

End of an Era, or Just Another Inflection Point in the History of Newborn Screening in the United States?

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abstract We have reached another inflection point—perhaps the end of an era—in state newborn screening programs. Frequently touted as one of the great achievements in public health, newborn screening in the United States is remarkable in part because of the role of state government in routine pediatric care and because of the role of the federal government in the standardization of health policy through an evidence-based, publicly engaged process. These and other long-standing characteristics of newborn screening are being challenged by the extraordinary realignments in federal government and by the long-awaited advances in screening technologies, gene therapy, and other novel therapies. This is not the first time state newborn screening programs seemed precarious; in fact, there have been challenges to state newborn screening nearly every decade since it began in the 1960s. However, recent disruptions to key components of newborn screening in the United States have the potential to radically reshape the future of public health approaches to newborns with rare diseases.

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INTRODUCTION

State newborn screening (NBS) programs are an anomaly in the US health care system because of the combination of state involvement and evidence-based policy formation. Every day in the United States, nearly every baby born receives sophisticated laboratory screening tests done despite having no symptoms of disease. This medical practice is driven by state public health law, achieving near-universal access but without true informed consent. Unlike much US health policy, the approach to adding medical conditions to state screening panels has typically been informed by a national process that is evidence based and deeply influenced by clinicians, researchers, industry, family advocates, and public health professionals. NBS is hailed as a great public health achievement because of its impact on infants with rare and ultrarare diseases. It is also unusual because it demonstrates how the United States can go beyond market forces and instead organize public and private resources to meet the health needs of its citizens.

We have reached another inflection point in the history of NBS in the United States. Disruptive advances in scientific medicine such as whole genome sequencing (WGS), gene therapy, and patient-targeted therapies have created the possibility that hundreds if not thousands of rare conditions could be added to state NBS panels.^{1–5} An unexpected but perhaps more fundamental challenge concerns the role of the government—and federal government in particular—as it undergoes a generational change. Federal government funding of scientific research is being transformed, federal expert opinion committees have been reshaped or terminated, and the federal government's support for uniform state NBS is unclear in the midst of many other changes to education and health care policy.

The questions we face are fundamental. Will a centralized, evidence-based approach to adding conditions to state NBS panels prevail? Will the adoption of WGS result in a fundamental shift in the public-health mandate for NBS, whereby it becomes a provider of information that is valued by some families but not medically urgent? Will the decades-long consensus among state NBS programs that no baby be left behind survive the current disruptions in scientific medicine and in government policy?

THE ROLE OF STATE GOVERNMENT IN NBS PROGRAMS

Most health care in the United States is delivered through a market-driven approach, with variable access based on insurance and other factors. State NBS programs are unique in part because state governments ensure nearly universal access. Every day, nearly 10 000 infants are born in the United States. In the day or so after birth, a nurse or technician gathers a few drops of blood from their heels with special filter paper. The specimen is transported to a centralized laboratory, and within a few days of that infant's

birth, laboratory technicians run dozens of complicated tests to search for rare and ultrarare conditions such as phenylketonuria (PKU), sickle cell disease, or methylmalonic acidemia. If the result suggests the possibility of one of these conditions, state public health personnel alert the family or clinicians as soon as possible. The latter are responsible for performing diagnostic tests and initiating treatment, sometimes within days when early intervention is critical. Treatment could be relatively simple, such as starting thyroxine for hypothyroidism, or complex with substantial risks, such as hematopoietic stem cell transplantation for Krabbe disease or severe combined immunodeficiency.

States laws govern this process, and state NBS programs take different approaches to ensure that nearly every baby, regardless of income or health insurance, receives the test. Some states have their own laboratories; others contract with nearby states or private laboratories. Most charge hospitals or other providers for the tests, and health insurance frequently covers costs for screening based on federal law. All states strive to meet national benchmarks for timeliness and quality of screening tests and for appropriate notification of families and clinicians. States take responsibility for testing specimens 5 to 7 days of the week, and all have contingency plans for any outages including disasters such as hurricanes and floods. Many states with laboratories struggle to maintain sufficient personnel, given low salaries for highly specialized skills. All states confront variations in budgets and contracting procedures that challenge their ability to test every newborn and notify parents of results within the first few days of life.

Federal agencies have provided essential support to state newborn screening programs since the 1960s, and in the past 2 decades through specific federal NBS legislation. The National Institutes of Health (NIH) has funded basic and translational research to develop screening techniques and novel treatments. The Centers for Disease Control and Prevention has provided state laboratories with technical assistance, proficiency testing materials, and standards-setting essential to specialized testing platforms that require continuous quality control and improvement. The Food and Drug Administration helps guide the commercial development of screening and diagnostic tests as well as medications used to treat children with newborn conditions. The Maternal and Child Health Bureau (MCHB), part of the Health Resources and Services Administration, has offered grants to state NBS programs and to professional organizations such as Association of Public Health Laboratories (APHL), the American Academy of Pediatrics (AAP), and the American College of Medical Genetics and Genomics (ACMG) to educate professionals, implement screening for new conditions, address quality and timeliness of short-term follow-up, and improve access to clinical care.

None of this is easy. For example, state NBS programs have historically focused on identifying any child who could benefit from early detection. When screening test results are set to maximize sensitivity, however, false-positive screening results are common, especially because NBS conditions are so rare. States strive to balance not missing an affected child with minimizing false positives, which can alarm families and use valuable health care resources. States must also have a process to rapidly find a family after they have left the hospital when there is a positive result for a time-critical disorder; this requires relying on information from diverse partners. Finally, states have some obligation to ensure that clinical specialists are available to confirm the diagnosis and initiate treatment.

Unlike many other health care practices in the United States, there is evidence that state NBS programs effectively reach nearly the entire target population.⁶ States report data to a centralized national organization, APHL, which MCHB funds to maintain a national dashboard to track system function (NewSTEPS⁷). Continuous quality improvement efforts have led to 99% of all infants in the United States being screened, with most states reporting timely notification of families and clinicians for at least some conditions. Most states do not, however, take responsibility for treatment. After their infant is diagnosed with an NBS condition, families struggle with the same vagaries of health insurance coverage, availability of clinical specialists, and ready access to treatment that characterize the US health care system. Limited evidence on long-term follow-up suggests that many infants identified with NBS do not receive care according to clinical guidelines.^{8–10}

Despite shortcomings in long-term follow-up, NBS as a state responsibility is indeed a public health triumph. It did not have to be this way. Infant NBS could be just another clinical service, depending on the hospital where an infant is born. Indeed, when NBS for PKU began in the late 1950s, hospitals simply added the heel-stick test to the list of routine newborn clinical tasks, like checking blood glucose levels or monitoring weight. NBS became a state responsibility in the early 1960s, when family advocates convinced legislators to require universal testing of all newborns. Led by Robert Guthrie, the microbiologist who invented the filter paper method, families told emotional stories about their children being saved—or not—by NBS. They argued that newborn screening could help many children in the future.

In the mid-20th century, the idea of government being involved in health care, such as state-based NBS, was opposed by most doctors, including the AAP (at least initially) and the American Medical Association, as an unwanted incursion into the practice of medicine. This was a time before Medicare, Medicaid, and many of the laws that currently govern health care. State legislators were attentive to advocates' arguments, however, in part because state hospitals for the intellectually disabled were a large

budget item and because NBS held promise to reduce the number of future wards of the state. Hospitals were supportive because some faced lawsuits for missing cases of PKU. At a national level, President Kennedy and his formidable sister, Eunice Kennedy Schriver, were leading a national crusade to improve the lives of people with intellectual disability by investing in research, education, and clinical services. More broadly, there was remarkable faith in science and technology in the mid-20th century: polio and measles were rapidly disappearing because of vaccines, and nuclear power and space travel were changing how Americans viewed their future. These advances in science and technology were supported by government programs, and state NBS laws seemed yet another way to improve the health and well-being of all citizens.¹¹

THE RECOMMENDED UNIFORM SCREENING PANEL: EVIDENCE-BASED, TRANSPARENT HEALTH POLICY

From the late 1960s through the 1990s, state NBS programs expanded slowly to new conditions such as hypothyroidism and galactosemia via a “one condition, one new test” model. Such expansion came with challenges. Some states added new diseases, such as histidinemia, to their panels, only to stop a few years later because treatment was not necessary.¹² Screening for other conditions, such as sickle cell disease, led to concerns about racial bias. As the optimism of the 1960s faded and trust in government waned, some states wondered if NBS was worth the cost at all, especially because only a few babies were identified with NBS conditions each year. In 1975 an influential National Academy of Sciences report on newborn screening criticized how PKU testing had been instituted in states but ultimately affirmed the value of NBS programs.¹³ The report called for patient advisory committees and provided guidelines for adding a condition to state NBS programs: public benefit and acceptance, feasibility studies, satisfactory test methods, quality control, resources for follow-up and treatment, acceptable costs, effective education, and informed consent.

Some of these recommendations were implemented, and state NBS programs continued to grow slowly in the 1980s. They had the opportunity to substantially expand their screening panels in the late 1990s with the application of tandem mass spectrometry (MS/MS). Instead of separate tests for each condition, MS/MS gave laboratories the potential to detect dozens of conditions in a single platform, including amino acid disorders, fatty acid oxidation disorders, and organic acid disorders. As a few states expanded to dozens of disorders and others did not, the variability among state NBS panels became more obvious. NBS advocates and the news media highlighted disparities based on geography. Noting that their children would be alive if they had been born in another state, families pleaded with legislators to expand their states' panels to include more conditions.

To support states during this time of rapid expansion, MCHB worked with organizations such as the AAP and ACMG to establish guidelines for NBS programs.¹⁴ Based in part on recommendations of a 1999 AAP task force, MCHB contracted with ACMG to gather a group of clinical and laboratory experts who in 2004 identified 29 conditions that states should screen for based on feasibility of screening and the availability of treatment.¹⁵ MCHB also oversaw the creation of a federal advisory committee, the Advisory Committee on Heritable Disorders Newborns and Children (ACHDNC), to advise the Secretary of the Department of Health and Human Services (HHS) on universal newborn screening tests, technologies, policies, guidelines, and standards. Federal support of state NBS programs was explicitly delineated with the passage of the Newborn Screening Saves Lives Act in 2008. That same year, the ACHDNC formally recommended that the HHS Secretary use a uniform Recommended Uniform Screening Panel (RUSP) to guide states, based on the 29 conditions listed by the ACMG workgroup.

The RUSP has become a focus of state NBS programs, with more than a dozen states having laws requiring that their programs add or at least consider adding conditions on the RUSP to their state panels. Groups of clinicians, researchers, industry partners, and family advocates devote decades to pursuing the addition of even a single condition to the RUSP. It is not easy; the criteria are stringent. To warrant addition to the RUSP, there must be a clear clinical case definition, a well-functioning screening test, demonstration of feasibility of detecting at least 1 case at a public health scale, and published evidence that the benefits outweigh the risks of treatment initiated *before* a condition is identifiable clinically. Gathering the evidence for a condition to be considered for the RUSP is difficult because basic research and population-level pilot testing are time-consuming and expensive, there are limited research funds and thousands of potential conditions, and demonstrating the success of a treatment before clinical detection is nearly impossible for rare conditions if there is no state screening program running a universal pilot project.

Until the termination of the ACHDNC in 2025, the process for RUSP inclusion most often began with advocates gathering evidence and nominating a condition for the ACHDNC to review. If the ACHDNC voted to fully evaluate a RUSP candidate, MCHB contracted with an independent evidence review group to systematically gather and present all available evidence to the ACHDNC. In a series of public meetings that included input from stakeholders, the ACHDNC discussed and eventually voted whether to recommend to the HHS Secretary to include the new condition on the RUSP. When the Secretary adds a condition, each state has a process for deciding whether to include it on their state screening panel. Through the Affordable Care Act,¹⁶ the RUSP process also leads to most health insurance plans

being required to directly or indirectly cover the costs of state NBS without cost sharing by the family.

There have been challenges to this process over the last decade: advocates for specific conditions have argued that the criteria are too stringent, the process is too slow, and support for research is far too little. On the other hand, many state laboratories counter that the process is too fast for them to prepare and implement new conditions appropriately. More recently, the rapid advancement of next-generation genomic sequencing has created the potential for hundreds of rare conditions to be identified in a single platform. Meanwhile, the recent success of gene therapy and other interventions suddenly makes treatment at least theoretically possible for many metabolic conditions.^{1,17}

The potential disruption of using genomic sequencing as a first-line screening test is similar in some ways to MS/MS in the 1990s. The costs and technical challenges for state laboratories to adopt a complex new platform are daunting; to support states in planning for genomic testing, the NIH is funding research into the feasibility of state NBS systems to identify conditions through genomic sequencing. In addition to incorporation of new laboratory procedures, states will also have to determine which specific conditions, genes, and variants revealed by NBS have sufficient penetrance and expressivity to justify alerting families and clinicians that confirmatory testing is necessary. The issue of false positives has long been a problem for state laboratories and follow-up programs; incorporating genomic sequencing as the primary approach to identifying a condition could make this exponentially greater because of the number of conditions and amount of information generated by genomic sequencing. Evaluating the many infants potentially needing urgent diagnosis can quickly overwhelm the already limited number of pediatric subspecialists who can provide appropriate diagnosis and treatment.

Adding hundreds of conditions would also be a challenge to the federal evidence review process of assessing conditions for the RUSP by the ACHDNC, which historically required nearly a year for just 1 condition. One approach is to follow the example of research programs currently using genomic sequencing in the newborn period. In these studies, clinical and laboratory experts organize conditions into a few categories based on specific criteria for feasibility, seriousness, child onset, and potential for clinical intervention.^{18–20} Using such an approach, dozens of conditions could be added to the RUSP as a group, rather than one at a time. The role of an advisory committee would be to provide criteria to guide the process of placing each condition in a specific group.

These are all logistical challenges, however, and genomic sequencing could—like MS/MS—be incorporated into state NBS without changing the fundamental premise that a state public health program should focus on identifying conditions that require presymptomatic treatment. For example,

genomic techniques could be restricted to provide results only in instances of very high genotype-phenotype correlation. Such an approach would not reveal all or perhaps even many of the children who would eventually have the condition. This would be a fundamental shift for state laboratories, however, given their historical focus on making sure to identify any baby with a screened condition.

Genomic technologies can also provide significant information on future disease risk that at least some families would find valuable. Offering useful but not urgent information would further expand state NBS programs beyond their traditional mission. Such an approach would not be unprecedented: some states report sickle cell trait status and risk for late-onset conditions that have health implications. The sheer amount of information available in genomic sequencing would tip the balance, however, and possibly alter the role of NBS. It raises the issue that has recurred since the 1960s: how great does the potential benefit of screening need to be to justify state involvement through a public health screening program with minimal informed consent?²¹

One recent example of this debate occurred in 2023 and 2024 as the ACHDNC discussed whether to add Krabbe disease to the RUSP.²² Some committee members judged the potential net benefit of universal NBS to be insufficient because the published research suggested that treatment carried substantial risks and the benefits were unclear beyond extending the life of the child. Furthermore, NBS would reveal some later-onset types of Krabbe disease, and those infants would require burdensome follow-up with little evidence of benefit. In contrast, parents of children with Krabbe disease argued that they were in the best position to make judgments about the value of treatment, and state NBS detection was essential for them to be able to have the *choice* to intervene earlier enough for the treatment to work at all. Ultimately, the ACHDNC voted to recommend inclusion to the RUSP with the stipulation that screening be restricted to biochemical profiles that minimized providing some parents with uncertain results about later-onset versions of Krabbe disease.

Informed consent, whether for screening or treatment, is a key feature of this debate. Families of children with Krabbe disease were rightly pointing out that when reasonable people can disagree on the risks and benefits of a treatment for an infant, families are in the best position to decide whether to proceed. However, a long-standing basis for state NBS screening programs has been that they only screen for conditions that *require* treatment. Indeed, when state NBS began in the 1960s, no one imagined that a family would say no to treating PKU, in part because individual informed consent was not yet central to medical practice. Benign paternalism, in which the doctor knew best what patients needed, governed most clinical decisions. As patient autonomy and formal informed consent became

central to medical practice starting in the 1970s, the lack of explicit consent for NBS tests was justified by the idea that government has the duty to protect its citizens, in this case newborns with treatable conditions. If families can ethically refuse treatment, however, then logically there should be an opportunity to refuse state screening.⁶

The lack of rigorous informed consent in state NBS programs has not gone unchallenged, including by the 1975 National Academy of Sciences NBS report. However, obtaining consent in the newborn period is difficult. In the days after a birth, parents are rarely able to absorb highly technical information and to decide whether they would like to test for extremely rare conditions that are very unlikely to apply to their baby. Although state NBS programs make some efforts related to consent, in practice the heel-stick falls under orders for routine newborn care, and many parents do not recall the test at all. Prenatal or postnatal periods are likely better times for families to carefully consider their desire for all the information that NBS can provide; however, connecting those parental decisions to the perinatal collection of blood spots remains challenging. One solution may come with technological advances in prenatal genomics, which allow families to choose broad screening as a prenatal clinical service rather than through a state public health program.

CONCLUSION

By 1968, more than a million newborns had been screened for PKU in the United States through state- and hospital-based programs. A consensus conference of international experts debated phenylalanine cutoffs, the nature of hyperphenylalaninemia, proper treatment of PKU, and the patchwork of long-term follow-up options. In short, there was no consensus about any of the critical questions surrounding NBS for PKU. Joseph Cooper, a political scientist at Howard University who was also involved in the 1975 NAS report, had been invited to provide commentary on the broader implications of state NBS programs for PKU. He was incredulous that state governments were testing for PKU given the extraordinary events in the United States at the time—the assassinations of Robert Kennedy and Martin Luther King, civil unrest over Vietnam, and a general sense that the United States was so bitterly divided that it was falling apart. Was state NBS truly a priority?⁶

It is natural when viewing historical events, even in the recent past, as simple and perhaps obvious in their outcome. The ubiquitous statement in NBS articles that “state NBS is a great public health achievement” is true: it is exceptional for American society to pool resources, mandate a blood test on babies, and pursue a unified national strategy to improve the health of every infant using an evidence-based, transparent approach. Almost all other health care in the United States is individual, episodic, and highly dependent on social drivers such as access to insurance.

There have been clear benefits because of universal NBS programs, even if the full benefits of treatment remain unrealized.

The common statement about NBS also hides the deep and contentious debates since the 1960s. Advances in science, medicine, and technology, such as MS/MS, have periodically challenged state programs and clinical systems to expand their work. The criteria for adding conditions to state panels has varied over time, and lately it has included debates about conditions with different severity levels, variable ages of onset, and expensive or difficult treatments. Indeed, adding conditions to state panels has always been political in the best sense of the word: do our societal values align best with limiting state programs to public health emergencies, or should we also provide information to families that some families find valuable?^{23–25} Lastly, financial and legislative support of state NBS has waxed and waned; the Newborn Screening Saves Lives Act, for example, has enjoyed both broad bipartisan support and difficulties being reauthorized. Whereas some have been eager to expand the reach of state NBS, others expressed concerns about privacy and state overreach.

Perhaps we are in similar times, with disruptions in NBS akin to those of previous decades. Indeed, in late 2023 Congress recognized these challenges and appropriated funds for the National Academy of Science Engineering and Medicine (NASEM) to address persistent issues in state NBS programs. Charged with envisioning the future of NBS, the NASEM committee released its report in spring 2025.¹ It reaffirmed the fundamental value of state NBS and its focus on conditions that are serious, urgent, and treatable. The committee recommended increased cooperation among all stakeholders, research infrastructure to inform NBS, more support for families after screening, and improvements to federal systems including the evidence review process for RUSP expansion. Their recommendation to establish a multi-stakeholder group to address fundamental issues in state NBS can be successful if the membership appropriately reflects the various constituencies, the decision-making processes are transparent, and the conclusions are consistent with broadly accepted values of our nation.

Like the 1975 NAS report, the 2025 NASEM recommendations are a road map to expand what is good in state NBS programs. But this time feels different. None of the challenges of previous decades seriously threatened the nature of the enterprise, and recent events potentially undermine both the traditional public-health rationale for NBS and the federal role in promoting policy that is based on open and thorough discussion and the best available evidence. One critical difference in the 2020s is the loss of faith in science, medicine, and academic institutions more generally; whatever the crises of the 1960s, it was a time of broad consensus on the value of science and modern medicine. We are at yet another inflection point

in state NBS, and the question remains whether state NBS will continue to be an exemplary state public health program that provides clear benefits to thousands of children and their families every year. History teaches us that concerted efforts by individuals are necessary to shape policy to align with even self-evident values such as the health and well-being of children. NBS advocates have work to do.

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ABBREVIATIONS

AAP: American Academy of Pediatrics
ACHDNC: Advisory Committee on Heritable Disorders
Newborns and Children
ACMG: American College of Medical Genetics and
Genomics
APHL: Association of Public Health Laboratories
HHS: Health and Human Services
MCHB: Maternal Child Health Bureau
MS/MS: tandem mass spectrometry
NAS: National Academy of Sciences
NBS: newborn screening
NIH: National Institutes of Health
PKU: phenylketonuria
RUSP: Recommended Uniform Screening Panel
WGS: whole genome sequencing

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