newborn Screening Saves Lives Reauthorization of 2013 is an essential part of the legislation that will support research needed to develop new therapies for conditions for which we currently lack treatment. Some of our vexing conditions in the newborn, which we could readily detect through newborn screening, currently lack safe and effective treatment.

Now that we have treatments for conditions that can be diagnosed and treated as a result of newborn screening, we need additional support for the study of the long-term outcomes of infants treated as a result of newborn screening.

As other conditions are recommended for addition to the RUSP, we will need to identify funding and partners for large pilot research projects prior to the implementation of a program throughout the country. Prior to the full implementation of the newborn screening for severe combined immune deficiency, a large pilot study was carried out that was a great example of cooperation between the public sector organizations, and a not-for-profit Foundation.

Public information about newborn screening has been recognized for a long time as not only important but lacking. Some public concern about the use of residual blood samples has in my opinion been linked to a lack of understanding about the program itself. The HRSA Clearing House for Newborn Screening Information and the National Newborn Screening and Genetic Resource Center will go a long way to address these needs.

The CDC Newborn Screening Quality Assurance Program is known throughout the world for its excellent work. This program has been, and remains, vital to the entire newborn screening program. As I travel the United States as well as Europe, Asia and the Middle East to meet with local leaders dealing with newborn screening, this distinguished program is routinely identified as vital. And this group's provision of quality assurance materials is essential to the development of new tests, and the assurance that our testing procedures are working well.

It is critical to the health of our infants that the nation's newborn screening programs be reauthorized with the passage of the Newborn Screening Saves Lives Reauthorization Act of 2013.

In consideration of the life-altering potential advances on the near horizon for so many of our nation's children, I want to call particular attention to the new "Priority Review" section of the legislation which serves to strengthen the federal newborn screening program. In our current newborn screening programs, we are regularly concerned with delays of days during which an affected infant, if not identified, can die or be damaged. And in considering new treatments, if there is a beneficial treatment to be considered, delays mean lost lives.

Under the reauthorization, there will be consistent and predictable time period allowed completing the evidence review process. It will be most important that we work to ensure that sufficient funds are available for these costly and intense evidence reviews required by the Committee. I believe that the impact of these timelines will encourage nominees to develop and submit more complete nomination packages and will provide the review committee an appropriate period of time to thoroughly and completely review the nomination to determine whether the condition meets all of the critical scientific standards necessary to warrant addition to the RUSP. It will require a lot of hard work, and of course we cannot afford any shortcuts since babies lives are at stake.

Equally important, this legislation will encourage the committee to more closely align its activities with the development of new and emerging interventions to narrow the gap between the approval of new treatments and the ability to identify the babies who could be saved if identified through newborn screening—again without undermining or diminishing the role of science in the committee process.

The individuals who serve on the Secretary's Advisory Committee do an incredible job of balancing limited public health resources with the goal of identifying babies who could benefit from newborn screening. Not only does newborn screening save lives, the program actually represents overall cost savings to the American healthcare system especially important at this time of extraordinary restricted funds. Medical interventions following newborn screening can prevent or ameliorate severe, childhood-onset diseases and reduce the financial burden of intensive care hospitalizations.

SCID (severe combined immune deficiency) where infants are born lacking an immune system provides a very clear case study demonstrating the importance of newborn screening. If a baby with SCID is not diagnosed at birth, the outcome is death in infancy but only after weeks or months in a hospital intensive care unit battling life-threatening infections. In addition to the enormous emotional burdens to families as well as lost time at work for parents there are medical bills that routinely exceed hundreds of thousands of dollars. Unfortunately, in the end it is common that a baby with SCID doesn't survive this long hospital ordeal, so the devastating loss of a child is added to the family's burden. On the other hand, if a baby with SCID undergoes newborn screening and is identified at birth at a cost of no more than $20, the baby can receive a life-saving umbilical blood transplant in the outpatient clinic over a period of days at a total cost of around $50,000.

We are at a unique point in history. The mapping of the Human Genome is now complete. Genetics has moved out of the laboratory and into the clinic, where its applications can save lives every day. The current progress in the development pipeline of genetically targeted therapies is tremendous.

I am very proud of the Committee's work and thoroughness and believe that S 1417 builds on the accomplishments of the newborn screening program and will allow the Committee to continue to deliver the latest evidence- based diagnoses and treatments for now and in the future which holds tremendous promise for genetically based therapies that will benefit our nation's children and their families.