TESTIMONY OF
JOSEPH A. BOCCHINI, JR, MD
PROFESSOR OF PEDIATRICS
LOUISIANA STATE UNIVERSITY HEALTH SCIENCES CENTER AT SHREVEPORT

BEFORE THE ENERGY AND COMMERCE SUBCOMMITTEE ON HEALTH HEARING
"REAUTHORIZING VITAL HEALTH PROGRAMS FOR AMERICAN FAMILIES"
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Madam Chairman Eshoo and Ranking Member Burgess, Health Subcommittee Members

My name is Joseph Anthony Bocchini, Jr. I am a pediatrician and a pediatric infectious diseases specialist. I serve as a Professor of Pediatrics at Louisiana State University Health Sciences Center in Shreveport.

I have recently had the privilege of serving an eight-year term as Chairman of the Advisory Committee on Heritable Disorders in Newborns and Children, the Advisory Committee established with the original authorization of the Newborn Screening Saves Lives Act in 2008 and continued in its reauthorization in 2014. I have seen the benefits of this Act through the eyes of the Advisory Committee, in my practice, and in the infants whose lives have been improved and, in many cases, saved, through the prompt diagnosis and treatment of conditions which untreated, can cause serious health problems in infancy or childhood.

I am pleased to provide testimony today in strong support of the Newborn Screening Saves Lives Reauthorization Act of 2019 (H.R. 2507). This is a critical piece of legislation which supports the activities of one of the most successful public health disease prevention programs in the United States. The activities imbedded in this reauthorization provide the additional infrastructure needed to build on the successes of the past 11 years and address how to continue to adapt and adjust this rapidly growing and changing program.

Newborn screening began 56 years ago with the development of a heel stick blood test to screen for phenylketonuria, a metabolic condition which untreated, can lead to brain damage or death. With rapid advances in the understanding of the cause of multiple disorders, and the availability of new diagnostic testing platforms and treatments over the next decades, many other conditions were being considered for newborn screening. States were making individual decisions about which conditions to include in their screening programs without the benefit of a systematic review of the evidence as to whether an infant would benefit from being screened or treated for a particular condition. This resulted in considerable health inequities with children born in one state being screened for perhaps only one or two conditions, and just across the state line infants being screened for a much larger number of conditions, and without a good understanding of the outcome or benefits of either approach.

Congress first enacted the Newborn Screening Saves Lives Act (Pub.L.110-204) in 2008 with the realization that Federal input was essential to improving the evidence-based, universal application by states of the new technologies and treatments becoming available for a number of serious and life threating conditions affecting infants and children which are not apparent at birth.

Congress also recognized that Federal Agencies served an important role in supporting states through a variety of mechanisms including educational and training activities, research, technical assistance and infrastructure development. Over the past 11 years, Federal input from the Advisory Committee, approval of its recommendations by the Secretary of the Department of Health and Human Services (HHS), research supported by the National Institute of Health (NIH), laboratory improvement efforts at the Centers for Disease Control and Prevention (CDC), and funding to help improve state screening programs from the Health Resources and Services Administration (HRSA) have greatly benefited families by helping to advance this highly successful state-based public health system.

Although each of the conditions recommended for newborn screening are considered rare, 1 in approximately every 300 screened newborn infants is found to have a condition for which treatment is beneficial.

Today, almost every one of the approximately 4 million infants born annually in the United States undergoes newborn screening. In 2010, the Secretary of HHS officially adopted the first uniform newborn screening panel, now called the Recommended Uniform Screening Panel, or RUSP, which included 29 primary conditions and 25 secondary conditions. Within a few years, all states were screening for these conditions. With this recommended screening panel, each year approximately 12,500 newborn infants were being identified as having one of the screened conditions according to CDC research.[1] Early diagnosis enables these infants identified through newborn screening to receive the treatments necessary to prevent serious and often permanent complications, and in many cases, even death. The conditions for which we screen include genetic, endocrine and metabolic disorders, and hearing loss. For many of the conditions on the panel, early diagnosis and treatment not only benefits the infant, it is cost saving.

Since 2010, six additional conditions were recommended for inclusion on the RUSP and accepted by the HHS Secretary, increasing the primary conditions recommended for routine screening to 35.[2] Today, all states are screening for at least 31 of the primary conditions.

I would like to briefly tell you about two of the added conditions.

Severe Combined Immunodeficiency (SCID) was added to the RUSP in 2010. All states now screen for SCID. Infants with SCID are born without an effective immune system. The first infection they develop is often fatal. Infants identified with SCID because they have a serious infection often fail therapy and die of complications of the infection. Screening allows a diagnosis to be made in most cases before an infection occurs and allows for the therapy

needed to reconstitute the immune system. California[3] reported this year on the results of the first 8 years (2010 through 2017) of its newborn screening program for SCID. California identified 50 infants with SCID during this time period. Of the 49 available for treatment of the type of SCID identified, 46 of the infants (94%) survived. Another article detailing newborn screening for SCID in 11 states[4], reported an 87% survival rate in the 52 identified cases following the introduction of newborn screening for SCID.

Critical Congenital Heart Disease (CCHD) was added to the RUSP in 2011. Congenital heart disease is responsible for approximately 3% of all infant deaths in the first year of life. A number of infants born with critical congenital heart disease will have no symptoms in the newborn period and become critically ill within the first few weeks to months of life. A significant number of these infants can be detected in the newborn nursery by measuring their blood oxygen saturation before they are discharged home. A study[5] of infant cardiac deaths between 2007 and 2013 demonstrated a 33.4% decrease in early infant deaths from critical congenital heart disease in eight states after implementation of mandatory screening for CCHD.

Screening for CCHD is one of two 'point of care' tests on the RUSP that do not involve testing of a blood spot obtained from the heel of an infant. The other is hearing testing.

The other conditions added to the RUSP in recent years are:

- Pompe Disease (2013) – now being screened for in 19 state programs
- Mucopolysaccharidosis, type 1 (2016) – now being screened for in 17 state programs
- Adrenoleukodystrophy (2016) – now being screened for in 15 state programs
- Spinal Muscular Atrophy (2017) – now being screened for in 9 state programs

Much remains to be done to continue to improve the capacity and effectiveness of the newborn screening system and to meet the challenges of this rapidly changing field of health care.

H.R. 2507, as written, will strengthen the newborn screening program and will have a significant positive impact on the health and wellbeing of the nearly 4 million children born each year in the United States and its territories.

It will strengthen the efforts to bring new conditions to the newborn screening program by increasing needed funding for the efforts of HRSA, NIH and the CDC to improve state developmental readiness and training opportunities. The additional funding will allow for enhanced technical assistance and financial support for states, which will reduce barriers to implementation of new conditions and shorten the time needed for states to begin screening once a condition is approved for addition to the RUSP.

In addition, as scientific advances and the ability to utilize new technologies such as genomic sequencing are evaluated, additional research, ethical and clinical questions will need to be answered. These technological advances could significantly alter the approach to newborn

screening in the coming years. The inclusion in the reauthorization of a request for the National Academy of Science to evaluate our current screening system is timely and is likely to provide many relevant policy recommendations and/or identify areas of further study.

Once again, I thank you for the opportunity to provide testimony in support of the Newborn Screening Saves Lives Reauthorization Act of 2019. I look forward to your questions.

References

1. MMWR. 2012;61(21):390-393
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3. Pediatrics. 2019;143(2):e20182300
4. JAMA. 2014;312(7):729-738
5. JAMA. 2017;318(21):2111-2118