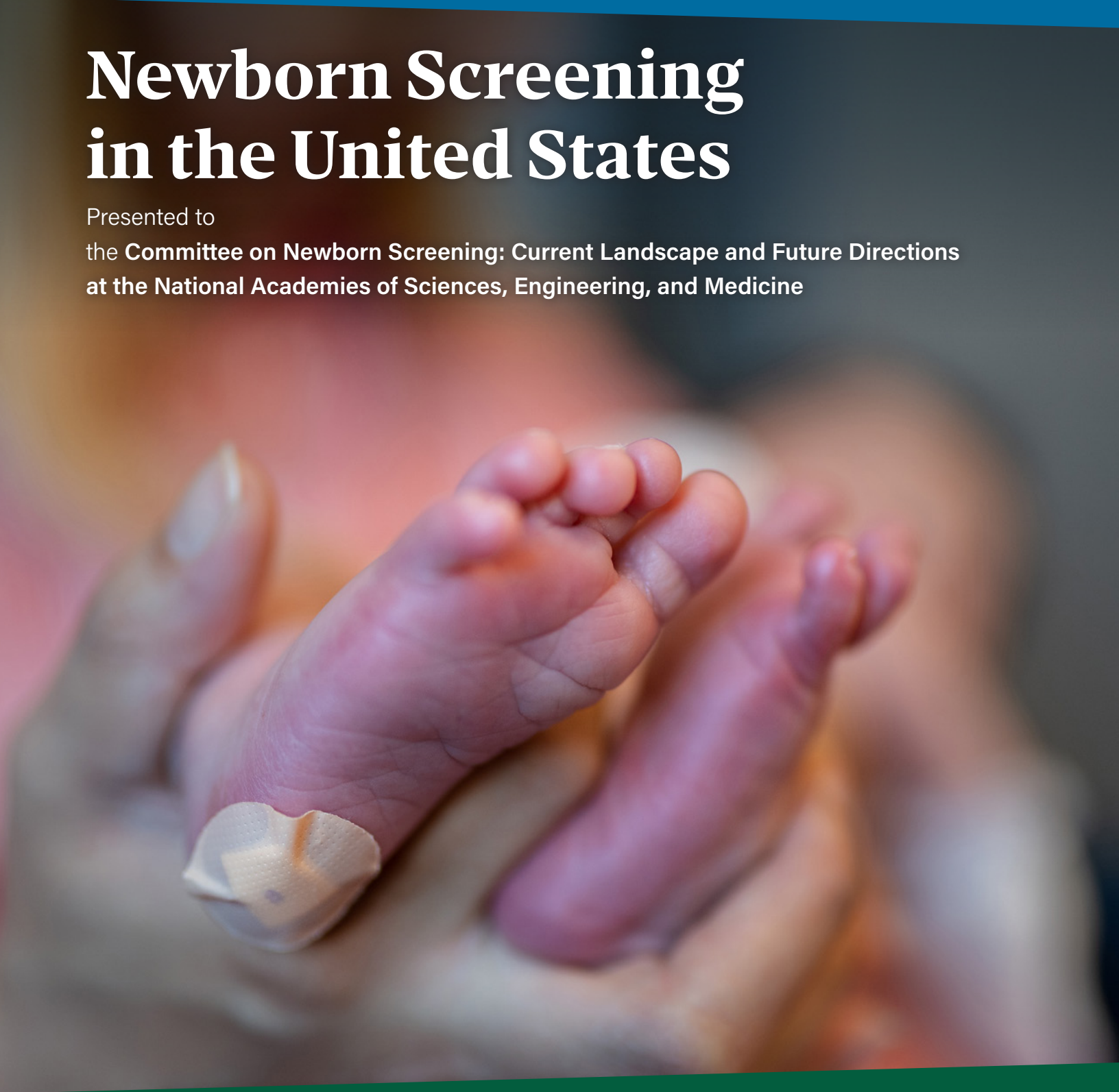
What We Heard

Executive Summary

Newborn Screening in the United States

Presented to
the **Committee on Newborn Screening: Current Landscape and Future Directions at the National Academies of Sciences, Engineering, and Medicine**



September 2024 | Prepared by Susanna Haas Lyons Engagement Consulting

SUSANNA HAAS LYONS

Thank You

This report is written with appreciation for the almost 600 questionnaire respondents, nearly 100 people who attended a 2.5-hour online listening session, 12 National Academies staff who facilitated and took notes for the listening sessions, and 14 members of the National Academies Committee on Newborn Screening: Current Landscape and Future Directions who are devoting hours of time to examine the strengths of existing programs and future opportunities to improve newborn screening for all babies born in the United States.

In particular, we thank The Akari Foundation and REACHUP, Inc., who helped connect the National Academies with communities whose voices might otherwise be less heard in a process like this.

We also thank the following organizations for helping to share the National Academies' call for input on this project. Please note that the information presented in this paper does not reflect the views of any organization or agency that helped share the call for input.

Akari Foundation
Alabama Rare
American Academy of Pediatrics
American College of Obstetrics and Gynecology
American Public Health Association – Public Health Nursing, Community Health Worker, and Maternal and Child Health Sections
Association for Creatine Deficiencies
Association of Maternal and Child Health Programs
Association of Public Health Laboratories
Children's Sickle Cell Foundation, Inc.
E.WE Foundation
EveryLife Foundation for Rare Diseases
Expecting Health
March for Moms
Gaucher Community Alliance
Genetic Alliance
Global Genes
Hunter's Hope
March of Dimes
Muscular Dystrophy Association
National Coordinating Center for Regional Genetic Networks
National Niemann-Pick Foundation
National Organization for Rare Disorders
National Organization of African Americans with Cystic Fibrosis
Parent Project Muscular Dystrophy
Rare Disease Diversity Coalition
Rare Diseases Clinical Research Network
REACHUP, Inc.
Sickle Cell Reproductive Health Education Directive

Suggested Citation

Susanna Haas Lyons Engagement Consulting. 2024. *Executive Summary: What We Heard: Engagement Summary for Committee on Newborn Screening: Current Landscape and Future Directions.* National Academies of Sciences, Engineering, and Medicine. Washington, DC.

About Susanna Haas Lyons Engagement Consulting

Susanna Haas Lyons Engagement Consulting designed and implemented the multi-party process that collected and analyzed the data in this report. This consulting group develops strategy and provides training for better conversations between the public and decision-makers. Susanna Haas Lyons Engagement Consulting has over 20 years of international leadership experience in designing, facilitating, managing, and evaluating complex civic engagement efforts.

Authors and data analysis team for this report are Susanna Haas Lyons, Civic Engagement Specialist; Anson Ching, Civic Engagement Specialist; and Kiana Alaei, Data Analyst.

The engagement team's medical advisor is Charlotte Hobbs, MD, PhD, Executive Associate Dean for Clinical and Translational Research, University of Arkansas for Medical Sciences. The engagement team's health equity advisor is Nicole Farkouh, Founder, CultureWonk.

SUSANNA
HAAS LYONS

Executive Summary

Newborn screening (NBS) programs touch almost every family with children in America, identifying babies at risk of serious but treatable conditions. There are three NBS tests: dried blood spot, hearing loss, and congenital heart defect testing. The engagement activities described in this report focused only on the collection and analysis of newborn bloodspots, during which a small amount of blood is collected from a baby's heel shortly after birth and screened for a set of conditions. NBS enables doctors to diagnose conditions quickly and start treatment as soon as possible.

Recently, Congress directed the Department of Health and Human Services' Office on Women's Health to commission a study with the National Academies of Sciences, Engineering, and Medicine to identify key state and federal actions that could help to modernize these programs. Supplemental funding was provided by the Chan Zuckerberg Initiative to enable enhanced community engagement.

This committee's report will provide both short-term options to strengthen existing NBS programs as well as a vision for the next 5-15 years.

For more information about the committee's work, please visit nationalacademies.org/newborn-screening.

INPUT FROM PEOPLE IMPACTED BY NEWBORN SCREENING

Although committee members bring a wide range of expertise and perspectives on NBS, it was essential for them to hear from people who are personally and professionally affected by NBS in the United States.

Two main engagement activities were carried out for the committee. The first was a series of online listening sessions, held in May and June 2024, and the second was an online questionnaire hosted on the project website, which was open from April 18 to May 26, 2024.

The committee's core questions to engagement participants were: *How can we strengthen today's newborn screening programs? What changes would you like to see in the future?*

A total of 570 questionnaire responses were complete (93%) or partially complete (7%) and included in the analysis. About 100 people participated in virtual listening sessions.

Perspectives represented in this document do not represent a statistical sampling of the American public. Respondents were mostly white (77%), well-educated (70% graduate or professional degree), women (76%), from urban or mostly urban areas (80%). Responses to the questionnaire were largely from people who primarily identified as parents (general parents 16%; parents of a child with rare disease 14%), healthcare providers (27%), or persons impacted by or advocating for rare diseases (a total of 20% have a rare disease, are parents of a child with a rare disease, or are rare disease advocates). Demographic information was not collected from the 97 individuals who participated in the listening sessions. Limited participation from diverse and often underrepresented individuals means these activities may have missed certain perspectives. Further work is needed in this space to ensure an understanding of the full breadth of perspectives on newborn screening, a public health program serving all babies born in the United States.

WHAT WE HEARD

Engagement participants (questionnaire respondents and listening session participants) shared their perspectives on the strengths and challenges of current NBS programs in the United States, as well as on short-term and long-term improvement opportunities. As a whole, the input highlighted some key tensions and sometimes contradicting positions. This dynamic is reflective of the diverse range of key actors, groups, and communities who have strong stakes in NBS, as well as the fact that NBS in the United States relates to moral, political, and medical considerations.

NBS Purpose

Engagement participants were asked what they think should be the purpose of NBS in the United States. The majority of participants suggested that NBS should exist to save lives and prevent rare diseases from having serious consequences—by **detecting rare diseases with effective treatments as early as possible**. Others indicated that NBS should serve the broader purpose of supporting parents and providers to make informed decisions about a baby's healthcare. Additional purposes mentioned include the role of NBS in equitable health outcomes and its importance in supporting access to follow-up care.

Strengths of Existing NBS System

When asked about the strengths of the existing NBS system, the majority of engagement participants highlighted that **all babies receive NBS** in the United States, except for families who opt out. A number of participants pointed to the **follow-up on screening test results** with families as a strength. Input also pointed to the fact that, in most cases, NBS is effective in leading to proactive identification of screened rare diseases and conditions that will benefit from early treatment. Questionnaire respondents also specifically described the practicality and cost effectiveness of bloodspot collection in NBS programs.

Current Challenges

Engagement participants were asked about NBS challenges, in the spirit of looking for opportunities to strengthen the system. Participants said the leading challenge of the existing NBS system is the process for **adding new conditions** to NBS programs. A number of participants also highlighted **parent education and support** as a challenge. Other concerns included state-level variation, follow-up care, state administration of NBS programs, research to develop new screening tests, federal guidance on conditions to be screened, and data management and administrative inefficiencies.

Adding Conditions to NBS Programs

Input from engagement participants provided several nuanced views regarding the challenge of adding rare diseases and disorders to NBS programs in the United States. For some, the issue lies with the **criteria of the Recommended Uniform Screening Panel (RUSP)**. Those participants see the RUSP as too restrictive, currently creating a longer diagnostic process for parents and their babies who have rare diseases not included in NBS screening. Some of these participants want the RUSP to test for all screenable conditions so parents can be informed in their efforts to care for their children. A major tension with this view, however, is the view of other participants that **NBS programs need to be evidence-based**, have existing treatments for the condition, and have reliably accurate screening methods available.

Parent Education and Support

The need to support parents **to understand and navigate NBS** was strongly emphasized by engagement participants. Many highlighted that parents rarely know about NBS until they receive a screening result that needs further evaluation; they recommended earlier and more robust parent education. Participants also called for ways to address public distrust in government health programs and apprehension about genetic information sharing. Participants suggested more nuanced **privacy protections** and policies for transparent storage and use of NBS samples and data. The need to address socioeconomic, geographic, and racial and ethnic **inequities** was also discussed in the listening sessions, especially in relation to NBS test accuracy and all forms of follow-up care. In addition, some listening session participants mentioned an unfair advocacy burden on parents and rare disease groups and highlighted the financial and emotional burdens of a child's diagnostic odyssey.

State-level Variation

One of the most prominent themes to come out of this engagement input was how NBS programs differ across the United States. Many engagement participants pointed out that even though the RUSP recommends diseases and conditions for states to consider incorporating in their NBS programs, the **processes and timelines for adding new conditions vary drastically across the country**. This disparity was often connected to the observation that states vary in their healthcare workforce capacity and NBS infrastructure. NBS variation was also noted by some as being more noticeable within states with large **rural-urban divides**. Other participants emphasized that having national NBS standards could lead to more equitable health outcomes. Some called for increasing the

standardization of test implementation and for strengthening the connections and coordination from screening in state-run NBS programs to follow-up care settings. Another potential solution suggested by participants is increased regional and **inter-state collaboration** for testing and follow-up care.

Barriers to Follow-up Care

Both questionnaire respondents and listening session participants emphasized the importance of follow-up after NBS screening results, with many highlighting that substantial **barriers prevent timely disease intervention** and support for patients, parents, and families. Some of the barriers include geographic challenges, logistical and capacity issues, a lack of health insurance, and communication issues with payors. The matter of **health insurance** in the United States was particularly noted by both questionnaire respondents and listening session participants as a barrier to effective NBS follow-up.

Data Management and Administrative Inefficiencies

Data management and administration challenges described by participants include a **lack of standardization** in data collection and analysis, as well as **limited data sharing** across NBS sectors, organizations, and state programs. Participants described that these data challenges can limit screening accuracy, research, coordination of follow-up care, and timely and effective intervention. Increased communication and collaboration within the broader NBS system were identified as key underlying needs.

Staffing Limitations

Many NBS and healthcare participants shared how NBS programs across the country are suffering from **understaffing and staff burnout**. Input also described increasing challenges of staff capacity and readiness in the face of new conditions being added to screening panels.

Insufficient NBS Funding

Engagement participants also expressed concerns about insufficient funding for NBS programs, which **affects the maintenance, expansion, and updating of services**, contributing to disparities in NBS programs. Funding shortfalls were also linked to state-level variations, workforce challenges, limitations in follow-up care, and research barriers. Some participants called for more federal funding and resources to support NBS programs, emphasizing that insufficient funding affects all aspects of the NBS system—from NBS laboratories to testing technologies, follow-up care, and research.

As part of a thought experiment, questionnaire respondents were asked to choose one funding priority for the US NBS system. Respondents were split between the option of investing in treatment for those with rare diseases identified through NBS, and the option of investing in improvements to the current NBS system. Slightly fewer respondents prioritized adding conditions not currently included in NBS.

Uncertain Screening Results

Listening session participants were asked for ideas on how to address uncertain or unclear NBS results. Most suggested strengthening the NBS **follow-up and referral system** for confirmatory diagnostic testing, counseling, and care. Some also emphasized the need for **systematized longitudinal follow-up**.

Participants generally encouraged efforts to limit unclear results. For some, the solution would be to avoid screening for diseases without reliable screening tests. Others suggested that ambiguity in screening results could be accepted as a potential outcome of a changing NBS system.

Bloodspot Research

Residual blood spots are dried blood spots that are 'left over' after all screening tests have been completed. These samples are deidentified and used to improve NBS for future infants and can also be used for other types of research. Regulations for research uses vary state to state. Questionnaire respondents were asked, as a thought experiment, to share their views on the role of consent for different scenarios involving the use of residual blood spots for research. Respondents' perspectives on the role of consent in this process were split, with some indicating that parents should be notified and asked for consent regarding storage and secondary research use of dried blood spots and others indicating that notification and consent were not essential.

Collaboration and Communication

Participants recommended **greater collaboration across the NBS system**. Seeing **families as partners** was recommended for both the determination of which diseases to screen for, as well as developing ways to improve families' experiences with NBS. Other input described the benefits of collaboration among federal and state governments, state NBS programs, advocacy groups, advisory bodies, medical professionals, NBS professionals, industry, universities, researchers and others.

Final Advice to the Committee

To strengthen current NBS programs, engagement participants suggested that the committee consider the following input and ideas:

- Address health outcome **disparities resulting from state-level variation**;
- Improve **equitable access to treatments** using the lenses of race, income, and geography;
- Consider **regionalization for aspects of the NBS system** including testing, follow-up, and/or access to specialists;
- Enhance **parent awareness and education** about NBS before and after birth;
- Promote **collaboration** at all levels of the system in planning the future of NBS;
- **Incorporate innovations in NBS testing**, such as genomic sequencing;

- → Consider ways to **screen for a wider range** of rare diseases and conditions in NBS programs, such as **reviewing and refining the RUSP** more frequently;
- → Continue to be mindful of the role of **data privacy** in NBS in data collection, longitudinal research, and follow-up care; and
- → Increase **funding**, training, and support for the NBS workforce and programs.

Participants called for more **collaboration and communication** in many areas associated with NBS. To better support families, a number of participants called for mechanisms to help people navigate the system, tie community-based resources to clinical care, and leverage public health departments' educational and home visitation systems. Government partnerships were recommended by many as a key solution. Some also suggested collaboration among and between industry, universities, and other researchers.

Lastly, participants emphasized the need to **engage with parents, families, and patients** in planning the future of NBS. Many also highlighted the need to engage **all key actors and groups** representing specialists, pediatricians, minority and Indigenous communities, medical organizations, patient safety organizations, and patient advocacy organizations.

CONCLUSION

Learning about the diverse perspectives of a wide range of people affected by NBS across the country is a critical first step for assessing what might strength NBS in the United States. The participants' diverse input helps articulate key points of consideration—including where there are strong tensions—that the committee will consider when developing both short-term options to strengthen existing NBS programs, as well as a vision for NBS in the United States in the next 5-15 years.

NEXT STEPS

The National Academies committee will carefully review this report and the other data gathered by the committee. The committee will produce a report with its recommendations in 2025.

Full Summary of Input

For the full summary of input shared by engagement participants, please visit nationalacademies.org/newborn-screening.

SUSANNA
HAAS LYONS