Leveraging Evidence-Based Public Policy and Advocacy to Advance Newborn Screening in California

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abstract

In 2016, the EveryLife Foundation for Rare Diseases, in partnership with Dr Pan, who is a pediatrician and state senator in California, launched legislation to advance and expand newborn screening. Researchers have shown that newborn screening can be cost-effective and can greatly improve health outcomes for patients with rare diseases. However, adding additional diseases in newborn screening is a long process, requiring legislative approval in addition to new state funding. Such process delays can lead to protracted diagnostic odysseys for patients, especially those with rare diseases. These delays can result in irreversible morbidity and, in some cases, early mortality for patients. To improve this process, legislation known as Senate Bill 1095 was introduced to require California to adhere to the latest federal recommendations for newborn screening within 2 years. We provide insight and describe the process of advancing state legislation, coalition building, and managing opposition. Senate Bill 1095 would become law in 2016, requiring California to screen for 2 new rare diseases by August 2018: mucopolysaccharidosis type I and Pompe disease. This case study can serve as a model for advocates looking to expand state newborn-screening programs.

THE NEWBORN-SCREENING LANDSCAPE

Newborn screening has been a key pillar in improving and advancing public health in pediatric patients in the United States for decades.[1] The first newborn-screening programs began at the state level with a technique for detecting phenylketonuria in the late 1950s and would eventually evolve and expand into a broader system with key components at both the state and federal levels.[2] Over time, additional diseases, such as a variety of hemoglobinopathies, would be added across states, dramatically improving diagnosis for these diseases and significantly lowering mortality rates overall.[3,4]

Today, every state in the United States has a newborn-screening program. State-controlled departments of public health manage both the implementation of newborn-screening programs and the laboratories used for processing the blood spots. Newborn screening has also been expanded beyond use of blood spots to include hearing tests as well as pulse oximetry tests, which can be used to detect critical congenital heart disease.[5] Not all states maintain and operate their own newborn-screening laboratories, but many do. The laboratories process blood spots and provide results to health care providers and families. Meanwhile, at the federal level, various agencies provide support to advance state newborn-screening programs. Most visibly, under the auspices of the US Department of Health and Human Services (HHS), the Secretary of HHS convenes the Advisory Committee on Heritable Disorders in

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Mr Bronstein helped lead the advocacy work on behalf of Senate Bill 1095, drafted the manuscript, and recruited co-authors; Dr Pan led the legislative initiative for advancing Senate Bill 1095, carrying the bill, and testifying in the state legislature and provided additional content and edits to the manuscript as a co-author; Mr Dant provided testimony to the California state legislature in support of advancing newborn screening and Senate Bill 1095 and provided additional content and edits to the manuscript as a co-author; Dr Lubin was a former member of the Board of Directors of the EveryLife Foundation for Rare Diseases, provided strategic direction on advancing the legislation, has been an advocate for advancing children's health in California, played a key role in enabling newborn screening for hemoglobinopathies, and provided additional content and edits to the manuscript as a co-author; and all authors approved the final manuscript as submitted and agree to be accountable for all aspects of the work.

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Newborns and Children to provide advice and recommendations for states on implementing novel tests for newborn screening. Once a screening recommendation is made by the committee, the Secretary of HHS has 120 days to make a decision regarding endorsement, at which point it becomes part of the official list of diseases recommended for screening.[6] Members of the advisory committee include leading public health experts with deep knowledge of newborn-screening programs and the benefits they can provide for patients. Updates to the Recommended Uniform Screening Panel (RUSP) contain a list of diseases that are recommended for screening. At the time of this writing, the committee currently recommends screening for 34 core conditions,[7] with the latest disease, spinal muscular atrophy, being recommended for screening on February 8, 2018.[8] The recommended diseases vary widely in terms of prevalence (spanning rare and ultrarare diseases) and severity. For a disease to be recommended for addition to the RUSP, the committee follows specific criteria, which include the following: availability of a Food and Drug Administration–approved treatment of the disease in question, a reliable and scalable assay for detecting the disease, and evidence that screening provides a net benefit to patients.[9] These criteria must be fully explored when submitting a nomination package to the RUSP, which requires data on the outcomes of early treatment versus delayed treatment in patients who have not benefited from newborn screening.[10] Of the ∼4 million infants born each year in the United States, 12 500 are diagnosed via newborn screening with 1 of the core conditions listed on the RUSP.[11]

States, however, have different responses to the federal recommendations and a range of diverse considerations. As a result, there is wide variation among the states as to which diseases are screened for and the policy and legislative mechanisms by which new screens are adopted. In some states, new legislation is required to permit the screening of a novel disease, and separate legislation may also be required to outlay funds to cover the additional costs of screening. In other states, screening decisions to add novel diseases are made at the discretion of agencies of public health but must be supported by additional state funding (requiring legislative action) or increases in newborn-screening fees, which are covered by public and private payers. These mechanistic and legislative discrepancies among states as well as various layers of legislative and regulatory bureaucracy have led to a patchwork of newborn-screening approaches across the United States.

The authors of a recent publication in *Pediatrics* illustrated the degree to which state screening panels lag behind the RUSP recommendations. State adoption of novel newborn-screening tests is at a median delay 4.4 years for severe combined immunodeficiency (SCID) and 3.2 years for critical congenital heart disease, with additional time delays being required for implementation.[12] This has created a patchwork of newborn-screening approaches across the United States rather than a science-based standard for newborn screening (Fig 1). Unfortunately, this dynamic results in potentially devastating health outcomes for both patients and families. Many infants will be born with conditions that are both detectable and treatable, but if they are born in a state that has not yet begun to screen, those infants are at great risk of severe, irreversible morbidity and, in some cases, early mortality.

In Fig 1, we highlight the variation that exists among states in screening for X-linked adrenoleukodystrophy, mucopolysaccharidosis type I (MPS I), and Pompe disease, all of which are listed on the federal RUSP. Although each of these diseases is rare, early detection and early treatment made possible through newborn screening can be lifesaving for patients with the most severe forms of these diseases and can help delay and, in some cases, prevent disease progression.[13]

Cost-benefit considerations are key decision-making inputs as states evaluate current newborn-screening programs and potential expansion. There is a significant and growing body of research exploring this area, which has been a critical tool for advocates in advancing newborn screening. In 1 such study published in *Pediatrics*, Carroll and Downs[14] explored the cost-effectiveness of newborn screening for inborn errors of metabolism and found that for the majority of diseases screened, such interventions would be both beneficial to patients and likely to save money for society over the long-term. In another analysis published in *Value in Health*, Tiwana et al[15] examined whether significant expansion of newborn screening in Texas (from 7 to 27 disorders) was cost-effective. This expansion of newborn screening was implemented in 2007, and the authors concluded that although costs increased to payers, the increases in quality of life for patients as well as the avoided morbidity and mortality proved to be cost-effective for Texas. Cost-effectiveness research will continue to play a key role as states evaluate expanded newborn programs.

ADVANCING AND IMPROVING NEWBORN SCREENING IN CALIFORNIA

A few states, however, have adopted an approach to help eliminate legislative delays to adhere to the most up-to-date standards recommended by the federal RUSP. What follows is a policy and legislative case study in which we highlight a unique and successful partnership between newborn-screening stakeholders in California.

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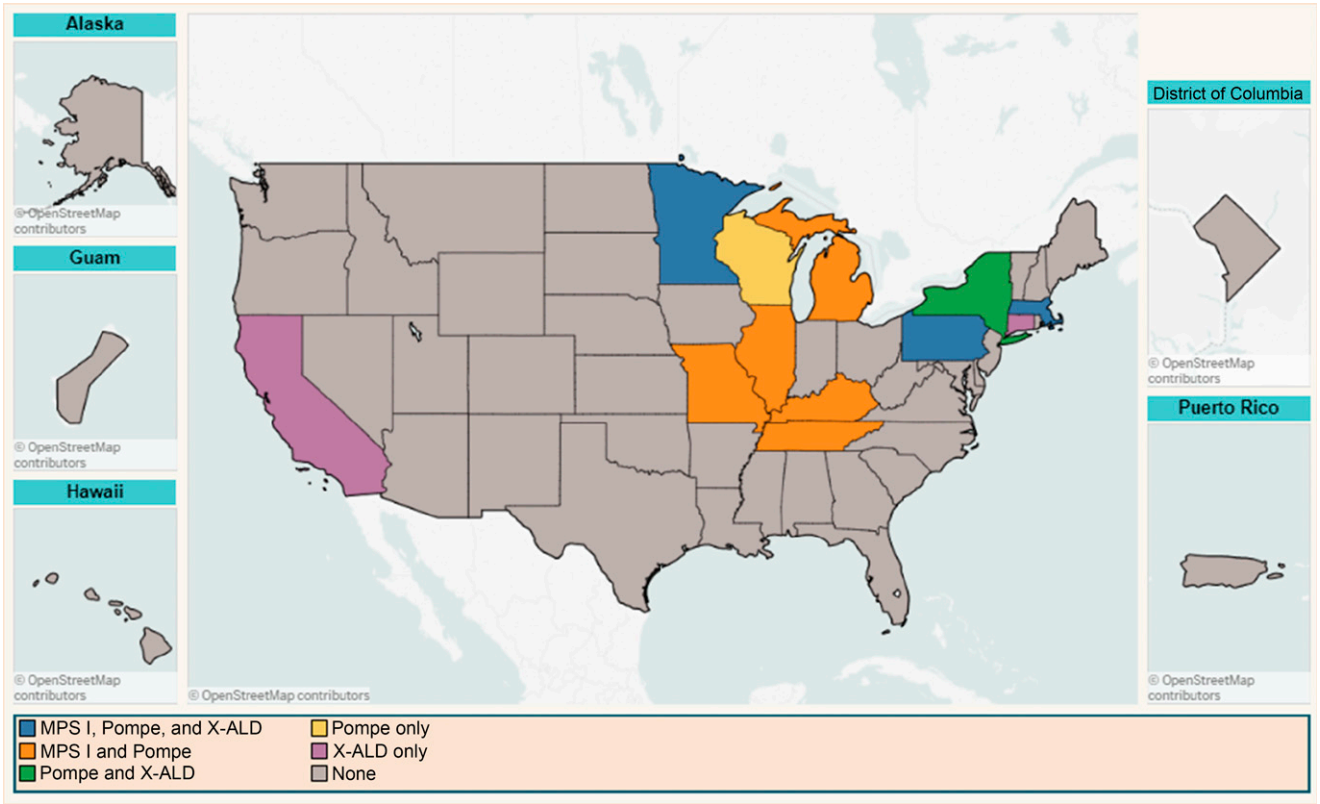

FIGURE 1

State and territory implementation of newborn screening for MPS I, X-linked adrenoleukodystrophy (X-ALD), and Pompe disease (2018) (Adapted from Association of Public Health Laboratories NewSTEPs Program. Newborn screening status for all disorders. 2018. Available at: https://www.newsteps.org/resources/newborn-screening-status-all-disorders. Accessed April 13, 2018).

Our goal is for this case study to serve as an example for how patients, patient organizations, state lawmakers, physicians, and academics can work together to dramatically improve newborn screening and health outcomes in the United States.

Late in 2015, the EveryLife Foundation for Rare Diseases (a nonprofit organization based in Novato, CA) identified Dr Pan, a California state senator and pediatrician, as a potential partner for advancing and improving newborn screening in the state.

Dr Pan had a long track record as an advocate for advancing children's health and would become a key partner in crafting and advancing the legislation that would become known as Senate Bill 1095 (SB 1095).[16] Dr Pan previously authored laws to add SCID and adrenoleukodystrophy to the California newborn screening panel, and his legislative experience guided strategy to pass SB 1095. In addition, his success in authoring nationally recognized child-health legislation, including abolishing nonmedical exemptions for vaccination, gave him tremendous credibility as a pediatrician and legislator. An initial meeting was convened with Dr Pan and key EveryLife staff to discuss the possibility of partnering on newborn-screening legislation. Both parties were keenly interested in designing a bill that would help ensure that California added diseases for newborn screening without the need to run individual bills for each disease, which historically has proven to be a risky and time-intensive approach with uncertain outcomes. At this stage, EveryLife (in partnership with Dr Pan) opted for an approach that would help ensure California's leadership in newborn screening in perpetuity.

Ultimately, the bill was designed to institute a process for the addition of new screens to the state screening panel. Specifically, SB 1095 required California to screen for any disease(s) recommended by the federal RUSP with an implementation deadline from the time of the federal recommendation. The bill also contained vital language requiring the state to outlay funding or appropriations for implementation. This ensures that monies are available to purchase new equipment, train or hire new staff, and for other related expenses required for the successful implementation of new screens.

This approach offered a variety of benefits:

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1. Eliminate the legislative delay that had hampered the addition of new screens in part because state lawmakers would no longer need to introduce and steward individual bills for the addition of a single new disease.
2. Ensure the availability of funding to add new screens. Many states fail to expand newborn screening on the basis of a lack of available funding.[17]
3. Transition the federal recommendations into requirements. This helps ensure that the state would be a leader in advancing newborn screening and children's health in perpetuity.

NEWBORN-SCREENING ADVOCACY IN ACTION

Before the introduction of SB 1095, the EveryLife Foundation for Rare Diseases had assembled a coalition of stakeholders, including >100 patient-advocacy organizations, who would be key partners for promoting, supporting, and shepherding the bill through the legislative process. Patient advocates and patient-advocacy organizations were identified that would benefit directly from the addition of new screens (eg, MPS I and the National MPS Society) and would play a leadership role in the advocacy effort. It's also important to point out that dozens of other rare-disease organizations with no direct connection to the diseases in question or to newborn screening stepped up to endorse the bill. This broad endorsement was made possible by ongoing grassroots outreach conducted by EveryLife and demonstrated strong support from patient organizations that broadly recognized the public health imperative of expanding and improving newborn screening. This coalition would play a key role throughout the legislative process, providing support and grassroots outreach at key junctures. EveryLife would periodically convene webinars and/or conference calls to help keep the coalition informed about and engaged in the effort.

SB 1095 was officially introduced in the California state legislature on February 16, 2016, by Dr Pan.[16] To become law, the bill would have to be voted on on 6 separate occasions and would require majority votes in the committee and on both floors of the California senate and assembly (Table 1). Assuming that these votes were successful, the bill would then advance to the governor for signature. There is, however, a strict timeline for the completion of this process. In California, for example, the voting would have to occur before the August 2016 deadline for transmitting bills to the governor.

From February 2016 through August 2016, the foundation, Dr Pan, and the coalition employed a variety of tactics to help build support and momentum. Some examples include the following:

1. electronic action alerts coordinated by the EveryLife Foundation for Rare Diseases, whereby constituents would send dozens of letters of support to their representatives in the California legislature;
2. a group sign-on letter from >100 patient organizations addressed to state policy makers calling for support and swift passage of SB 1095;
3. in-person meetings convened with key members of the legislature to raise awareness about the legislation and the potential benefits it could provide to infants born in California; and
4. as committee hearings on SB 1095 were convened, patient advocates provided oral testimonies in which they highlighted the importance of newborn screening for rare diseases.

What follows is an excerpt of the oral testimony provided by Mark Dant, father and caregiver of Ryan Dant, a young man affected by MPS I:

TABLE 1 Legislative Pathway for SB 1095

Date	Legislative Action	Outcome
February 17, 2016	Senator Pan introduces SB 1095 in senate	Referred to Senate Health Committee for analysis
April 14, 2016	Senate Health Committee hearing; rare disease advocates testify in support of the legislation	Passes Senate Health Committee 9–0 and referred to Appropriations Committee (fiscal committee)
May 27, 2016	Senate Appropriations Committee hearing	Passes Senate Appropriations Committee 7-0
June 2, 2016	Full senate vote	Passes senate on consent 39-0; ordered to assembly
June 22, 2016	Assembly Health Committee convenes public hearing; rare disease advocates testify in support of the legislation	Passes Assembly Health Committee 17-0; referred to Appropriations Committee
August 12, 2016	Author amends bill to address Department of Public Health concerns; assembly Appropriations Committee hearing	Passes assembly Appropriations Committee as amended 20-0
August 18, 2016	Full assembly vote	Passes assembly 78-0
August 24, 2016	Senate concurs with bill as adopted by assembly; bill is engrossed and enrolled	Senate concurs with assembly amendments 39-0; enrolled and transmitted to governor
September 16, 2016	Rare disease advocates and Dr Pan meet with governor's staff; governor reviews bill	Signed by the governor; SB 1095 becomes public law

Adapted from California State Legislature. Complete bill history. Available at: www.leginfo.ca.gov/pub/15-16/bill/sen/sb_1051-1100/sb_1095_bill_20160916_history.html. Accessed August 2, 2018.

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Our son's diagnostic journey pales in comparison to the often lengthy quest parents undergo to find the right diagnosis. As the executive director of the National MPS Society, I am in constant contact with parents who spend years moving from doctor to doctor, specialist to specialist, often times being misdiagnosed and watching their child's actual disease worsen as they are treated for diseases they do not have.

[The diagnostic odyssey of] Krystal Gonzales from Chula Vista, California, . . . is sadly not an isolated story. Krystal's mother, Linda, took her to a pediatrician when she was 8 years old because she had not yet received an answer to why Krystal was developing so differently than any of her schoolmates. Her pediatrician suggested she take her to a rheumatologist because of Krystal's stiff joints and short stature. The rheumatologist treated Krystal's hands with multiple rounds of steroid injections in each finger, which did nothing to treat her disease. After about 2 years with no improvement, the rheumatologist suggested Linda take her to a geneticist, who after examination diagnosed Krystal with Noonan syndrome and started her on the protocol to treat Noonan [syndrome], which included a low-dosage [chemotherapy] pill as well as Enbrel shots once or twice a week. Linda remembers that the methotrexate caused Krystal to lose a great deal of her hair as well as nearly constant nausea. In addition to these treatments, Krystal had contractures on both knees and walked on her toes, so she was casted twice for 6 weeks then recasted for 8 weeks. This treatment occurred twice, both times prior to the proper diagnosis of MPS I, and of course, both times, the casting did nothing to help correct the problem.

Krystal was nearly 12 years old before a physician correctly diagnosed her with MPS I. Within weeks, Krystal began receiving the appropriate drug to treat her disease, and within weeks of proper diagnosis, Krystal's life journey changed. Linda reports that Krystal feels better and is stronger each day but has more surgeries ahead of her to try to correct the 12 years of no and wrong diagnosis . . . 12 years of not being on the treatment that would have most probably changed Krystal's quality of life forever.

We cannot go back and change what Krystal and Ryan missed all those years without treatment, but what we can do is keep other parents from watching their children suffer day after day as they seek the right diagnosis. We have a treatment that is approved to help these children the very day they are diagnosed. Please help them find the proper diagnosis at birth through newborn screening.

It is important to emphasize how critical the testimony provided by Mark Dant and by other parents was in building support among California lawmakers. The stories resonate powerfully with elected officials, many of whom are parents themselves. The personal and emotional nature of testimony illustrates the impact of the disease burden as well as the potential for dramatically improving health outcomes with expanded newborn screening. Advocates particularly emphasized that for many metabolic diseases detected by using newborn screening, significant and irreversible patient injury already occurred by the time symptoms manifested, making newborn detection critical to initiating treatment in time to prevent harm. In addition, fact sheets were developed for policy makers to highlight potential cost savings from adding additional screens along with case studies of contrasting patients who had the benefit of newborn screening versus those who did not. The combination of stories from individual patients along with data revealing the potential cost-effectiveness for the state was a key tool in persuading state lawmakers.

UNDERSTANDING THE POLITICAL DYNAMIC

Although the legislation enjoyed broad support from state lawmakers, there were still concerns raised regarding the bill. The bill would have to pass appropriations committees in each house, where the state cost of the legislation would be determined. As new screens are added, states incur additional costs of purchasing testing kits, new equipment, and hiring additional staff. In anticipation, the coalition prepared cost-benefit research before the introduction of SB 1095 that would be provided to committee staff when they prepared bill analyses. A previous publication in *Pediatrics* revealed that at the state level, the introduction of tandem mass spectrometry in California's newborn-screening program was estimated to save the state $0.27 in health care costs for every dollar spent on screening.[18] A similar analysis of Washington state's newborn-screening program revealed that every dollar spent on SCID screening saved an average of $0.43 in treatment costs.[12] Decision makers often request information on the cost savings, cost-effectiveness, or cost/benefit ratio of public health programs. In practice, quantifying the health and economic benefits of population-level screening programs, such as newborn screening, is challenging. However, there is ample evidence that the cost of completing the federally recommended screens can save health care dollars in addition to preventing undue suffering.[19]

Concern was also raised that requiring public health laboratories to add a slew of additional screens as innovative drugs gained Food and Drug Administration approval would place undue burden on the state. However, research indicated that the federal RUSP makes relatively few recommendations on an annual basis[20] and therefore would not outpace the state's ability to implement new screens. Some were also concerned with the implementation timeline for the addition of new screens as additional recommendations are made by the federal RUSP. In particular, state officials wanted to ensure that the public health laboratories would have sufficient lead time to purchase new equipment as well as hire and/or train new staff. Ultimately, through constructive negotiations and discussions with the Department of Public Health, a compromise was reached that would allow for 2 years from the time of federal RUSP recommendations to implementation in the state.

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Finally, the governor needed to be persuaded to sign the bill. Although the bill received unanimous support in the legislature, Governor Edmund (Jerry) Brown, Jr frequently expressed opposition to legislation that resulted in ongoing state spending, and a key part of Governor Brown's legacy was restoring budget stability after the Great Recession and fiscal preparation for the next economic downturn. However, advocacy efforts with the administration, including the Department of Public Health, which administered the newborn-screening program, advanced parallel to the advocacy efforts with the legislature. A variety of factors would ultimately influence the governor's decision-making on SB 1095, but among the most important factors were the following: (1) unanimous support in the legislature, (2) analyses revealing that additional newborn screening would be cost-effective for the state, and (3) in-person meetings between key advocates and the governor's health staff. The governor ultimately signed SB 1095 into law on September 16, 2016.

IMPLEMENTATION OF NEW SCREENING LAWS AND NEW SCREENS

With the adoption of SB 1095, California would then have 2 years (ie, a deadline of September 16, 2018) to comply with the latest RUSP recommendations. For California, this meant the implementation of 2 new screens: MPS I and Pompe disease.

At the time of this writing, the California Department of Public Health has purchased new equipment and trained staff to help manage the additional screenings for MPS I and Pompe disease. In the first year of expanded screening, we expect that ~4 patients with MPS I will be detected along with as many as 50 patients with Pompe disease (some with infantile onset and others with late onset).[21] Early diagnosis and access to enzyme-replacement therapies for both MPS I and Pompe disease can dramatically improve health outcomes and, in some instances, can be lifesaving.[22–25]

CONCLUSIONS AND LESSONS LEARNED

Since the passage of SB 1095, other states, such as Florida,[26] have passed similar bills to help advance and improve newborn screening. However, 41 states are still not screening for Pompe disease, 42 are not screening for MPS I, and none are screening for spinal muscular atrophy. Additional advocacy is needed as states weigh novel newborn-screening legislation, and it will be especially vital for policy makers to hear from patients and health care providers alike to understand the burden of these diseases and the impact that newborn screening can make to improve health outcomes. We believe the process of identifying legislative champions, building coalitions, and executing advocacy campaigns described provides guidance to a variety of stakeholders who may wish to expand and improve newborn screening in their states as well as have an impact on children's health more broadly. In particular, coalitions that comprise a broad group of stakeholders but include representation from health care providers, such as treating physicians, patients and/or patient organizations, academics, and others, can be highly effective in advocating within state legislatures. However, it is important to note that the outcomes of the legislative process are often uncertain and, at times, based on factors outside of evidence available to policy makers.

Cost-effectiveness will continue to be an issue in state legislatures and within state departments of public health, so advocates must be prepared to offer data revealing how early diagnosis through newborn screening can be cost-effective for the state. However, it is important to note that for some diseases, it may be difficult to show cost-effectiveness because of the cost of treatments or other factors. Although cost is a critical determinant in newborn-screening decision-making, it is just 1 of several inputs that policy makers consider when weighing whether to add additional screens. Furthermore, the federal RUSP is now playing a more central and empowered role as states look for additional guidance and advice on expanding screening programs. This has the potential to introduce a true standard for states to follow, but the RUSP may lag behind the latest science. In such cases, states should be willing to expand screening beyond current recommendations or to engage in pilot programs to research the viability of screening for new diseases. The data generated in such pilot studies can play a key role as input during the RUSP application process and as other states seek evidence in support of expanding newborn-screening programs.

States are poised to play a leading and growing role in improving newborn screening across the United States but will need to hear from advocates, researchers, and health care providers as changes are considered and implemented by state departments of public health. Such changes will help to ensure the legacy of newborn screening in the United States as a leading public health intervention with the ultimate goal of ensuring the best possible health outcomes for pediatric patients.

ABBREVIATIONS

HHS: Health and Human Services
MPS I: mucopolysaccharidosis type I
RUSP: Recommended Uniform Screening Panel
SB 1095: Senate Bill 1095
SCID: severe combined immunodeficiency

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FINANCIAL DISCLOSURE: Mr Bronstein is employed by and owns stock options in Audentes Therapeutics, which is conducting preclinical research for a Pompe gene therapy treatment; and Dr Pan, Mr Dant, and Dr Lubin have indicated they have no financial relationships relevant to this article to disclose.

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