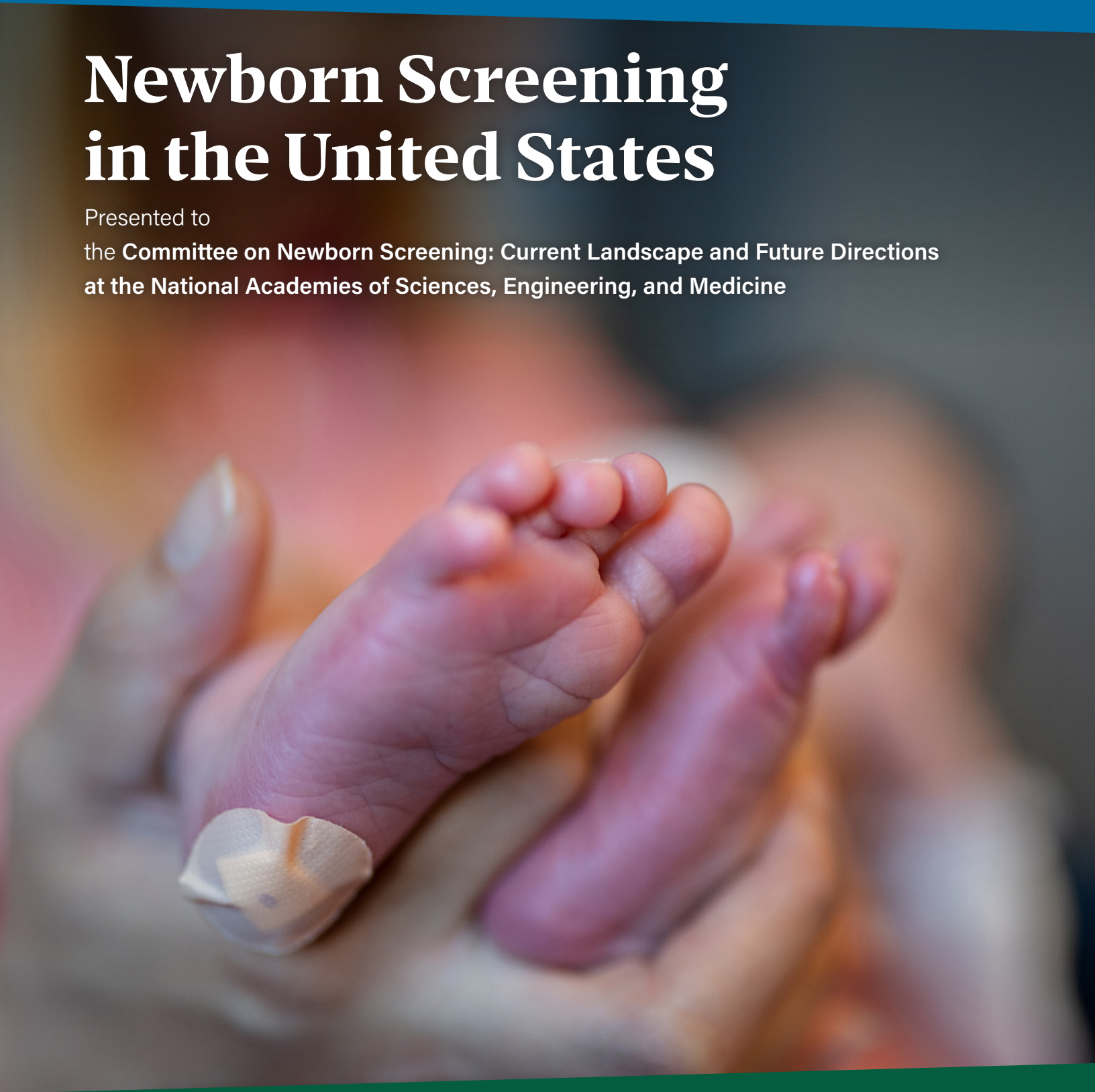
What We Heard: Engagement Summary

# 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

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# Contents

# 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. *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.

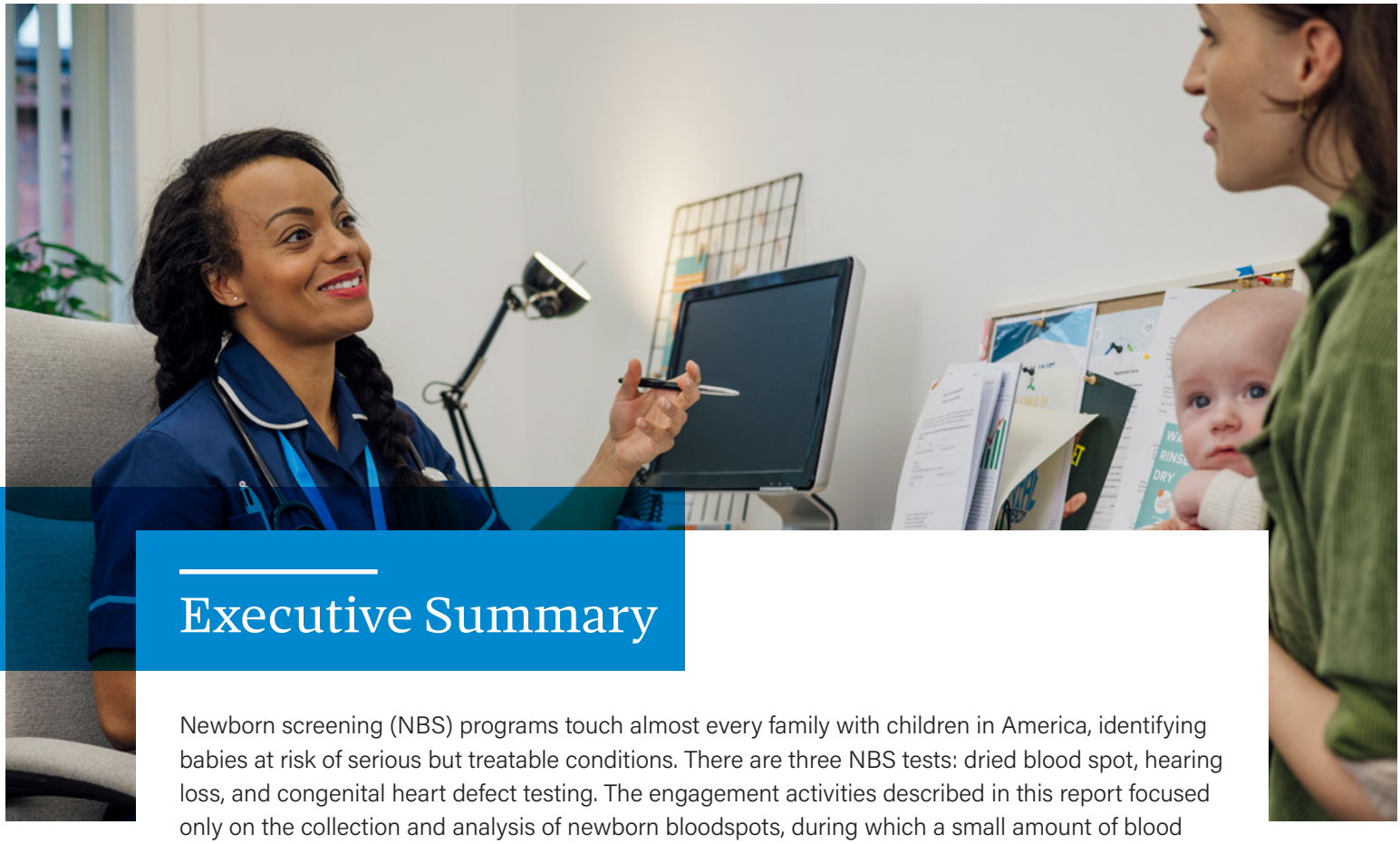
The engagement team's medical advisor is Charlotte Hobbs, MD, PhD, Executive Associate Dean for Clinical and Translational Research, University of Arkansas for Medical Sciences. The engagement team's health equity advisor is Nicole Farkouh, Founder, CultureWonk.

# Abstract

Following a Congressional request, the National Academies of Sciences, Engineering, and Medicine convened a committee on newborn screening (NBS) to identify key state and federal actions that could help to modernize NBS programs in the United States. People who are personally and professionally impacted by NBS in the United States were invited to give input to the committee, and about 670 people were involved through online listening sessions and an online questionnaire. During spring 2024, participants shared their views on how to strengthen today's newborn screening programs and changes they would like to see in the future.

According to participants, early detection of diseases leading to intervention is a key strength of NBS programs. Input about NBS also described challenges of parent education, state-level variation, follow-up care, program administration, data management, research, and administrative inefficiencies.

The committee will carefully consider the input received from the engagement and other sources and produce a report with its recommendations in 2025.



# 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.

## 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.

## GLOSSARY[1]

| | |
|---|---|
| ACHDNC | Advisory Committee on Heritable Disorders in Newborns and Children—a federal committee made up of doctors, scientists, parents, ethicists, and researchers. This group advises the United States Secretary of Health and Human Services about newborn screening and related topics.[2] |
| Blood spot | The sample of blood collected on filter paper cards used in newborn screening. |
| Committee | National Academies committee on Newborn Screening: Current Landscape and Future Directions. |
| Diagnostic testing | Also called confirmatory testing. Testing performed after a baby has a positive result from newborn screening. This test confirms whether the baby has the suspected condition. |
| False negative result | A result indicating that the newborn screening is normal, when a disease is in fact present.[3] |
| False positive result | When a child with an out-of-range newborn screening result has a follow-up test result within the normal range, it is sometimes called a "false positive". The child does not have the condition that the original screening indicated was a possibility.[3] |
| Follow-up care | This term can be used to describe a number of situations after a newborn screening result, including follow-up testing (e.g. confirmatory testing leading to a diagnosis); follow-up communication to inform a healthcare provider and/or the family about NBS results; follow-up visits involving primary care and specialists; longer-term specialty follow-up to meet the needs of the identified baby over an extended period; and/or follow-up data collection by the public health program to monitor disease prevalence and outcomes. |
| Follow-up, short-term | The process of ensuring that all newborns are screened, an appropriate healthcare provider is informed of the results, confirmatory testing is completed, and the infant receives a diagnosis for the existing health condition and, if necessary, treatment.[3] |

1 Glossary entries without citations were developed by the project team for the purpose of the engagement sessions and this engagement summary.
2 Health Resources and Services Administration. (n.d.). *Glossary.* Accessed 22 July 2024. https://newbornscreening.hrsa.gov/about-newborn-screening/glossary
3 Baby's First Test. (n.d.). *Glossary*. Accessed 22 July 2024. https://www.babysfirsttest.org/newborn-screening/glossary

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| Genetic testing | A laboratory method that looks for changes in genes, gene expression, or chromosomes. These changes may be a sign of a disease or condition. They may also be a sign that a person has an increased risk of developing a specific disease or condition.[4] |
| Genomic sequencing | Also called DNA sequencing. A general laboratory technique for determining the exact sequence of nucleotides, or bases, in a DNA molecule. The sequence of the bases (often referred to by the first letters of their chemical names: A, T, C, and G) encodes the biological information that cells use to develop and operate. Establishing the sequence of DNA is key to understanding the function of genes and other parts of the genome.[5] |
| Health equity | Where everyone has a fair and just opportunity to attain their highest level of health.[5] |
| Healthcare industry | Businesses and related organizations that provide medical services and goods. |
| Healthcare provider | Someone with special training in health-related areas, such as a doctor, nurse, physician's assistant, or genetic counselor.[3] |
| In-range screening result | Also referred to as a negative result. A result that indicates that the baby's blood test did not show any signs of the conditions included on the newborn screening panel.[3] |
| National Academies | National Academies of Sciences, Engineering, and Medicine. |
| Negative screening result | Also referred to as an in-range result. A result that indicates that the baby's blood test did not show any signs of the conditions included on the newborn screening panel.[3] |
| NBS | Newborn screening. |
| Newborn screening panel | A list of conditions that a baby will be screened for after birth. Each state has its own panel.[3] |
| Newborn screening program | Checking (screening) babies for certain serious conditions; identifying those few who might have one of these conditions; and helping to connect babies with the early care treatment, and/or intervention they need to give them the best chance at a healthy life.[6] |

4 National Cancer Institute. (n.d.). NCI Dictionary of Cancer Terms. Accessed 22 July 2024. https://www.cancer.gov/publications/dictionaries/cancer-terms/

5 Centers for Disease Control and Prevention. (2024). *What is Health Equity?*. Accessed 22 July 2024. https://www.cdc.gov/health-equity/what-is/

6 Health Resources and Services Administration. (2023). *About Newborn Screening.* Accessed 22 July 2024. https://newbornscreening.hrsa.gov/about-newborn-screening/glossary

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| Newborn screening system | The informal network of sectors and partners that support the identification, follow-up, and treatment of babies with certain serious conditions. These partners include public health laboratories and professionals, care providers, payors, advocacy groups, patients, parents, regulatory agencies, and industry and academic researchers, among others. |
| Pathogenic variant | A change in the DNA sequence of a gene that causes a person to have or be at risk of developing a certain genetic disease or condition. Not everyone who has a pathogenic variant will develop the disease.[4] |
| Positive screening result | This result indicates that the baby's screening exam identified that they may be at higher risk of having one or more of the conditions included on the newborn screening panel. A positive result does not mean that the baby has a medical condition. Follow-up testing must be performed immediately to determine if a condition is actually present.[3] |
| Quality assurance | A dynamic process of defining the quality of performance required for each step in the testing process.[3] |
| Quality control | The mechanisms for monitoring the degree of adherence to defined criteria, taking corrective action when the system fails, and documenting relevant events to convey the total quality of performance.[3] |
| Rare disease | An uncommon disorder that affects the ability of the human body to function normally.[3] |
| RUSP | The Recommended Uniform Screening Panel is the list of conditions for which the United States Secretary of Health and Human Services recommends newborns receive screening.[2] |
| Residual dried blood spot | The small amount of dried blood that remains on the filter paper cards after newborn screening has been performed.[3] |
| Treatment | In the context of NBS and RUSP condition review, the treatment approved by the United States Food and Drug Administration to improve morbidity and/or mortality.[7] |
| Variant | Gene variants are different versions of the same gene.[2] |
| Variant of unknown significant | Also known as variant of uncertain significance. When analysis of a person's genome identifies a variant, but it is unclear whether that variant is actually connected to a health condition.[8] |

7 Health Resources and Services Administration. (2022). *Key Questions Considered by the Committee*. Accessed 22 July 2024. https://www.hrsa.gov/advisory-committees/heritable-disorders/key-questions

8 National Human Genome Research Institute. (n.d.). *Talking Glossary of Genomic and Genetic Terms*. Accessed 22 July 2024. https://www.genome.gov/genetics-glossary

# Introduction

An ad hoc committee of the National Academies of Sciences, Engineering, and Medicine (the National Academies) has been tasked with examining the current landscape of newborn screening (NBS) systems, processes, and research in the United States. The committee on Newborn Screening: Current Landscape and Future Directions (committee) will make recommendations for future improvements to help modernize NBS. The recommendations will aim to make NBS more adaptable, flexible, coordinated, and communicative; enhance its ability to efficiently and sustainably incorporate new conditions and technologies; and ensure it becomes a more equitable public health program from which all infants benefit.

## ABOUT NEWBORN SCREENING

NBS programs are **state-run public health programs** that identify babies at risk of rare, serious, but treatable conditions before symptoms develop. These conditions are often described as rare diseases—uncommon disorders that affect normal functioning of the human body. Identifying these conditions may help prevent brain damage, physical disabilities, or death. Nearly every family with children in the United States has been touched by NBS; over 98%[9] of the approximately 3.6 million babies born annually are screened.[10] Although NBS encompasses dried blood spot, hearing loss, and congenital heart defect testing, **the scope of the committee's work is limited to dried blood spot screening**.

9 Centers for Disease Control and Prevention. (2012). *CDC Grand Rounds: Newborn screening and improved outcomes.* Accessed 20 June 2024. https://www.cdc.gov/mmwr/preview/mmwrhtml/mm6121a2.htm

10 Centers for Disease Control and prevention. (n.d.). *NVSS - Birth Data.* Accessed 20 June 2024. https://www.cdc.gov/nchs/nvss/births.htm

NBS is typically performed in the first 24–48 hours after birth. Staff from the hospital or birth center collect blood from the baby's heel on a special card. Samples are sent for screening, and then the results are sent to the baby's healthcare provider.

Each state or territory independently determines the conditions for which its program will screen. However, a Recommended Uniform Screening Panel (RUSP) is provided by the US Secretary of Health and Human Services with the support of the federal Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC). Conditions added to NBS tend to meet the following criteria:

- **Urgent**—requiring treatment as early in life as possible;
- **Severe**—producing serious health effects if untreated; and
- **Treatable**—having an effective medical treatment available.

Ultimately, states and territories choose conditions to address the needs of their populations and comply with factors such as legislative requirements, budget, workforce availability, and technological resources.

State-run NBS programs work with partners in a large system that supports research, identification, follow-up, and treatment. These partners include regulatory agencies, clinical care, advocacy groups, researchers, industry, payors, patients, and parents, among others.

## TASK OF THE COMMITTEE

**The committee's work focuses on the following tasks:**

1. Examine state and federal capacities to strengthen current screening processes and implement screening for new conditions, including considerations for future conditions added to the RUSP.
2. Review existing and emerging technologies that would permit screening for new categories of conditions and describe:
   a) how these new technologies may impact states;
   b) changes to public health infrastructure needed to incorporate new technologies while upholding and implementing the required components of NBS;
   c) options for incorporating new technologies to allow for screening of additional conditions; and
   d) research, technological, and infrastructure needs to improve diagnosis, follow-up, and public health surveillance.
3. Review NBS data collection processes for tracking disease prevalence, improving health outcomes, conducting longitudinal follow-up, ensuring health equity, defining the natural history of conditions that can be screened for, and measuring quality of life.

4. Examine the review and recommendation processes for the RUSP, including the process for:

   a) selecting new conditions that could be added to the RUSP;

   b) conducting reviews of the evidence to support adding new conditions;

   c) scaling up these review and recommendation processes to efficiently handle the review of potentially hundreds of conditions; and

   d) considering whether additional factors should be included in the analysis of harms and benefits (e.g., societal harms such as financial cost or opportunity costs, and family benefits such as avoiding the "diagnostic odyssey").

## COMMITTEE REPORT

**The committee will write a report that describes:**

- **Short-term options** that could be implemented at the state and/or federal level over the next 2-3 years to help strengthen existing NBS programs and address the current challenges facing state programs; and

- A **vision for the future** of NBS and a roadmap for how to implement and achieve that vision over the next 5-15 years.

The report will include options for how to implement longitudinal follow-up data collection to improve understanding of the impact of NBS on infant health outcomes (including morbidity and mortality, and quality of life for screen-positive infants). The committee will also consider the resources required for implementation, such as changes to the current NBS system that will need to occur, the feasibility of implementing the future vision, and the challenges and barriers that may arise when trying to implement the roadmap.

## COMMITTEE MEMBERS

The 14-person committee is chaired by Dr. Jewel Mullen. Dr. Mullen is the Associate Dean for Health Equity and Associate Professor of Population Health and Internal Medicine at the Dell Medical School, University of Texas at Austin, and a public health leader with federal and state-level experience.

Committee members include experts in NBS systems, lived and parental experience, bioethical and legal issues, existing and emerging technologies, health systems, health economics, and clinical care disciplines, among other areas. They bring differing perspectives on the mission and expansiveness of NBS programs, and how to balance benefits, harms, and implications of screening. Learn more about the committee at nationalacademies.org/newborn-screening.

# Engagement Process

While the 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 the topic. The engagement process was designed to **gather input from people and organizations interested in, impacted by, or involved with** NBS about current and future approaches to NBS programs in the United States, as part of the information-gathering to inform the study.

**The committee's core questions for engagement participants were:**

- ➔ *How can we strengthen today's newborn screening programs?*
- ➔ *What changes would you like to see in the future?*

The National Academies' Committee to Review Studies on Human Subjects, acting as the National Academies' Institutional Review Board (IRB), reviewed the engagement approach and related materials in spring 2024. After revisions, approval of IRB exemption was given for the proposed engagement activities.

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

The listening sessions and questionnaire were promoted to people across the United States. The committee and project staff identified associations, organizations, and other kinds of groups interested in or impacted by NBS, and then reached out to these groups to invite individuals who could participate in the listening sessions or respond to the questionnaire. Information about the project, along with ways to participate, was disseminated to the identified groups.

In addition, information about the listening sessions and questionnaire was disseminated through National Academies' Health and Medicine Division email and social media channels, and made available on the study website (nationalacademies.org/newborn-screening). The aim was to obtain input from individuals reflecting a diversity of NBS experiences in the United States.

This engagement process enabled the committee to gain insight from parents of children with a rare disease or condition, general parents, persons with a rare disease or condition, rare disease advocates, health administrators, healthcare providers, health researchers, health industry representatives, NBS lab professionals, NBS follow-up professionals, public health professionals, payors, privacy advocates, and the general public. Engagement was conducted in English and Spanish.

> **How can we strengthen today's newborn screening programs? What changes would you like to see in the future?**

## QUESTIONNAIRE

An online questionnaire was open for public input for 38 days (from April 18 to May 26, 2024). The questionnaire was developed using Alchemer, a feedback and data collection platform. It was available in both English and Spanish. The questionnaire had general questions, as well as targeted questions that used branching logic to show specific sets of questions according to a participant's category in relation to their connection to NBS. Respondents were asked limited demographic questions at the end of the questionnaire for the purpose of understanding and reporting what types of people participated.

## LISTENING SESSIONS

The National Academies held four virtual listening sessions between May 1 and May 13, 2024. These sessions were professionally facilitated and held online using Zoom. Participants were invited to register in advance via the National Academies' project website. The main purpose of the listening sessions was for participants to discuss:

- Key challenges that they experience or see in the current NBS systems, processes, and research context in the United States;
- Explore potential responses to the challenges; and,
- Criteria that should be involved in adding new conditions to the RUSP.

In advance, participants received a "What to Expect" overview of the Listening Sessions (see Appendix G), which included an agenda with the questions to be discussed. A short presentation was shared at the start of each of the listening sessions to ensure that all participants had at least a basic understanding of NBS and the work of the committee.

Each listening session brought together a specific category of participants to enable focused discussion. A fifth session to hear from general families was anticipated but not held due to low registration. The dates and categories of the listening sessions are in the following table. The first two sessions were fairly homogenous, while the second two were more mixed, with about two-thirds of participants identifying with the sector focus, and others coming from mixed sectors.

| | |
|---|---|
| **Wednesday, May 1, 2024** | Newborn screening laboratory and follow-up professionals |
| **Sunday, May 5, 2024** | Rare disease patients, families, and advocacy organizations |
| **Monday, May 6, 2024** | Health administrators, payors, and health industry professionals |
| **Monday, May 13, 2024** | Healthcare providers |

To support participation by historically marginalized groups, two virtual listening sessions were convened with the assistance of organizations that helped the project connect with communities whose voices might otherwise be less heard:

| | |
|---|---|
| **Thursday, May 23, 2024**<br>Convened with the assistance of The Akari Foundation | Spanish-speaking parents of rare disease patients. |
| **Tuesday, June 11, 2024**<br>Convened with the assistance of REACHUP, Inc. | Families with children who are two years old or younger and are eligible for Medicaid or lack insurance altogether. |

# Who Participated

Those who took part in the listening sessions and questionnaire were people with personal and professional experience with NBS in the United States:

- Public health professionals and experts, such as NBS laboratory and follow-up professionals;
- Healthcare providers, such as clinicians, nurses, and birth workers;
- Health administrators and payors;
- Health industry professionals;
- Rare disease patients; and,
- Parents, caregivers, or representatives of advocacy organizations for people with rare diseases.

**Perspectives represented in this document do not represent a statistical sampling of the American public.** The committee wanted to hear from people with expertise and experience across the NBS ecosystem, as well as from families of children with a rare disease, and the general public. Participation was voluntary and intended to provide views and input to inform the study. Participant demographics provided below and in Appendix B are reflective of who shared their views, including the high number of NBS and healthcare professionals who participated.

## QUESTIONNAIRE PARTICIPANTS

A total of 570 questionnaire responses were complete (93%) or partially complete (7%) and included in the analysis. See the Input Analysis Approach section below for further details.

Questionnaire respondents do not reflect a random sample. Respondents came from a few sources: individuals who accessed the questionnaire via a link shared by organizations partnering with the National Academies to disseminate the call for input (see the Thank You section for details); individuals who accessed the link via the National Academies' consensus study webpage; and individuals who were made aware of this opportunity through National Academies' Facebook advertisements, emails, newsletters or social media posts.

## Group Affiliation

The questionnaire asked respondents to answer two questions about sectoral groups with which they identify (Figure 1). The first question allowed respondents to select 'all that apply', while the second question asked them to select the group with which they most identify. Results from the second question (primary group affiliation) are used when reporting questionnaire responses by affiliation group. Those who replied "other" as their primary sectoral affiliation include educators, grandparents, retired NBS and health professionals, and others.

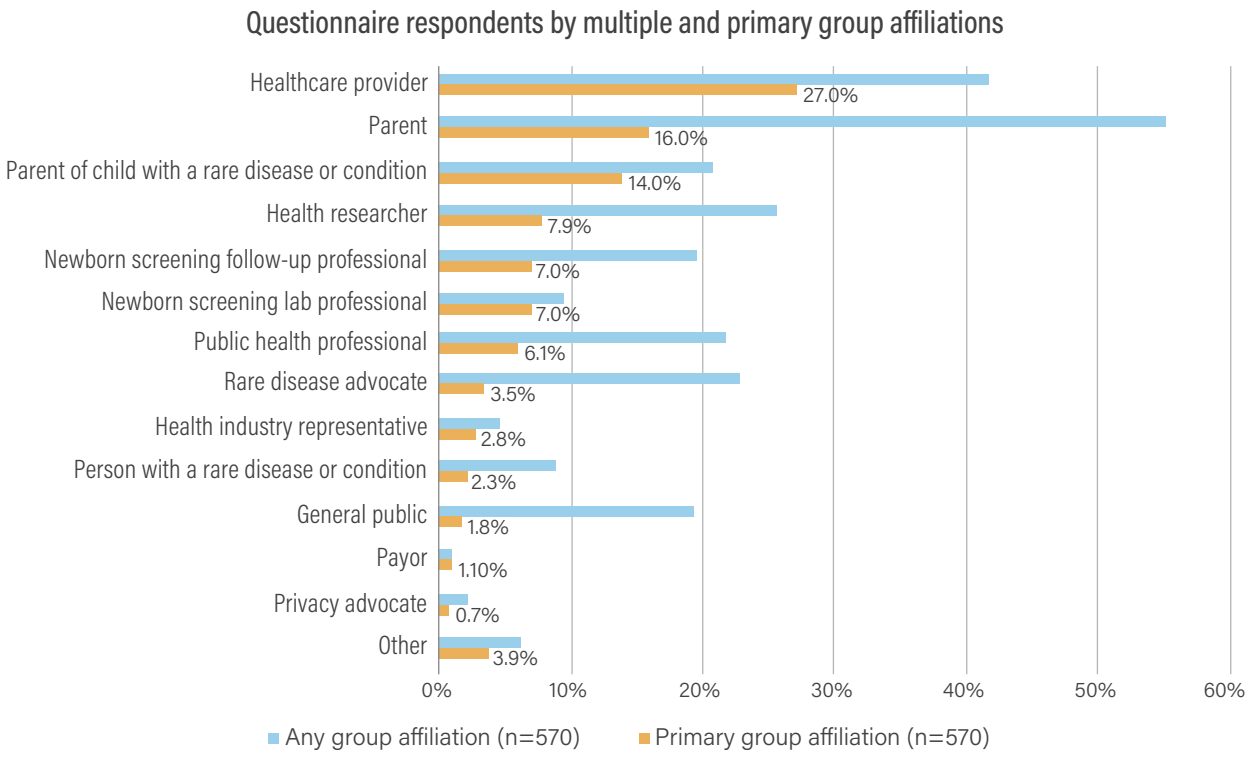



*Figure 1. Questionnaire respondents by multiple and primary group affiliations.*

237 respondents identified themselves as healthcare providers. They were then asked to choose the role that best describes them. The figure below shows the distribution of these roles among the respondents.

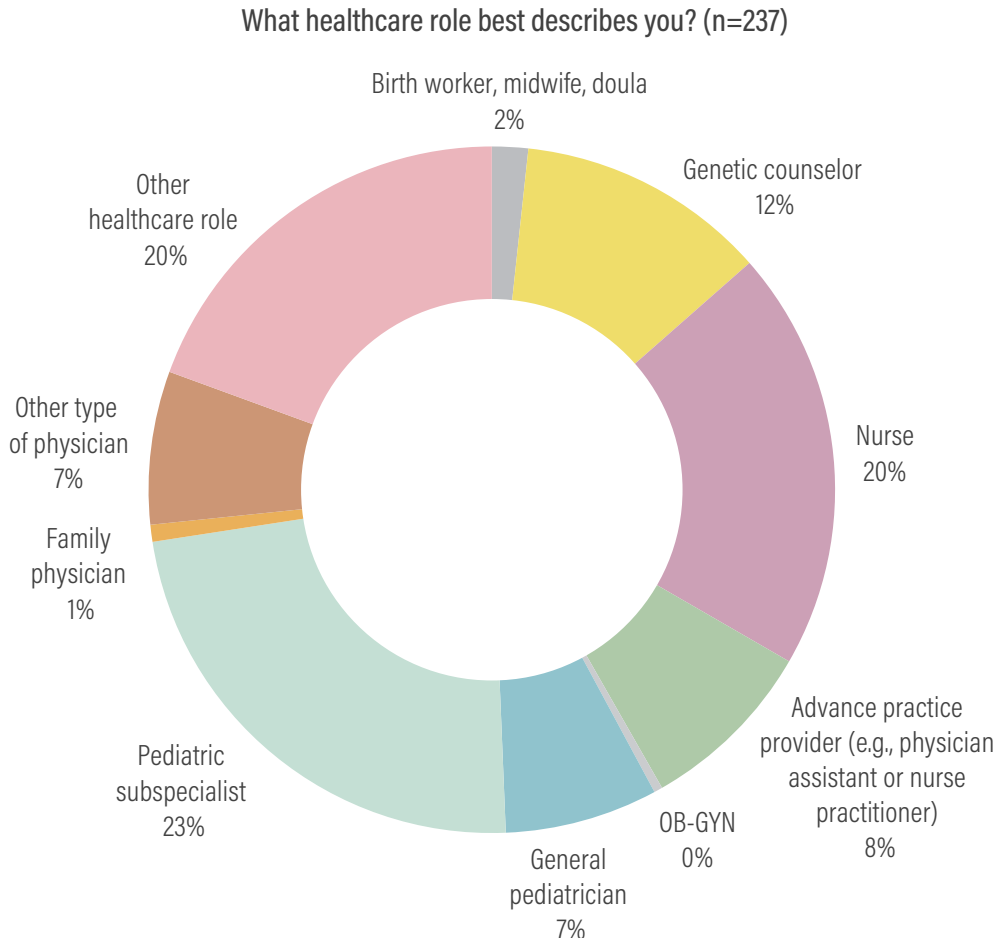



*Figure 2. Questionnaire respondents' healthcare roles.*[11]

11 *One respondent identified as an OB-GYN and this is rounded down to zero in the chart.*

## Rare Disease

20% of respondents identified themselves as a person with a rare disease, parent of a child with a rare disease, or a rare disease advocate. They were then asked specifically: Which rare disease(s) or conditions(s) are you impacted by or engaged with? Adrenoleukodystrophy (ALD) was mentioned frequently. Other rare diseases or conditions commonly mentioned include CTNNB1 Syndrome, metachromatic leukodystrophy (MLD), mucopolysaccharidosis (MPS), sickle cell disease, Duchenne muscular dystrophy, Krabbe disease, cystic fibrosis, Phelan-McDermid syndrome, Menkes disease, and severe combined immunodeficiency (SCID). Approximately 30 other conditions were mentioned at least once.

## Location

About 98% of questionnaire respondents were residents of the United States and territories, involving residents of every US state but one.

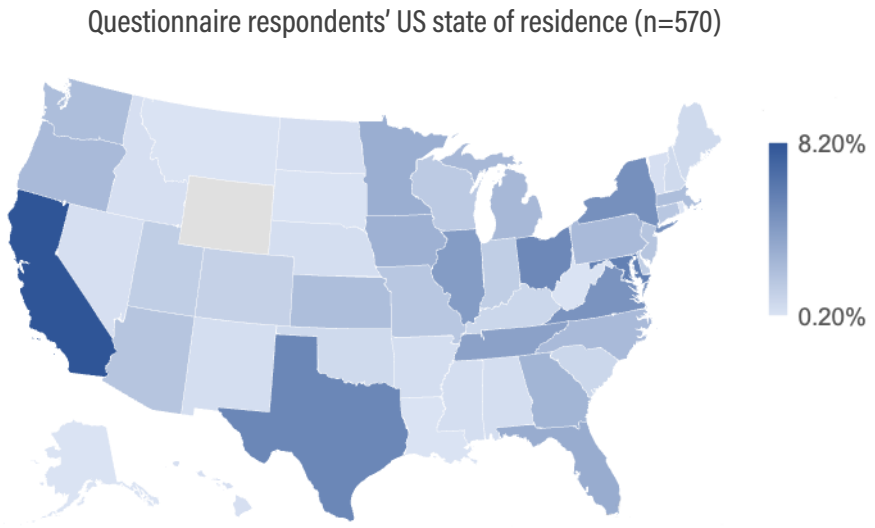



*Figure 3. Questionnaire respondents' location of residence. This map does not display the 2% of respondents outside of the US.*

### Additional Demographics

At the end of the questionnaire, additional demographic questions were asked to better understand the makeup of participants. The demographic questions were prefaced with a disclaimer that the questions were anonymous and voluntary, and these questions were asked at the end of the questionnaire. Fewer answers were received to these questions in comparison to the questions about NBS. A total of 88% of the participants responded to the demographic questions.

Additional demographic figures can be found in Appendix B. As discussed in the Limitations section, participants do not reflect a representative sample of the American population, as the committee wanted to hear from those with lived and professional experience across the NBS ecosystem.

Of those who responded to the demographic questions:

- **Gender:** Three-quarters (76%) of respondents identified themselves as women, about a fifth (15%) as men, and about 1% are non-binary or prefer to self-describe. One percent of respondents indicated that they have lived experience as a trans person.
- **Ethnicity:** 6% of respondents indicated they are of Hispanic, Latino/a/e/x, or of Spanish origin; 2% indicated they are of Middle Eastern or North African origin.
- **Race:** Three-quarters (77%) of respondents identified themselves as White; 7% as Asian or Asian American; 6% as Black or African American; and 1% as American Indian, Alaska Native, Indigenous, or Native American.
- **Urban versus rural:** About 80% of respondents indicated that they live in urban or mostly urban areas. 18% live in rural or mostly rural areas.
- **Education:** Over two-thirds (70%) of respondents indicated that they have a graduate or professional degree, compared to one-fifth (21%) that have a bachelor's degree. 4% have an associates or technical degree; 2% have completed some college, but did not obtain a degree; and 2% have completed some high school or have a high school diploma.

## LISTENING SESSION PARTICIPANTS

Six listening sessions were held in May and June 2024, involving 97 people. Four sessions were convened to hear from:

- NBS laboratory and follow-up professionals
- Rare disease patients, families, and advocacy organizations
- Health administrators, payors, and health industry representatives
- Healthcare providers

Participants of the NBS professionals and rare disease community sessions were almost entirely affiliated with the intended sectors for those sessions. The other two had a mix of participants. The reporting below reflects the composition of these sessions.

To support participation by historically marginalized groups, two virtual listening sessions were convened with the assistance of The Akari Foundation and REACHUP, Inc.:

- *Spanish-speaking parents of children with a rare disease.* This session was convened with the assistance of The Akari Foundation. The Akari Foundation is an organization that educates and empowers the Hispanic community on rare diseases, specializing in Duchenne muscular dystrophy. It offers resources, awareness, advocacy, and education.
- *Families with children who are two years or younger and are eligible for Medicaid or lack insurance altogether*. This session was convened with the assistance of REACHUP, Inc., a Florida-based organization that advocates for and mobilizes resources to help communities achieve equality in healthcare and positive health for families.

## Affiliation

The three largest groups of participants across the six sessions were NBS follow-up professionals, NBS lab professionals, and parents of a child with a rare disease.

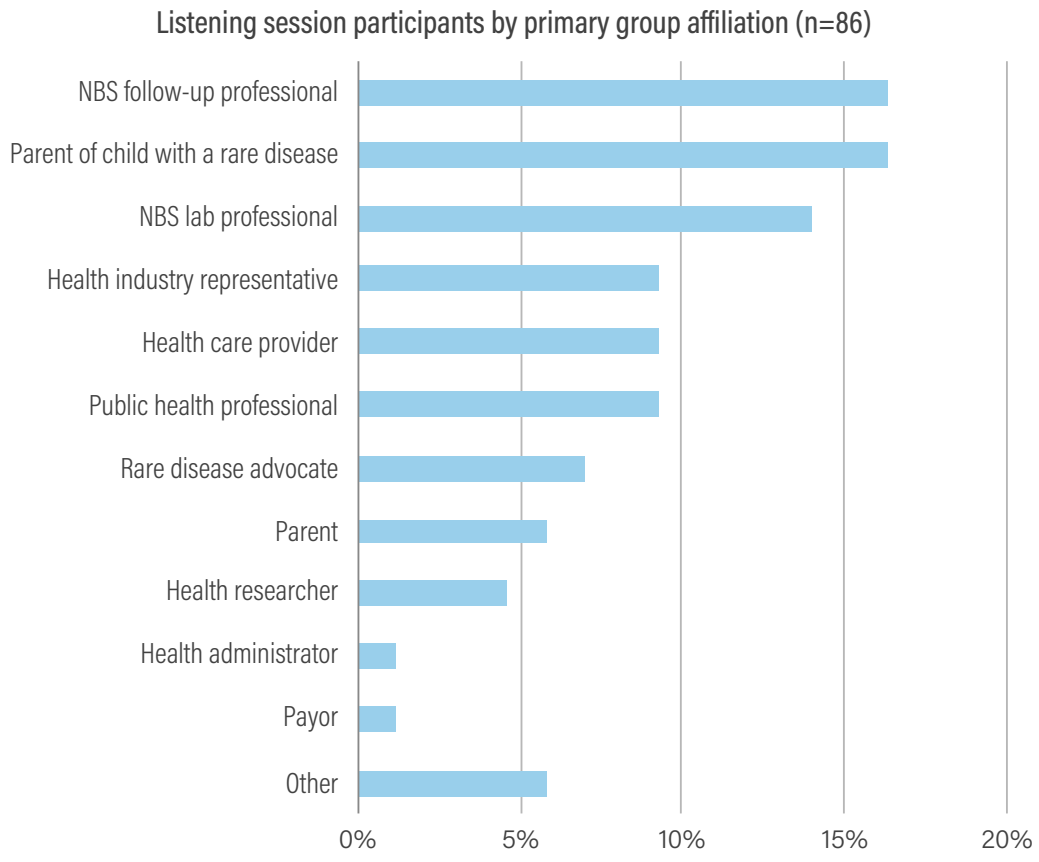



*Figure 4. Listening session respondents by primary group affiliation.*

## Location

Listening session participants reside in a mix of regions, including Puerto Rico, with the most coming from the southern region of the United States.

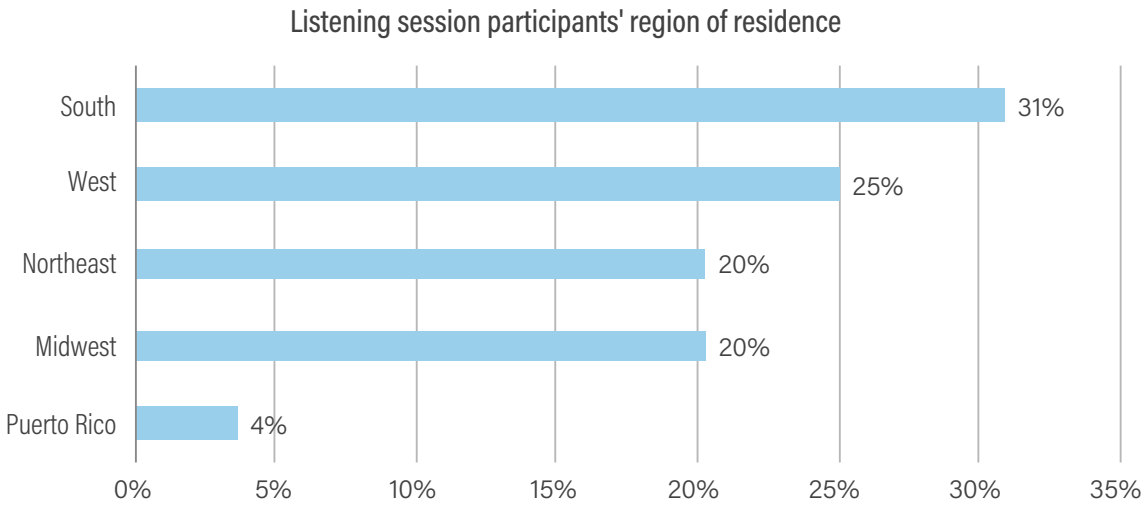
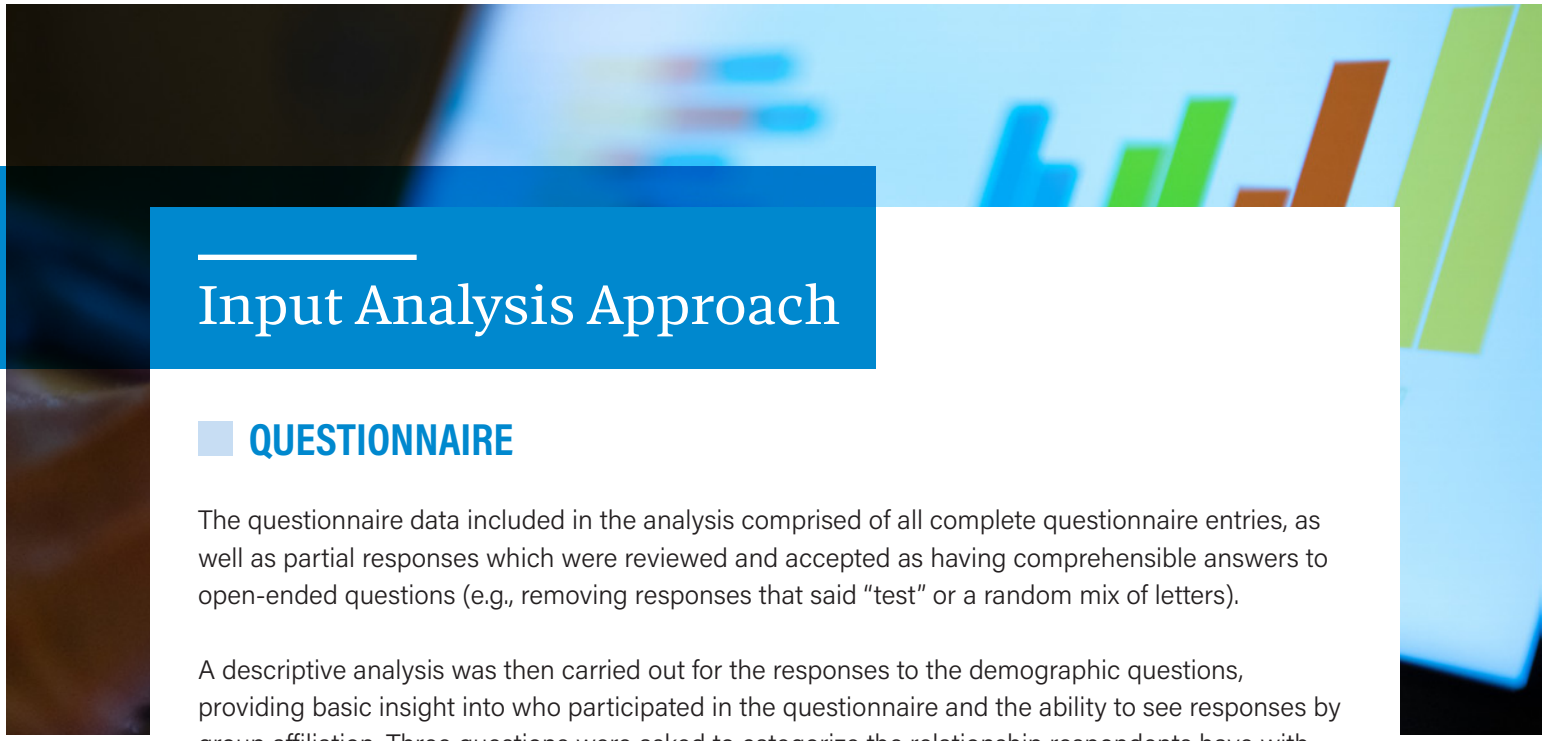



*Figure 5. Listening session participants by region of residence.*



# Input Analysis Approach

## QUESTIONNAIRE

The questionnaire data included in the analysis comprised of all complete questionnaire entries, as well as partial responses which were reviewed and accepted as having comprehensible answers to open-ended questions (e.g., removing responses that said "test" or a random mix of letters).

A descriptive analysis was then carried out for the responses to the demographic questions, providing basic insight into who participated in the questionnaire and the ability to see responses by group affiliation. Three questions were asked to categorize the relationship respondents have with NBS in the United States and their location of residence. Six optional questions were asked about age, gender, ethnic and racial identity, and level of education.

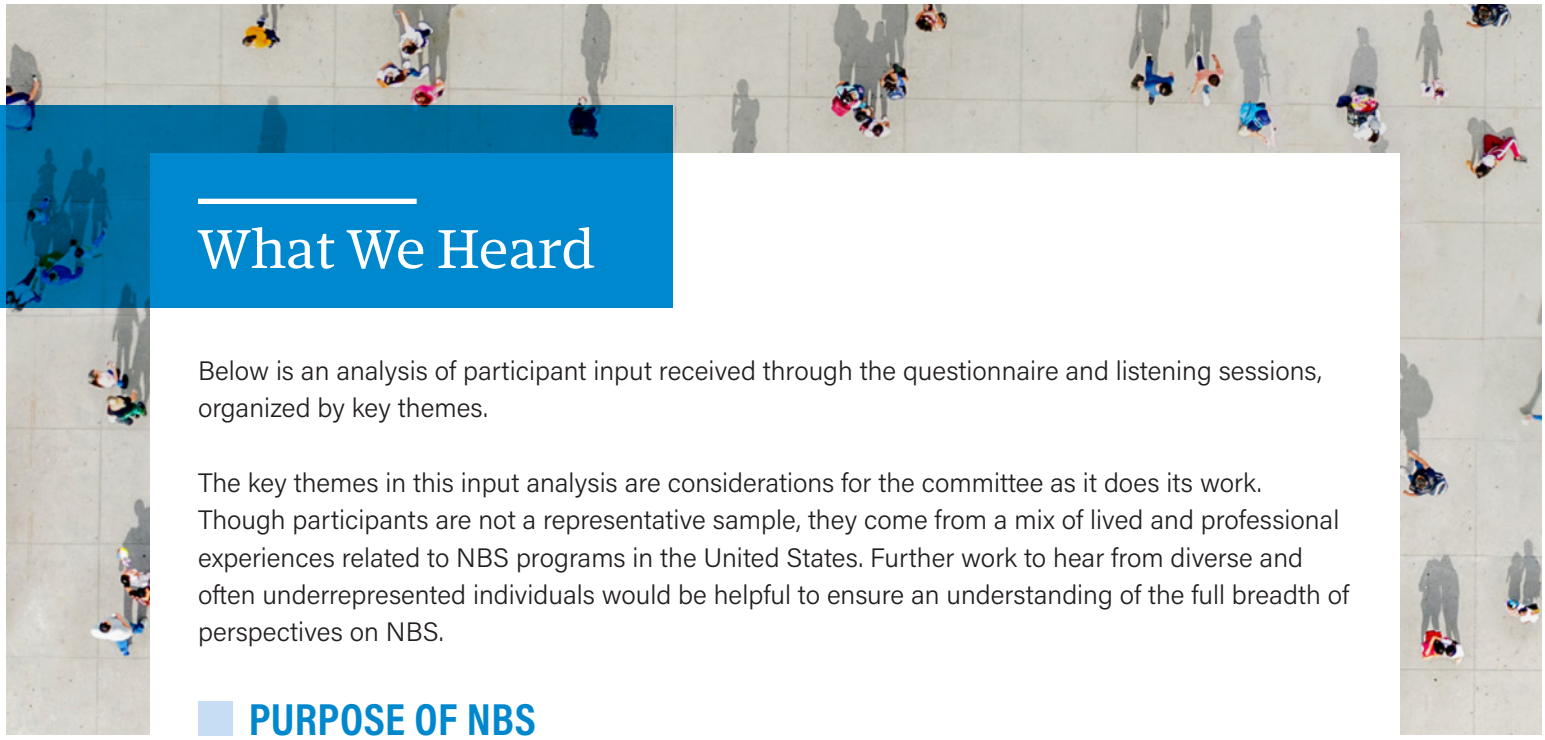
The remaining input gathered from the questionnaire included both closed-ended and open-ended responses to questions on respondents' experiences in relation to NBS in the United States. The responses for the closed-ended questions were analyzed by the engagement team using the built-in data visualization, subgrouping, and cross-tabulation features in Alchemer and Fathom. For the open-ended responses, answers were first grouped thematically using Fathom and then underwent textual analysis conducted by the engagement team.

## LISTENING SESSIONS

Notes from the listening sessions were not attributed to individuals. Input from the listening sessions was captured by notetakers during the plenary and breakout room discussions, as well as the saved Zoom chat. Backup notes were generated by Otter.ai, a software which specializes in producing speech-to-text transcripts, based on the audio recording of the Zoom sessions.

Both the transcripts and notetakers' notes were collated and cleaned (e.g., correcting misspellings, abbreviations, and acronyms) by the engagement team. The gathered input from the listening sessions was then analyzed for key themes and major topics of discussion.

The engagement team generated themes by first uploading the notetakers' notes into Fathom to generate suggested key themes and topics. These themes were then reviewed by the engagement team. As the engagement team actively facilitated the plenary sessions and participated in the breakout room discussions of every listening session, they had the required understanding to review, adjust, and regroup the themes and topics suggested by Fathom, as well as add to them. The weight or relevance of themes and key topics were then analyzed through the data visualization, subgrouping, and cross-tabulation features of Fathom to inform the summary of themes and findings from the listening session discussions.



# What We Heard

Below is an analysis of participant input received through the questionnaire and listening sessions, organized by key themes.

The key themes in this input analysis are considerations for the committee as it does its work. Though participants are not a representative sample, they come from a mix of lived and professional experiences related to NBS programs in the United States. Further work to hear from diverse and often underrepresented individuals would be helpful to ensure an understanding of the full breadth of perspectives on NBS.

## PURPOSE OF NBS

Overall, the majority of participants suggested that the main purpose of NBS should be to save lives and prevent rare diseases from having serious consequences—to detect rare diseases for which there is effective treatment as early as possible. This echoes the main criteria of the RUSP—urgent, severe, and treatable.

A number of participants of the listening session for rare disease patients, families, and advocacy groups, as well one-third of questionnaire respondents, indicated that NBS serves the broader purpose of supporting parents and providers to make informed decisions. Additional purposes mentioned were the need for equitable health outcomes and the importance of long-term follow-up care.

### Questionnaire

In the questionnaire, respondents were asked the open-ended question: *From your perspective, what should be the main purpose of newborn screening?*

The overwhelming majority of questionnaire respondents supported the common view that the main purpose of NBS should be **to identify serious diseases and conditions**. Respondents differed in whether screened conditions should have an available medical treatment, with some believing NBS should only be for conditions that can be treated, and with others believing screening is an essential information opportunity for all conditions.

About half of respondents emphasized that the purpose of NBS is to detect a rare disease or condition as early as possible, including one parent of a child with a rare disease who responded, "Detectar desde temprano cualquier enfermedad" [Detect any disease early]. The respondents described that this **allows for early and more effective intervention and treatment**. One NBS lab professional expressed this sentiment by saying that the purpose is for "early identification of heritable metabolic disorders in infants to provide timely intervention and treatment in an effort to prevent serious medical consequences or death."

About one-third of questionnaire respondents said NBS should also screen for diseases that do not have existing effective medical treatments. Many of these questionnaire respondents indicated that NBS should be done because it can support **benefits beyond treatment or prevention** of disease symptoms. These respondents described the importance of, in the words of a parent of a child with a rare disease, "information for parents to make the best possible choices for the health of their baby."

Although the emphasis on the treatability of screened-for diseases was much higher in the responses made by NBS professionals compared to the rest of the respondents, about one-fifth of NBS professionals indicated that they were sympathetic to a broader, more informative interpretation of NBS' purpose.

A small number of questionnaire respondents reflected on a more systems-oriented perspective, emphasizing that NBS enables long-term follow-up. A small number of others mentioned equitable NBS access as a purpose.

## Listening Sessions

At the start of the listening sessions, participants were prompted to use the Zoom chat to respond: *From your perspective, what should be the main purpose of newborn screening?*

Most participants suggested that the primary purpose of NBS is to detect rare diseases to facilitate intervention as early as possible—to **identify treatable conditions early**, enabling prompt treatments and supportive care that can lead to improved health outcomes and quality of life for infants. Many also noted that NBS should be considered "preventative medicine" in that early detection can potentially prevent disability and save lives. Rare disease patients, families, and advocacy groups emphasized the benefit of early or presymptomatic detection of conditions.

Rare disease participants also described NBS' purpose as **informing parents and providers**, and moreover, empowering parents with the necessary information to make decisions, even if treatments are not necessarily available.

Some participants also noted the need for **equitable health outcomes** to come out of NBS programs across the country. In the words of a participant from a mixed listening session, the purpose of NBS is "to ensure improved clinical outcomes for *all* children."

## STRENGTHS OF THE EXISTING NBS SYSTEM

When asked about the strengths of the existing NBS system, the majority of participants pointed to the fact that all babies are screened except for families who opt out. A number of all listening session participants also described follow-up with families after screening test results as a strength. Questionnaire respondents highlighted the financial and practical strengths of collecting bloodspots through heel sticks for NBS.

### Questionnaire

To highlight the strengths of the current NBS system, questionnaire respondents were asked: *Which parts of the current newborn screening systems are most effective at achieving the main purpose of newborn screening?* Figure 6 shows responses to this closed-ended question. The questionnaire also invited respondents to follow up with their rationale for their selection.

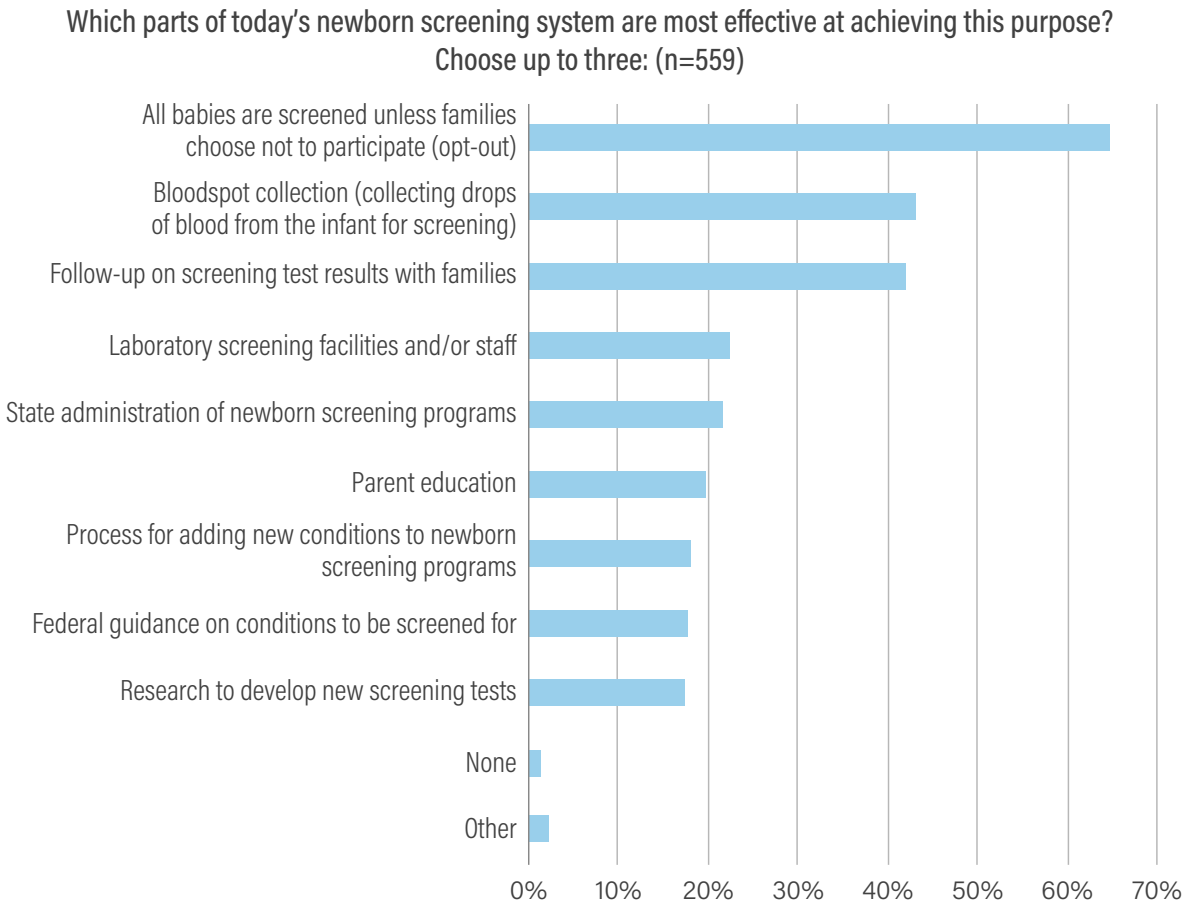



*Figure 6. Most effective parts of today's NBS according to questionnaire respondents. Respondents do not represent a statistical sampling of the American public.*

The majority of questionnaire respondents indicated that the most effective part of the NBS system is the public health goal of **screening all babies** unless families opt out. The widespread support for this approach highlights these respondents' acceptance of population-level mandated screening.

**Bloodspot collection**, the method of using heel sticks to collect drops of blood onto a special collection card (a Guthrie card) allows for ease of newborn blood collection and long-term storage. Over two-fifths of respondents selected dried bloodspot collection as a strength of NBS. One NBS follow-up professional qualified their answer in this regard by saying, "Collection of dried bloodspots is relatively easy to perform, which improves compliance." A rare disease advocate reflected, "There are very few alternatives that would allow NBS to take place without it." Many questionnaire respondents discussing bloodspots also pointed to the ease of storing and transporting these samples.

Two-fifth of respondents identified **follow-up on screening test results with families** as an NBS strength. Ensuring that families are promptly informed of concerning screening results and guided through subsequent steps is seen as a crucial strength of the NBS process. Many questionnaire respondents linked this follow-up aspect of NBS with the importance of enabling parents to be informed and supported with resources to navigate their child's diagnosis journey.

Several other NBS system components offered as answer options received moderate levels of selection among questionnaire respondents, including laboratory screening facilities and/or staff, state administration of NBS, parent education, the process for adding new conditions to the screening program, federal guidance on conditions for which to be screened, and research to develop new screening tests.

> **"Collection of dried bloodspots is relatively easy to perform, which improves compliance.**
>
> —Rare disease advocate

## Listening Sessions

In the listening sessions, participants were asked to briefly share their thoughts on what is working well about NBS in the country: *In your experience, what is working well about newborn screening in the US today?*

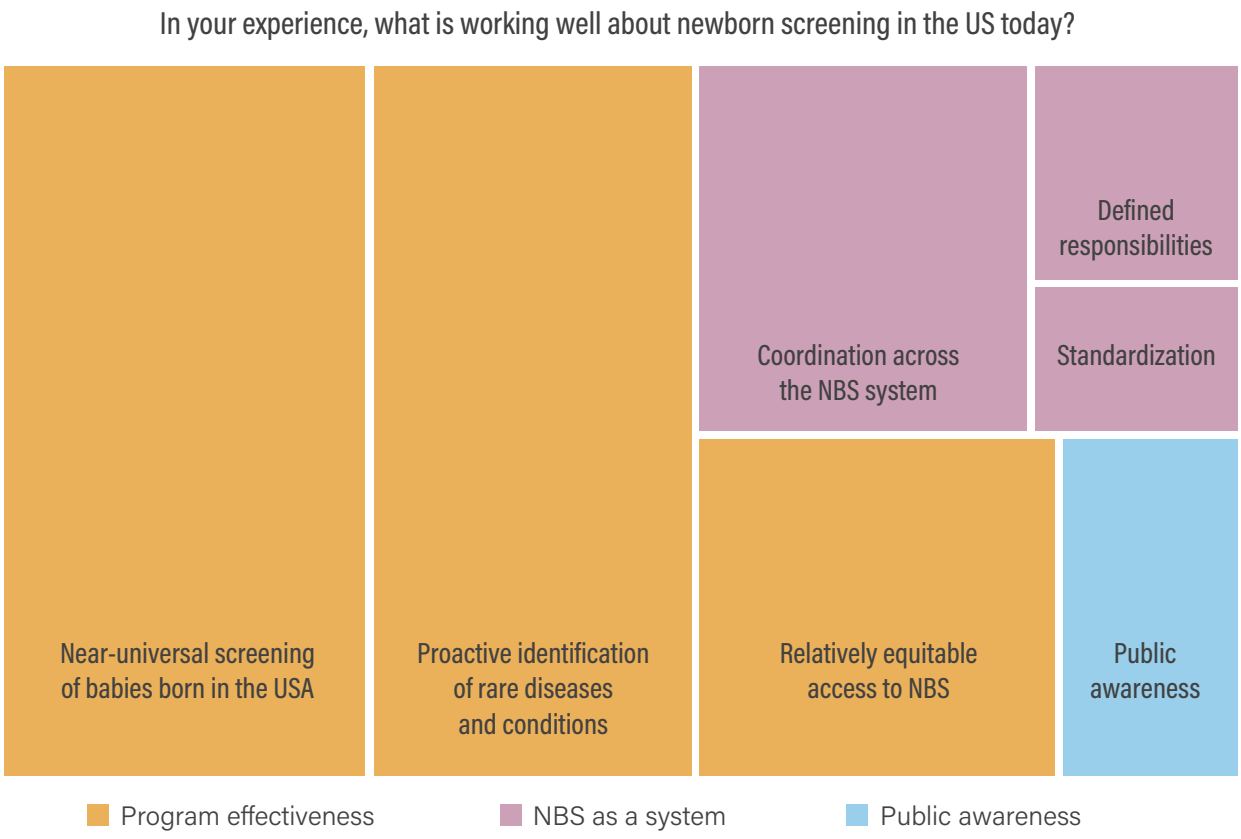



*Figure 7. Most effective parts of today's NBS system according to listening session participants. The box size corresponds with the frequency of comments on the subject. Participants do not represent a statistical sampling of the American public.*

Two-thirds of listening session participants discussed themes related to the effectiveness of NBS programs as a strength. Specifically, one-third of comments pointed to the fact that NBS in the United States has **near-universal coverage**—about 98% babies born each year in the United States are screened.[12] In addition, nearly one-third of comments noted that NBS leads to **early intervention** by identifying and treating babies with one of the diseases or conditions screened for by NBS as early as possible. A public health professional summarized this sentiment, saying, "Almost all infants are screened, and most conditions identified receive interventions and have good outcomes."

12 Centers for Disease Control and Prevention. (2012). *CDC Grand Rounds: Newborn screening and improved outcomes.* Accessed 20 June 2024. https://www.cdc.gov/mmwr/preview/mmwrhtml/mm6121a2.htm

Relatedly, a parent of a child with a rare disease talked about the **near-universal access**, saying, "Lo que funciona es que uno no tiene que pagar por esa prueba, no importa si tienes plan médico privado o plan médico del gobierno, es totalmente gratis" [what works is that you don't have to pay for that test, it doesn't matter if you have a private health plan or a government health plan, it is totally free].

Participants of one mixed listening session frequently mentioned **systemization** in their assessment of what is working well. Many of them reflected that cross-sectoral coordination is working well across the NBS system, including "from hospitals, providers, labs, follow-up and administration," as shared by a public health professional. Another participant from the healthcare industry added that clearly defined roles and responsibilities among NBS providers is a strength, as it "addresses equitable access, systematic coordination of cascade activities, and is funded by states."

> "Lo que funciona es que uno no tiene que pagar por esa prueba, no importa si tienes plan médico privado o plan médico del gobierno, es totalmente gratis.
>
> What works is that you don't have to pay for that test, it doesn't matter if you have a private health plan or a government health plan, it is totally free.
>
> —Parent of a child with a rare disease

## CHALLENGES OF THE NBS SYSTEM

Both questionnaire respondents and listening session participants were asked to share their views about the main challenges facing the current NBS system. The themes from their input are described immediately below and further explored in subsequent report sections.

### Questionnaire

In the questionnaire, participants were asked to select the top challenges of the existing NBS system in the United States, and then to describe their rationale. Almost half of respondents identified the process for **adding new conditions** to NBS programs as a challenge. Respondents described concerns regarding the complexity and efficiency of incorporating additional conditions into existing screening protocols.

Over two-fifths of respondents highlighted **parent education** as a challenge. These respondents called for better resources and strategies to inform and educate parents about NBS processes, benefits, and follow-up actions. Similarly, follow-up with families on test results is a challenge raised by some, with respondents describing gaps in communication and support provided to families after initial screening results.

**State administration o**f NBS programs was an additional issue identified by about one-quarter of respondents, who described challenges in the management and implementation of these programs at the state-level. **Federal guidance** on conditions to be screened for was a challenge identified by about one-quarter of respondents, suggesting a need for clearer and more consistent federal policies to guide state programs. Additionally, **research** to develop new screening tests was identified as a challenge by about one-quarter of participants, who underscored the importance of ongoing innovation and development in screening technologies and methodologies.

Laboratory screening facilities and staff was seen by some respondents as a key challenge, though to a lesser extent compared to the top concerns. Bloodspot collection and the policy of screening all babies unless families opt out were identified as challenges by a smaller proportion of respondents, indicating specific procedural and policy-related concerns.

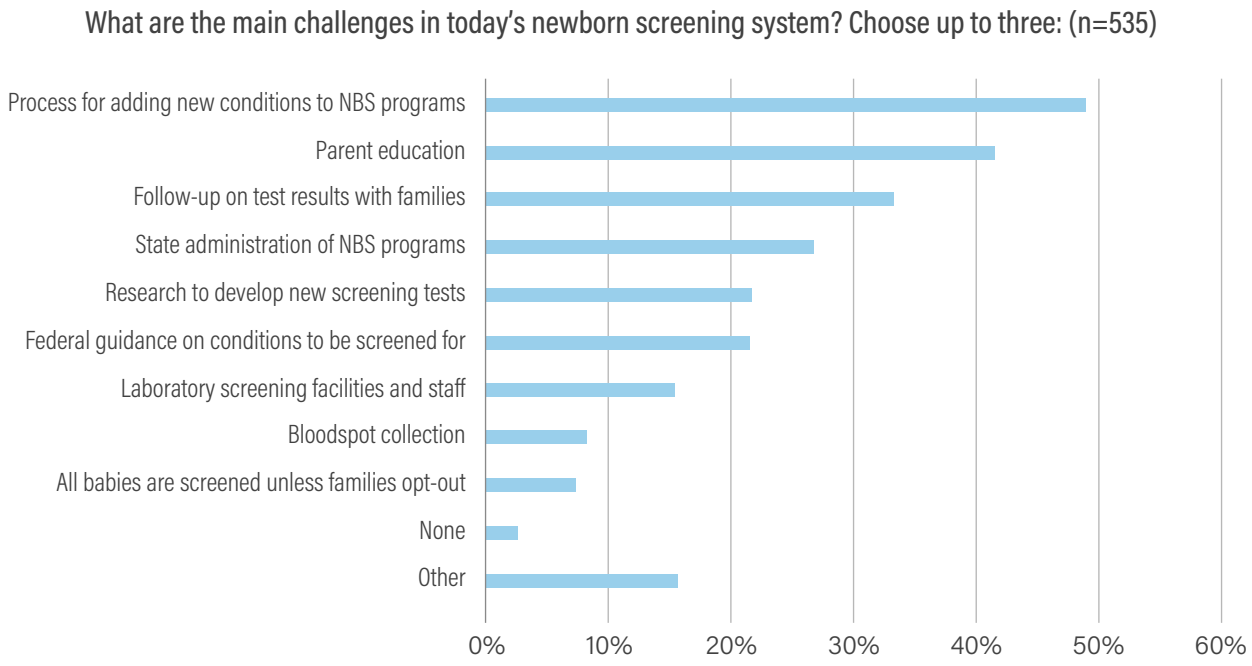



*Figure 8. Main challenges in today's NBS system according to questionnaire respondents. Respondents do not represent a statistical sampling of the American public.*

A total of 32 respondents chose "other" in response to this question, describing challenges in their own words. Funding was the top challenge identified in these responses, followed by a call for better healthcare provider training and educational initiatives for the public. Additional challenges raised include workforce challenges, healthcare access disparities, technology and infrastructure limitations, data access and utilization, timely follow-up and referrals, and operational difficulties in screening.

### Summary of Challenges Explained

Following the question above, questionnaire respondents were asked an open-ended question: *Looking at what you chose in the question about main challenges in today's NBS screening system, why are these part(s) a challenge?* After analyzing the responses, the following themes emerged:

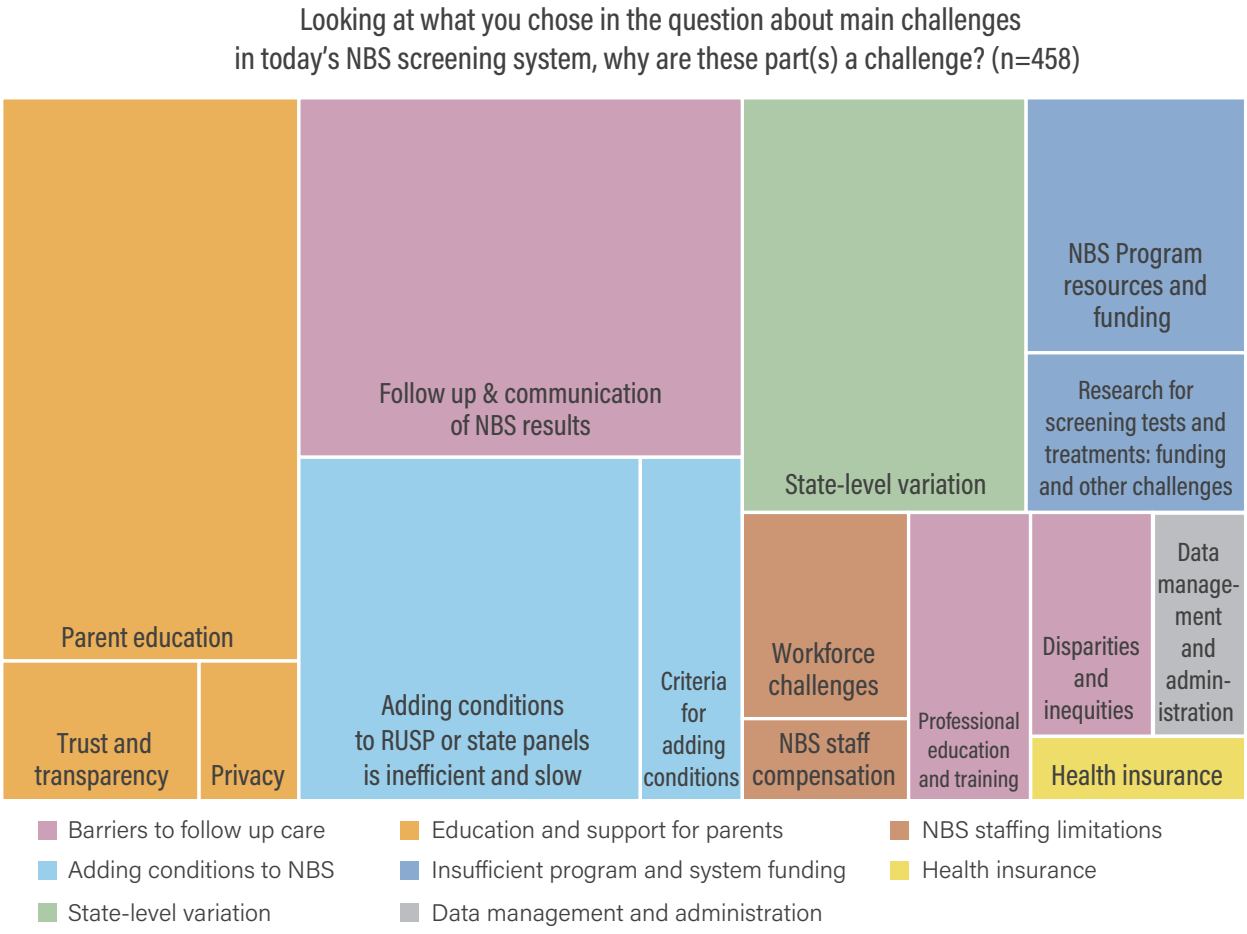



*Figure 9. Questionnaire themes from open ended answers about NBS challenges according to respondents. The box size corresponds with the frequency of comments on the subject. Respondents do not represent a statistical sampling of the American public.*

## Listening Sessions

Listening session participants were asked to identify challenges they see or experience with NBS. The NBS challenges identified by listening session participants differ in some ways from questionnaire respondents. **Barriers to follow-up care**, especially in terms of the communication of NBS results, was one of the more frequently identified challenges. The next most frequently discussed challenges were **parent education and support**, followed by the challenges of **state-level variation** and **adding conditions** to NBS programs. Some of the listening sessions also generated

discussions about **data management and administrative inefficiencies**. The challenge of **follow-up care**, although frequently discussed in several listening sessions, was not as prominently featured in discussions across all the listening sessions. This is in comparison to its importance among questionnaire respondents, selected as one their top three choices for main challenges.

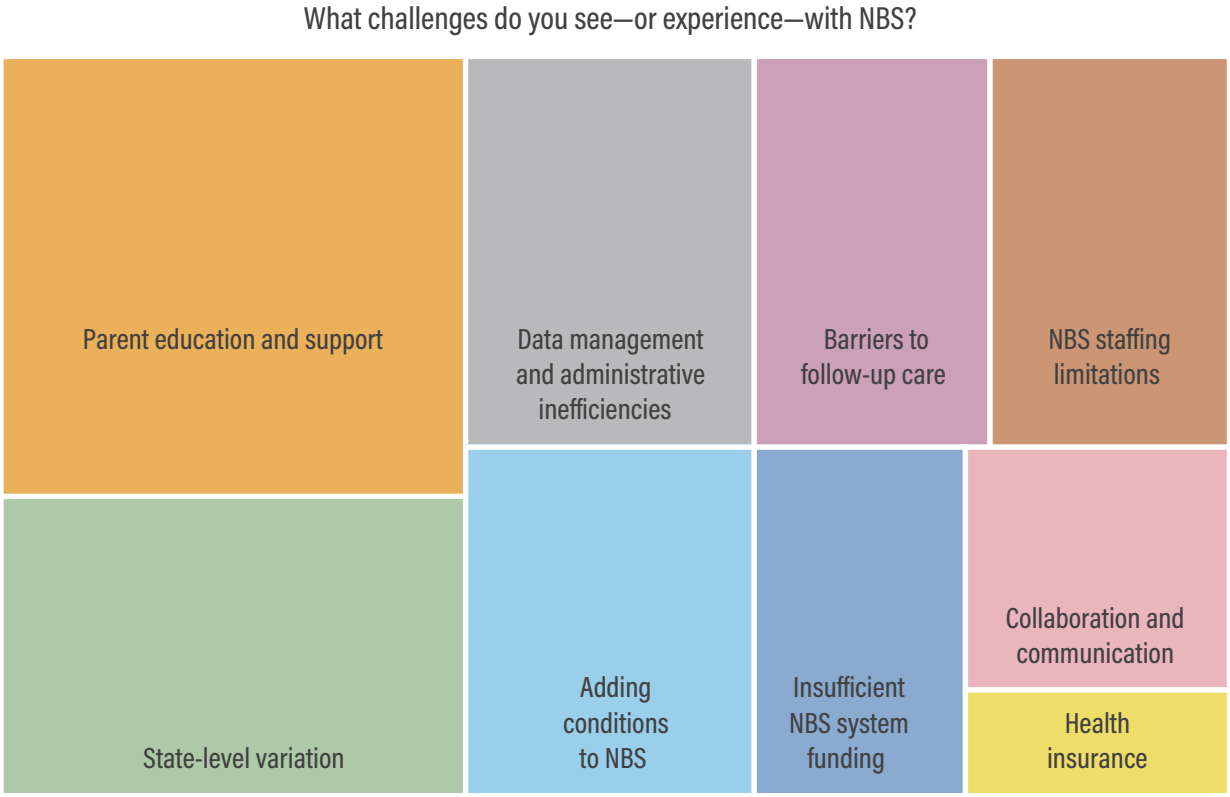



*Figure 10. Listening session themes from discussion about NBS challenges. The box size corresponds with the frequency of comments on the subject. Participants do not represent a statistical sampling of the American public.*

## ADDING CONDITIONS TO NBS PROGRAMS

Participants shared 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 RUSP for adding new conditions. The current RUSP emphasizes conditions that are urgent, severe, and treatable.[13] Some respondents indicated that these criteria are far too restrictive, potentially resulting in poor **health outcomes** for babies who have rare diseases that are not screened by NBS. A major tension with this view, however, is the perspective from other participants that NBS needs to be based on sound evidence, particularly in the **availability of treatment** for the condition and the **accuracy of the screening methods** available at the time. These discussions often touched on

13 Health Resources & Services Administration. (n.d.) *Recommended Uniform Screening Panel*. Federal Advisory Committees. Accessed July 19, 2024 at https://www.hrsa.gov/advisory-committees/heritable-disorders/rusp

ideas about engaging more with parents and advocacy groups and developing research incentives for generating the necessary evidence involved in adding new conditions. Other discussion points were also raised around genomic sequencing, and on considering the incidence, onset time, and prevalence of a condition.

Many respondents and participants also pointed out that even though the RUSP recommends diseases for states to consider adding to NBS programs, **approvals of new conditions vary** state-by-state. Participants with a professional NBS affiliation often connected this with variations in the healthcare workforce and the NBS infrastructure of each state. Discussions on this theme often touched on ideas around federal funding for state NBS programs and the uniqueness of each state's challenges. This topic is also further explored in the section on state-level variation.

## Questionnaire

The leading challenge identified by questionnaire respondents is **adding conditions** to NBS programs. Addressing the challenge is a priority for many families with rare diseases, as one parent of a child with a rare disease put it simply: "Que pueden incluir más enfermedades a la lista para detectar en bebés" [So they can include more diseases in the panel to detect these conditions in babies].

Open-ended responses show a mixed level of comfort with the existing **RUSP criteria**. For example, in the words of a parent of a child with a rare disease: "The criteria that states the condition must currently be treatable does not account for the impact that has on families trying to get their kids into clinical trials." Others shared a different view; one health researcher expressed, "If there is no treatment, we should not be using screening in this manner." Another health researcher remarked that the "addition of new conditions may be political rather than scientific."

> **"The process for adding new conditions is cumbersome, onerous, and takes too much time and effort on the part of patient advocates who are already spread thin.**
>
> —Parent of a child with a rare disease

Navigating **state approval processes** across the country to add new conditions to many states' NBS programs was identified by respondents as a key challenge. "A valid NBS test and valid treatment may exist for a disease," said a parent of a child with a rare disease, but "it takes the better part of a decade to get it done, and taxes the advocacy organizations who can least afford to spend their valuable time and resources on this."

Adding conditions to the RUSP or to state NBS panels is **inefficient and slow**, according to many questionnaire respondents. "The process for adding new conditions is cumbersome, onerous, and takes too much time and effort on the part of patient advocates who are already spread thin," said a parent of a child with a rare disease. Another parent of a child with a rare disease pointed out, "Advocates must repeat the process 50 times, and no state operates the same way. Children are dying during this delay."

A repeated recommendation was for sufficient **federal funding for states** so that all states have the ability to add a condition within 3 years of being added to the RUSP. A number of respondents described that the state-level variations in screening panels are, in the words of a health researcher, "exacerbated by the chronic underfunding of state public health programs, meaning it takes much longer for states to add new conditions to their NBS panels because they **lack the resources** to hire new staff or buy new equipment."

Echoing this, other respondents pointed out that, even if a state mandates adding a new condition to its NBS panel, state laboratories are often not ready to add more conditions without support. "It can sometimes make things more difficult for state laboratories if new conditions are added before the lab is ready or if … they have to put their priorities on the back burner in order to implement legislatively mandated additions," said a public health professional. Respondents identified a need for **financial, staffing, space, and logistical support for state labs** that have, as one NBS lab professional said, "limited resources and daily responsibilities of screening and following thousands of newborns."

## Listening Sessions

In each listening session, participants were asked to specifically discuss: *What do you think should guide whether a disease is added to the newborn screening program?*

A number of **inefficiencies in adding conditions** to the RUSP were raised. An overarching concern from most participants was that the nomination process is subjective and takes a substantial amount of time and resources to accomplish—e.g., requiring extensive packages of evidence to be collected and reviewed. Others noted that the number of studies that may result in additional conditions meeting RUSP criteria has the potential to overwhelm the NBS system. Some participants in each listening session also reflected that it should not be the **burden of parents** to be the main driver in nominating a disease and bringing it into implementation.

Some responses varied among NBS professionals and the participants who attended the listening sessions as rare disease patients, families, and advocacy organizations. The latter group were more aligned with the idea of adding diseases to the RUSP if there are reliable, **accurate screening methods** with low false positive rates. NBS professionals raised more frequently the need to consider if approved and **evidence-based treatments** were available.

Rare disease patients, families, and advocacy organizations generally expressed less interest in screening accuracy and the availability or potential of medical treatments. Rare disease families and advocates more frequently suggested that the **RUSP criteria is too exclusive**. "If you can identify the disease you should screen for it," said one participant from the listening session for that group.

Others from the listening session for the rare disease community also emphasized that the **availability of treatment** or treatment potential should not be a determinant for NBS inclusion, as there can be alternative therapies or other benefits that can come from parents being empowered by receiving the initial screening results. Participants of the session convened with the assistance of The Akari Foundation encouraged, "Si hay enfermedades que no tienen tratamiento, también es bueno saber, porque de ahí puede nacer el interés por generar estas nuevas terapias o estos nuevos tratamientos para estas enfermedades, "[If there are diseases that have no treatment, it is also good to know, because from there the interest in generating these new therapies or these new treatments for these diseases can arise].

Participants from rare disease patients, families, and advocacy organizations also raised the **research incentives** that could come with adding a disease to the RUSP regardless of current treatment options. Many recognized that industry involvement could be incentivized this way, including pharmaceutical companies committed to developing therapies. Ideas put forward included having trial screening periods, prioritizing diseases with ongoing clinical trials, and leveraging data from screening to advance drug development. Some participants said that screening for diseases, even those without treatments, would open the door to companies developing treatments, as "in order for treatments to be developed, there needs to be patients identified for trials."

Those who attended one of the mixed listening sessions often indicated that adding a new disease required careful **consideration of the systems built around NBS programs**, from the general increase in demands to the healthcare workforce and NBS infrastructure to the **level of readiness of each state** for implementing the new screening required. Even with this in mind, a parent of a child with a rare disease asked, "Why do we have to reinvent the RUSP approval by doing it again at the state-level?" A member of the rare disease community suggested that all states should follow the Alabama three-year timeline to add diseases to the state screening program once they have been added to the RUSP. On the other hand, healthcare providers who participated expressed more interest in discussing the degree to which **referral and follow-up systems** are in place in each state.

Related to follow-up, apart from the listening session for NBS professionals, most listening sessions also touched on the need to **engage with parents and advocacy groups** when considering adding new diseases and conditions to the RUSP. One parent summarized the motivation behind their position on why engaging with parents is critical in this manner: "Whether treatment or no treatment, simply having information gives family or caregivers the knowledge that truly matters to plan financials, family support, clinical support, and insurance decisions."

> Whether treatment or no treatment, simply having information gives family or caregivers the knowledge that truly matters to plan financials, family support, clinical support, and insurance decisions.
>
> —Parent participant

Consideration of the disease itself was also raised during the listening session discussions. Participating NBS professionals raised the need to consider the **incidence and prevalence of a disease** as elements for whether to add it to the RUSP. Meanwhile, participants of the listening session for rare disease patients, families, and advocacy organizations were more keen on discussing the need to consider the **onset time of a disease**. A few participants across different listening sessions also suggested the concept of a two-tiered approach, based on both the level of severity and onset time.

Participants in each listening session also explored the promise and challenges of genomics in NBS. Sequencing could offer more clarity to families, some suggested. A key concern was **equitable access to genomic sequencing**, given the variability in both state capacity and family resources for private testing. Additional concerns raised include the need to ensure care for the anticipated increase in babies with positive screen results, the complexities due to variants of unknown significance, and the longer turnaround time for sequencing results.

Finally, while a few participants raised the need to consider the financial impact or burden on NBS programs when considering whether to add a disease to the RUSP, an NBS professional made a point that resonated well in their listening session: "Cost is not a consideration for ACHDNC. Can we not take care of our children? Cost is another system's problem to solve."

## PARENT EDUCATION AND SUPPORT

Many questionnaire respondents and listening session participants strongly emphasized the importance of supporting parents in understanding and navigating NBS. Many highlighted that parents are rarely educated about the NBS system until they receive a positive result, and they called for earlier and more robust parent education. Participants added that the need for more communication with parents is also driven by growing distrust in government programs and apprehension about genetic information sharing. Some called for more nuanced privacy protections and policies for the transparent storage and subsequent reuse of NBS samples and data.

The need to address racial, socioeconomic, and geographic inequities was also prominent in the responses. Participants flagged that test accuracy can be affected by the newborn's ancestry. There are racial and socio-economic disparities in access to follow-up and specialty care, with geography posing a further challenge for many rural families, particularly those with lower incomes. In addition, some participants mentioned an unfair advocacy burden on parents and rare disease groups and highlighted the financial and emotional burdens of a diagnostic odyssey.

## Questionnaire

One-quarter of questionnaire responses about the challenges of NBS pointed to the need for parent education. "Childbirth is a whirlwind experience," said one healthcare provider, "many parents are overwhelmed and often are **not aware this test is even performed** until results are shared. Thus, education must occur during **prenatal care** and reinforced to ensure families are making an informed decision."

Of those who identified as parents in the questionnaire, 88% said their child received NBS, 9% said they did not, and 4% were unsure. Recent data indicate that many parents may not be aware their child received NBS, given that over 98% of United States babies born annually are screened.[14]

Respondents also noted that, in the words of a person with a rare disease, "parents who may be **experiencing unmet social needs**, have lower literacy or health literacy, have lower incomes or education, may not be as prepared to absorb education around screenings." Therefore, education must be appropriate to all types of parents.

The key message about education from many respondents is captured by this healthcare provider's statement: "Parents are hardly ever educated about NBS until they receive a positive result." The need for a proactive approach to NBS education is particularly important given the **rise in misinformation** on social media, as suggested by a number of respondents.

> "Childbirth is a whirlwind experience, many parents are overwhelmed and often are not aware this test is even performed until results are shared.
>
> —Healthcare provider

Increasing mistrust was another theme found in participants' descriptions of the challenges of NBS. "There's a **lack of trust** by many about any government programs. Add to this the apprehension about genetic information being shared," said one healthcare provider. Better and earlier parent education was suggested as an opportunity to build trust in NBS among parents and the community. Another suggestion was to develop more **robust and explicit privacy protections** for NBS samples and data.

Although the storage and use of residual newborn bloodspots were seen by a number of respondents to "have amazing potential for equitable public health research," one health researcher warned that "the failure to protect bloodspots from misuse in criminal investigations and the lack of **transparency** in storing and using these bloodspots severely damages public trust." Respondents called for **nuanced policy** to address privacy concerns.

14 Centers for Disease Control and Prevention. (2012). *CDC Grand Rounds: Newborn screening and improved outcomes*. Accessed 20 June 2024. https://www.cdc.gov/mmwr/preview/mmwrhtml/mm6121a2.htm

> The failure to protect bloodspots from misuse in criminal investigations and the lack of transparency in storing and using these bloodspots severely damages public trust.
>
> —Health researcher

## Listening Sessions

**A lack of parent education and support** was a key challenge raised in all the listening sessions. Multiple participants said that parents are insufficiently educated about NBS, before, during, and after NBS takes place. "I think I do remember them taking blood from my second baby," said one mother, "but that was it. They never came back and never reported the results."

Many participants recommended prenatal education, as parents have a lot going on once the baby is born. Others recommended popular education like using social media campaigns or partnerships with search engines to make sure that reliable and accurate educational resources are appearing at the top of web searches. One participant from the session convened with the assistance of REACHUP, Inc., who learned about NBS through the listening session, had a different view and said that there is already enough information for new parents: "I kind of like that they don't approach you with anything if there's nothing to be worried about."

**Racial and socioeconomic inequities** were a concern also raised in all the listening sessions. Several participants pointed to the lower accuracy of the test results for people of non-European ancestry as well as disparities in follow-up timing based on racial differences. In terms of follow-up, "So much depends on the resources of the parents," a parent of a child with a rare disease pointed out, giving an example: "If you are lower-income, you may not be able to travel for treatment." Another issue raised was language barriers. A public health professional pointed out that "the healthcare system in general is not well suited for a diverse language demographic."

> I kind of like that they don't approach you with anything if there's nothing to be worried about.
>
> —Listening session participant

Some listening session participants noted that there is an **unfair advocacy burden** on parents and rare disease groups. "If there is no well-organized and developed advocacy group with the opportunity to develop resources and infrastructure, it causes disparity," said a participant in the listening session for rare disease families and advocacy groups. Participants in this session also noted that there have been important changes due to the work of parents and advocacy groups. "There's nothing we can do until we change the regulations, so there I went. Now we [our state] have actually one of the strongest programs in the nation," said a parent of a child with a rare disease.

For many participants, **public trust and transparency** are essential for NBS. "People ask more questions now and are not believing the answers," one pediatrician said, underscoring the need to "reestablish trust with the general public and this generation of parents." Relatedly, some participants noted that there appears to be an increase in the number of people opting out of NBS or expressing interest in doing so.

The financial and emotional burden of a **diagnostic odyssey** is a challenge for NBS as a whole, said a few participants in each listening session. This issue also has an equity element, and a participant in the listening sessions for rare disease families and advocacy groups noted that the "diagnostic odyssey can cost +$100,000 and about 42% of children are born onto Medicaid. It is highly unlikely that they will be able to pursue a diagnosis or pay out of pocket for services."

A solution suggested by some listening session participants is to see **parents as partners** in the process of diagnosis and care. Having a child with a rare disease is difficult for parents, who "need emotional support and mental healthcare to help the family process what's going on," said a participant of the rare disease families and advocacy groups listening session. **Connecting parents** going through similar experiences was recommended during the listening sessions by a number of parents of children with rare diseases.

> **"If there is no well-organized and developed advocacy group with the opportunity to develop resources and infrastructure, it causes disparity.**
>
> —Participant in the listening session for rare disease families and advocacy groups

## Support for Infants and Families

Questionnaire respondents were asked: *Which three of the following elements are most important to offer every infant and their parent/guardian?* Respondents had the option to select up to three items from the provided list.

Two-thirds of respondents chose the option of **timely follow-up** on screening results for the purpose of additional testing to confirm a diagnosis. Many respondents also chose connection to **pediatricians and specialist care**, if applicable; access to **affordable** treatment; and **screening** for all urgent conditions that have a medical treatment.

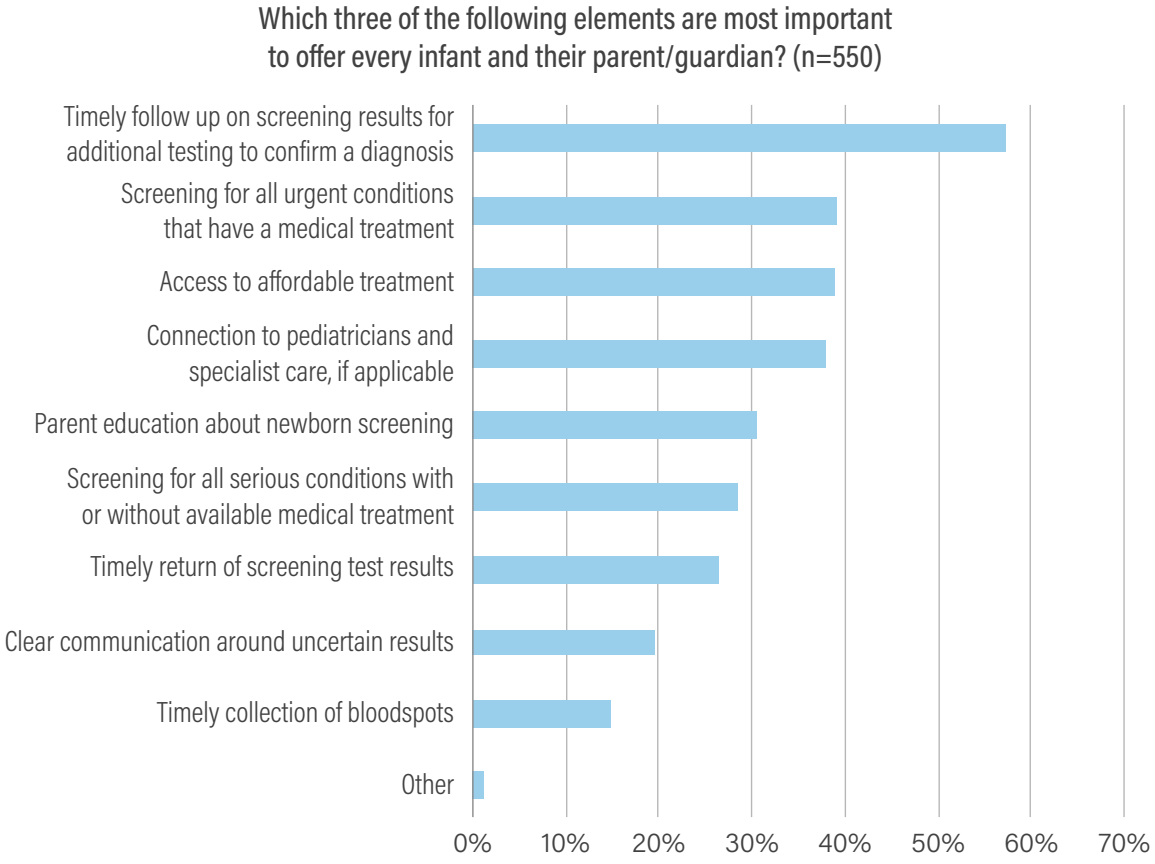



*Figure 11. Elements most important to offer every infant and their parent/guardian according to questionnaire respondents. Respondents do not represent a statistical sampling of the American public.*

## The Needs of Rare Disease Families

Those questionnaire respondents who identified as connected to the rare disease community—as parents, persons with a rare disease, or advocates—were asked: *What is your rare disease community's most important need regarding being able to identify and treat people affected by rare disease, and how do you think newborn screening could contribute to meeting this need?* Response frequencies for the following themes were fairly consistent across the three rare disease respondent groups.

What is your rare disease community's most important need regarding being able to identify and treat people affected by rare disease, and how do you think newborn screening could contribute to meeting this need? (n=171)

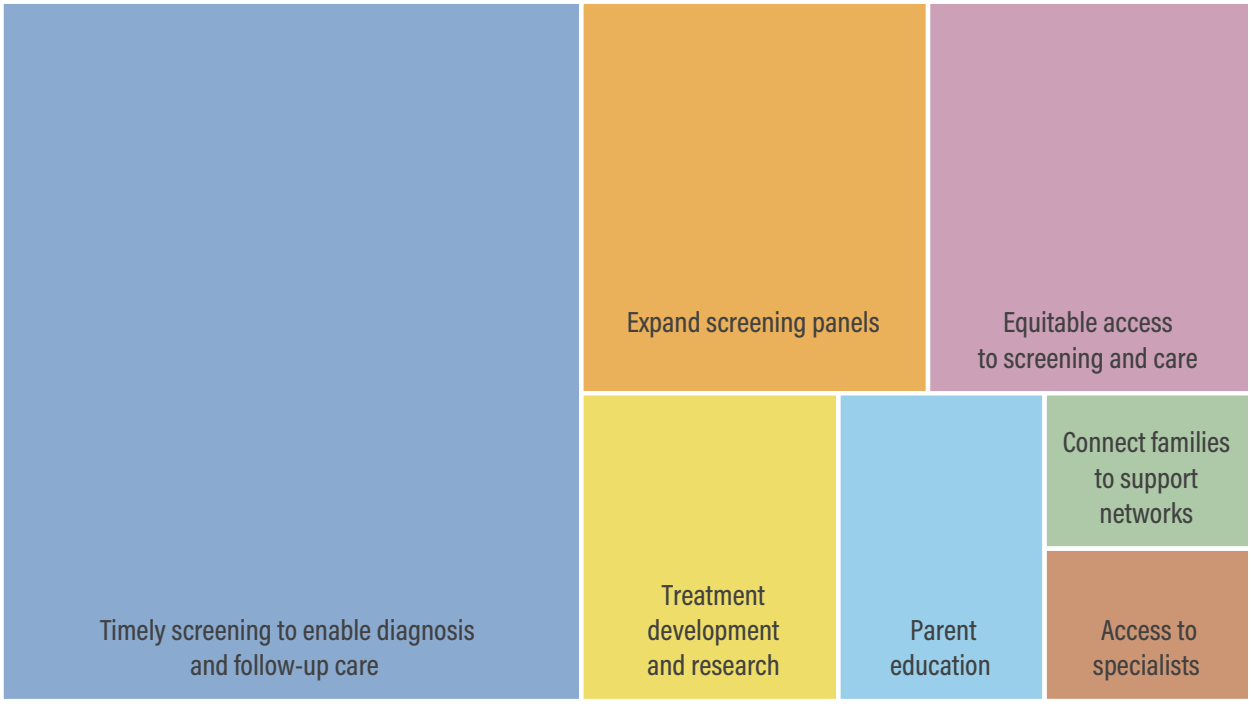



*Figure 12. Important needs of the rare disease community and the related role of NBS according to questionnaire respondents. The box size corresponds with the frequency of comments on the subject. Respondents do not represent a statistical sampling of the American public.*

Half of respondents with connections to the rare disease community underscored the importance of **timely screening to enable diagnosis and follow-up care**. As described by a person with a rare disease or condition, "Knowledge is power. Early diagnosis of a rare disease will enable families to avoid diagnostic odysseys, to obtain treatment or supportive care as early as possible, and to engage in informed future family planning."

**Equitable access to screening and care** was highlighted as a need in one-sixth of the responses to this question. "Healthcare in the USA is not fair or just and is very much affected by location, race and socioeconomic status," described a healthcare provider. A number of other responses

highlighted the importance of affordable care and treatments. The other focus of responses to this question was on state parity in testing. In the words of a parent of child with a rare disease or condition, "The most important need is to make all children born in [the] US [get] screening for all diseases or conditions no matter their state."

The need for treatment development and research, expanded NBS screening panels, and parent education were featured in about one-tenth of the responses. A parent of a child with a rare disease said, "The most important need of the rare disease community is **more effective treatments** for more diseases … Development of new treatment or more effective treatment of any rare disease depends on identifying patients early enough, ideally presymptomatic, so that the treatment benefits can be better demonstrated."

The call to **expand the conditions screened for** by NBS focused on the addition of specific conditions, as well as comments related to state-level variation in NBS.

Responses calling for **more parent education** focused on helping parents prepare for the process of follow-up testing. A rare disease advocate described the need for "education that a healthy-looking baby might have a rare disease and needs to get followed up for confirmatory testing."

Others called for more **healthcare provider education**, including one parent of a child with a rare disease, who said, "Que puedan educarse todos los profesionales de la salud sobre distintas enfermedades para que puedan detectar a tiempo una enfermedad o afección" [That all health professionals can be educated about different diseases so that they can detect a disease or condition in time].

A smaller number of responses identified the importance of **connecting families to support networks** or **connecting patients to specialists** to support diagnosis and care.

> **"Development of new treatment or more effective treatment of any rare disease depends on identifying patients early enough, ideally presymptomatic, so that the treatment benefits can be better demonstrated.**
>
> —Parent of a child with a rare disease

## STATE-LEVEL VARIATION

The challenge of NBS program variation between states was noted by many participants. Although the RUSP recommends conditions for screening, the state-run NBS programs are ultimately responsible for adopting and implementing NBS and there are differences between each state's system. Accordingly, many participants took issue with the disparities between states in terms of the set of conditions screened. Nearly half of questionnaire respondents emphasized that having national standards would lead to more equitable health outcomes. A potential solution to this issue, some pointed out, was the regionalization of NBS programs and increased inter-state collaboration for follow-up care.

Others, however, pointed out that state-level variation comes from the fact that states differ in their demographics, and their readiness and capacity regarding staffing and NBS infrastructure. For some, the issue was more noticeable among states with large rural-urban divides.

### Questionnaire

About one-fifth of respondents identified state-level variability as a key NBS challenge. They described that because conditions are not uniformly screened from state to state, **disparities differ based on where a baby is born**. "Death by zip code" was a term used by nineteen respondents. "Luck or happenstance shouldn't be a factor in determining if a child has a rare disease not immediately apparent at birth," said a respondent. One healthcare provider reflected, "I work in a border area with 2 other states, it is very disheartening to see kids whose diagnosis was missed just because they were born 20 min over the state line. Quite tragic."

> "I work in a border area with 2 other states, it is very disheartening to see kids whose diagnosis was missed just because they were born 20 min. over the state line.
>
> —Healthcare provider

One healthcare provider argued, "Each state is testing differently and that needs to be changed." A parent of child with a rare disease or condition supported this perspective, saying, "All states should test for ALL available conditions. It should be **mandated federally**." Relatedly, a rare disease advocate said, "Having each state responsible for their own NBS causes **inequality and delays**."

#### Roles of state and federal governments in NBS

The questionnaire also asked a closed-ended question about state-level variation: *The federal government recommends (but does not require) the conditions that should be included in newborn screening. States and territories choose which conditions to include in their program to address the needs of their populations and comply with other factors including legislative requirements, budget, workforce availability, and technological resources. Which statement best reflects your perspective?*

Respondents were asked to choose one of two closed-ended options, or "other". About three-quarters of respondents called for "every newborn in the United States and territories should receive the same newborn screening tests." Under one-fifth selected the statement: "Each state/territory should be able to choose the tests to include in their newborn screening." One-tenth selected "other".

This question about the roles of state and federal governments in NBS was followed up with the inquiry: *Why did you choose this answer?* Approximately three-quarters of questionnaire respondents described that they believe **national standards** are important, with nearly half of respondents also emphasizing that having national standards would lead to more equitable health outcomes. One parent, for instance, said, "The availability of a test for a disease should not be subject to luck of residing in a state that tests for it." A healthcare provider summarized their reasoning in relation to the tension between federal recommendations and state adaptability by saying, "While understandably each region may have higher prevalence of certain conditions than others, all individuals deserve to have the same access across the country to testing for all of the available conditions that can be detected on NBS."

Just under one-fifth of questionnaire respondents indicated that they believed **states should have autonomy** to decide which diseases to include in their NBS programs to address the needs of their populations and comply with other local factors and context. The group that most identified with state adaptability were public health professionals, with nearly half of these respondents suggesting that states need to be able to adapt their NBS panels for their population's needs. About one-third of NBS lab professionals, follow-up professionals, and health researchers also shared this kind of thinking. Only a few respondents gave other reasons, which were more about state rights or sovereignty.

## Listening Sessions

One of the most prominent themes to come out of discussions across all listening sessions was the **variability among NBS programs** across the United States. One participant of the listening session for NBS professionals, for example, reflected that in their region "all the medical facilities are in one state and people go get tested in that state." A particular phrase, **"death by zip code,"** was heard nine times across the various listening sessions, except for the session for NBS professionals. This phrase represents the way some participants saw the issue of unequal implementation of NBS across the country—that access to NBS testing and follow-up is dependent upon the state in which the baby is born. In addition to the differences in state NBS panels, participants noted that in areas with fewer resources, there are challenges in accessing or following up on NBS. Healthcare providers noted that the differing NBS standards for screening and specialty consultations in follow-up care between states can make it difficult to collaborate across state lines.

Most participants of the listening session for NBS professionals attributed state-level variations to the **rural-urban divide**. For these NBS professionals, state-level variation is a geographic issue within each state due to a lack of health centers in rural areas. One NBS professional reflected that the challenge is simply that "rural states [have to] minimize gaps for rural areas."

A number of participants across all listening sessions called for NBS to be standardized, so there would be **more uniformity across states** in what conditions are being screened for. "Standardization on the national level would be a great step," said one rare disease participant. A health industry representative suggested that "the RUSP should be an USP (uniform screening panel). Drop 'recommended."

Another suggested solution was the **regionalization** of NBS programs, where states can pool capacity and resources, look beyond state boundaries to help connect individuals with testing and specialists, coordinate more intensively, or operate with increased federal guidance. "Regionalization can also reduce overall cost and make screening more equitable across the country," recommended an NBS professional.

## BARRIERS TO FOLLOW-UP CARE

Participants emphasized the importance of NBS follow-up, with many highlighting that there are key barriers preventing timely intervention and support for patients, parents, and families. Some of the barriers include geographic challenges, logistical and capacity issues, as well as a lack of health insurance and issues in communicating with payors.

Note that participants may have used the term 'follow-up' to describe a number of considerations, including follow-up from an NBS screening result (e.g. confirmatory testing leading to a diagnosis); follow-up involving primary care and specialists; and, longer-term specialty follow-up of monitoring and care to support a child and their family. Where the participants' intended use is clear, the narrative below endeavors to clarify the term's meaning.

### Questionnaire

About one-third of questionnaire respondents flagged barriers to follow-up after a screening result as a key challenge for NBS. In the words of one healthcare provider, "follow-up is the most important interface with the families as this is highly sensitive and urgent." Yet, "there are significant gaps in follow-up," described a health researcher, echoing the voice of many respondents.

**Inequities** were a major gap in follow-up testing and care. "follow-up is where most babies get lost," said one healthcare provider, "parents may have no close facilities to do the testing or lack transportation, language skills, or health insurance to get the testing done." Many respondents noted that in **underserved areas** families have to deal with many more logistical issues and lack of access to providers. "While infants are generally screened universally," noted a healthcare provider, "the system for follow-up and treatment for screen positive infants **mirrors our healthcare system**. [Some] Infants are lost to follow-up or don't receive treatment in a timely manner."

**Communicating NBS results** and their implications was a key challenge identified in the questionnaire results. One specific challenge described, is in the words of a pediatric subspecialist, "Because NBS is a state-run program, there are no consistent policies for who is responsible for follow-up of abnormal NBS tests." Many also called for a **standardized follow-up process** to diagnose screening results that are abnormal and need confirmation, one that can be replicated in all states to avoid cases being lost to follow-up. "The notification of families should be standardized and all families should be notified of the results," said a public health professional.

Professional **education and training** are critical for NBS follow-up success, described a number of respondents. Healthcare providers are not always well informed about the rare diseases covered by NBS, said some questionnaire respondents. This can result, in the words of a healthcare provider, in "delayed diagnosis due to **[primary care physician's] lack of understanding of how to interpret results** and refer appropriately." NBS **specimen quality** is a problem described by some respondents from a range of sectors, who recommend better education and training for those involved in NBS. "Incorrect collection technique can delay results," said a healthcare provider. "Laboratory tests have to be accurate, facilities have to be up to date," a person with a rare disease argued.

Follow-up capacity is hampered by **insufficient funding and personnel**, described respondents. Follow-up, said one healthcare provider, "is dependent on the number of staff available for performing follow-up and … it is separately funded from laboratory testing, and prone to budget cuts by politicians." Others pointed out that follow-up testing and diagnosis following an abnormal NBS result and the communication with families can be a challenge for a busy primary care provider who has many other duties.

A number of respondents recommended responding to these follow-up challenges by seeing the **families of patients as partners**, and that family "perspectives and preferences be built into the system as partners, and not as subjects or just patients who receive instructions," said a public health professional.

> **"Follow-up is where most babies get lost. Parents may have no close facilities to do the testing or lack transportation, language skills, or health insurance to get the testing done.**
>
> —Healthcare provider

### Listening Sessions

A number of listening session participants noted that, for those who screen positive, a key barrier to follow-up care is having the **financial ability** and **insurance coverage** for accessing critical intervention therapies. A health insurance analytics expert noted that "28% of patients have a break in coverage during perinatal period, leading to follow-up challenges."

Many families, these participants said, also experience **delays** in seeing a specialist or slow physician response time to parent calls. "Timely follow-up and treatment for those with abnormal screening results," is a challenge identified by a participant of the rare disease patients, families and advocacy organizations listening session.

Barriers to follow-up care are exacerbated by **inequities**. In addition to spoken **language barriers**, as discussed above, some listening session participants also noted that there may be limited translated resources for various rare diseases and disorders. "There are also many languages that are spoken in the state [of Texas] and the medical field is not equipped to manage conversations beyond English," said a participant in the NBS professionals session.

Listening session participants also pointed out the challenge for some parents in accessing specialty care is **geographic distance**. Many noted the challenge of carrying out NBS follow-up for rural areas in their states. States with more rural and spread-out settlement patterns see this as a particularly critical challenge. Moreover, many NBS follow-up professionals also noted that state borders can confuse parents who travel out-of-state to give birth, as they may be unaware of the differences in NBS programs between the state where they gave birth and their home state, leading to logistical issues in follow-up.

**State disparities** in the number of available medical specialists or equipment are also barriers to accessing follow-up diagnostics and care. "The presence of children's specialists tends to be clustered around children's hospitals," noted a participant working at Medicaid. "If there isn't funding to add conditions," said a participant of the NBS professionals listening session, "then a baby born across a state border will not have access to the same screening as their neighboring state births. Financially, families struggle to travel long distances to reach specialty care."

## Health insurance

The matter of health insurance in the United States was a barrier to effective NBS follow-up according to participants of both the questionnaire and listening sessions.

### Questionnaire

Most questionnaire respondents who pointed to the challenge of health insurance underlined that the lack of health insurance is a barrier both to diagnostic testing and follow-up care. "Definitive testing is **dependent on insurance and resources**, both of which are not available to every child," said a healthcare provider.

If a disease is confirmed, "treatments for some of these conditions are **exorbitantly expensive**, not widely available, and insurance coverage can be a challenge," pointed out a genetic counsellor, representing a concern of other respondents.

> "Definitive testing is dependent on insurance and resources, both of which are not available to every child.
>
> —Healthcare provider

### Listening Sessions

Several listening session participants observed that few rare diseases have an associated ICD (International Classification of Diseases) code, which promote international uniformity in the collection, classification, processing, and presentation of health statistics.[15] Those in the listening session for rare disease families/advocates and the NBS professionals' session pointed out that the **lack of an ICD code obstructs further testing**, treatment, and billing, which are all essential elements of care.

Many listening session group discussions also focused on the **importance of insurance coverage** for rare diseases, with a number of participants calling for insurance coverage for treatments. "Rare disease treatment is often expensive because the market is small," shared a health policy researcher. As well, several participants pointed out that insurance responsiveness is essential, because, as a medical geneticist said, "having to get prior authorization for [confirmatory] testing could delay testing for a crucial couple weeks."

One potential solution raised by participants was a payors forum. As one participant from the rare disease families and advocacy groups listening session said, the goal would be to "**bring together insurance partners** and have conversations around coverage and ICD codes."

## DATA MANAGEMENT AND ADMINISTRATIVE INEFFICIENCIES

Participants pointed to a few key challenges in data management and administration for NBS programs. Common issues include a lack of standardization in data collection and analysis due to state-level variation. A substantial concern is the lack of data sharing across sectors, organizations, and states, which limits screening accuracy, research, follow-up care coordination, and timely and effective intervention. Moreover, the need for more communication and collaboration within NBS programs, and even across the country, was identified as a key underlying challenge.

This topic of discussion, in particular, yielded numerous ideas for solutions. Many called for greater data sharing, improved data management guidance, protection of genetic privacy, as well as data interoperability standards. Some respondents and participants suggested developing regional systems to improve consistency and establish data-sharing protocols, while others called for a national system to track patients and follow-up efforts.

15 Center for Disease Control. (n.d.) *International Classification of Diseases*. National Center for Health Statistics. Accessed July 19, 2024. https://www.cdc.gov/nchs/hus/sources-definitions/icd.htm

## Questionnaire

A small number of questionnaire respondents elaborated on challenges regarding data and NBS administration. State-variability means a **lack of standardization for data collection and analysis**, suggested some respondents. One healthcare provider suggested, "Perhaps we could develop regional systems to improve consistency and establish data sharing protocols."

A number of respondents highlighted that there is a **lack of data sharing** among NBS programs and related bodies, which limits research, care coordination and quality control. "We currently cannot assess whether these outcomes were realized or not because we do not have the structures and processes to assess performance," said an NBS lab professional.

**Specimen collection errors** and **clerical errors** were a challenge for a few respondents working in healthcare or rare disease advocacy.

Related, see the above section on parent education and support for concerns about data privacy.

## Listening Sessions

About an eighth of listening session comments about NBS challenges were about data management and administrative effectiveness. "We focus a lot on screening but there's no data or national system to count the number of kids who are screened positive. We need a **national system to track patients** and follow-up," recommended a medical geneticist participating in one of the mixed listening sessions. Participants described that states may not have the resources or technology for data capture and analysis which will have impacts on health equity.

Healthcare providers and administrators in particular called for greater **data sharing**, with the support of good guidance for **managing data** and **protecting genetic privacy**. Such participants also called for **data interoperability standards** and incentives for data collection.

> "We need a national system to track patients and follow-up.
>
> —Medical geneticist

A number of listening session participants also observed there is currently **missing and inaccurate data** in NBS and follow-up records which leads to gaps in screening, follow-up, research and planning. A number of participants noted that some **paperwork is handwritten** which leads to human error, recommending instead an electronic reporting system.

## NBS STAFFING LIMITATIONS

Participants identified workforce limitations as a key challenge for NBS, describing that many NBS programs suffer from understaffing and staff burnout. Comments also pointed to increasing challenges of staff capacity and readiness in the face of new conditions being added to screening panels, as discussed in the section on State-level Variation.

### Questionnaire

About an eighth of questionnaire respondents who are professionally connected to NBS discussed workforce challenges. "Having enough staff is critical for the successful implementation of newborn screening," said a public health professional, reflecting the views of a number of respondents. However, "most lab facilities are **underfunded, understaffed, and under supported**. They cannot compete with private labs to get and retain experienced staff," said an NBS professional.

There is a struggle to fill and retain NBS positions. "The workforce is aging, the **workload is increasing** as more programs add weekend staffing, and **salaries are not keeping pace** to attract younger workers," described a genetic counsellor. "Pay is not reflective of the individuals needed to run the program," said an NBS lab professional.

"As the RUSP grows and as testing becomes more complicated, the **personnel qualifications** become a hurdle (very few programs employ board certified laboratory geneticists)," said an NBS lab professional. One public health professional explained that because "hospital and clinic staff has had significant turnover, there is a **continual need to educate** the providers and hospital/clinic staff (lab and nursing) to ensure that parents are receiving good quality education about NBS."

Related, a clinical geneticist observed, "We don't have enough genetic educators, genetic counselors, and clinical geneticists to support the large expansion of genetic and genomic screening and diagnostic testing that we would like to see happen."

Some respondents called for more **federal guidance** on appropriate staffing and funding of NBS programs.

> "Most lab facilities are underfunded, understaffed, and under supported.
>
> —NBS professional

### Listening Sessions

Many NBS professionals and healthcare providers participating in the listening sessions said staffing is a "continual pain point." Others described the existing **staffing levels as inadequate**, while they are also experiencing increased workload. One NBS professional described the impact, saying, "it does not allow for the level of service that we feel we should provide."

Drivers of staffing limitations identified by participants include **subpar compensation levels**, lack of state job descriptions for roles like genetic counselors, **limited career growth** opportunities, current staff retiring, long work hours even on weekends and holidays, and more.

A number of participants noted that, together, these issues are contributing to NBS **workforce retention issues** and staff burnout. One of the NBS professionals explained that "rare disorder specialists are fatigued already and concerned with adding even more disorders."

## INSUFFICIENT NBS FUNDING

Questionnaire responses and input from the listening sessions highlighted concerns about funding for NBS programs. NBS lab professionals were particularly vocal about underfunding. Others noted that some NBS programs face financial pressures that hinders the maintenance, expansion, and updating of services, leading to disparities in screening programs. State disparities, staffing issues, follow-up care limitations, and research barriers were also linked to funding shortfalls.

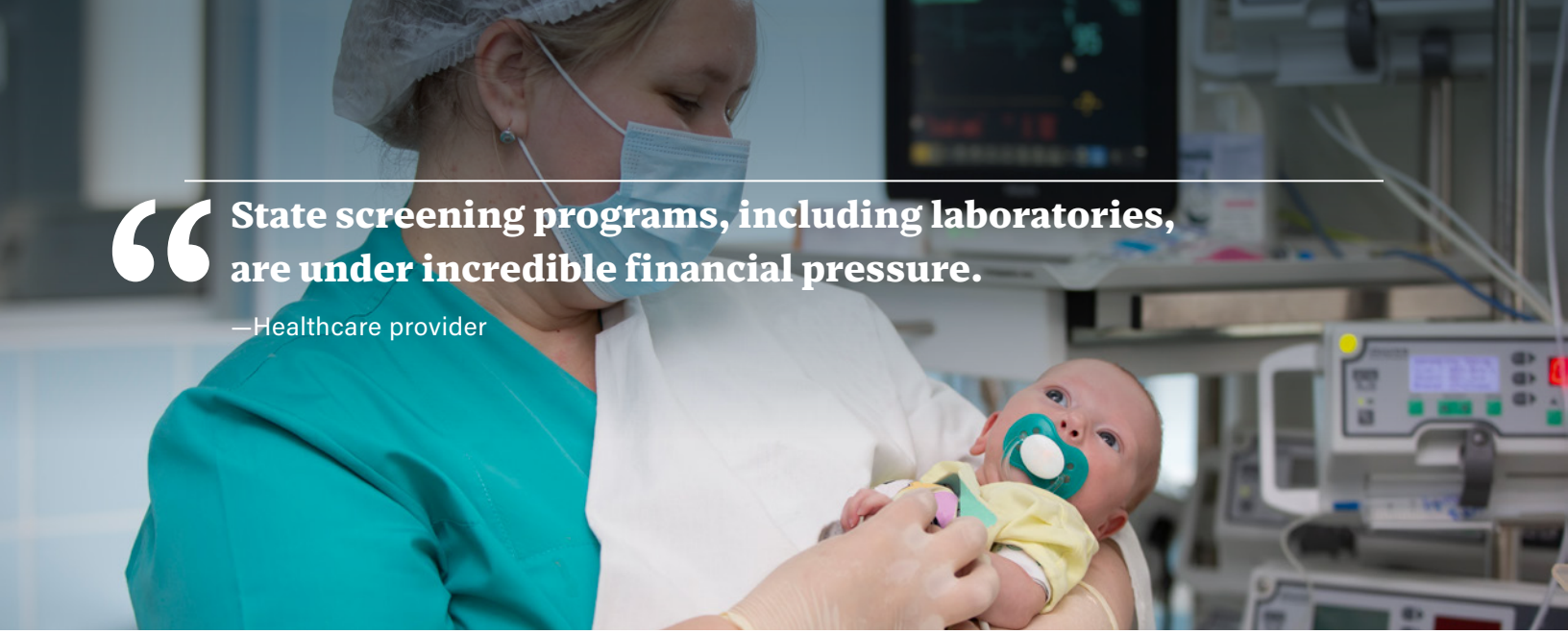
Some listening session participants called for more federal funding and resources to support NBS programs, emphasizing that insufficient funding affects all aspects of the NBS system, from laboratories to testing technologies, follow-up care, and research.

### Questionnaire

Each sector discussed insufficient NBS system funding, while NBS lab professionals and health researchers explored this more commonly in their comments. "The main challenges of the existing NBS is really the **lack of funding for labs** to support the conditions being added," shared an NBS lab professional.

"State screening programs, including **laboratories, are under incredible financial pressure**. It is impossible to maintain services and expand or update when needed due to financial constraints," described a healthcare provider.

Some respondents pointed out that the **state budgets** set by state administrators can be out of alignment with the requirements of running NBS.



> **State screening programs, including laboratories, are under incredible financial pressure.**
>
> —Healthcare provider

Respondents said that research for **developing new NBS tests takes time and money** but, in the words of an NBS lab professional, "The Newborn Screening system as a whole is funding-short, which significantly impairs both [the] development and adoption of new methods." This research is essential, as noted by a public health professional who said, "Most programs do not have qualified staff or the time and resources for method development." Instead, as some respondents observed, NBS research is mostly led by third-party private companies and universities.

### Listening Sessions

Participants suggested that the federal government needs to provide additional funding and resources to support the NBS system. **Insufficient NBS program** funding impacts laboratories; the availability of testing technologies; and follow-up care options, including telehealth, research, and more. One NBS professional observed that "some states are moving faster because they have resources or ability to compete for grants to get things done faster."

Relatedly, participants noted that adding new conditions to NBS **requires increased funding and capacity**. "We need equivalent funding [to] the cancer moonshot," recommended a medical geneticist. See above for challenges related to state disparities, staffing, follow-up care, and more.

## Funding Priorities for NBS

As part of a thought experiment, questionnaire respondents were asked where they would like to prioritize funding for the US NBS system. The opportunity to discuss priorities for funding the US NBS system was not a part of listening sessions.

Questionnaire respondents were somewhat evenly split among the three hypothetical options for funding priorities. Those who prioritized funding treatment for children with a diagnosis described the importance of affordable, equitable, and effective care. Those who prioritized funding to improve existing NBS programs emphasized the need to strengthen the current system before making other changes. Slightly fewer respondents prioritized funding for adding conditions not currently included in NBS; those who selected this option pointed to the importance of early detection. The results are outlined in the following table:

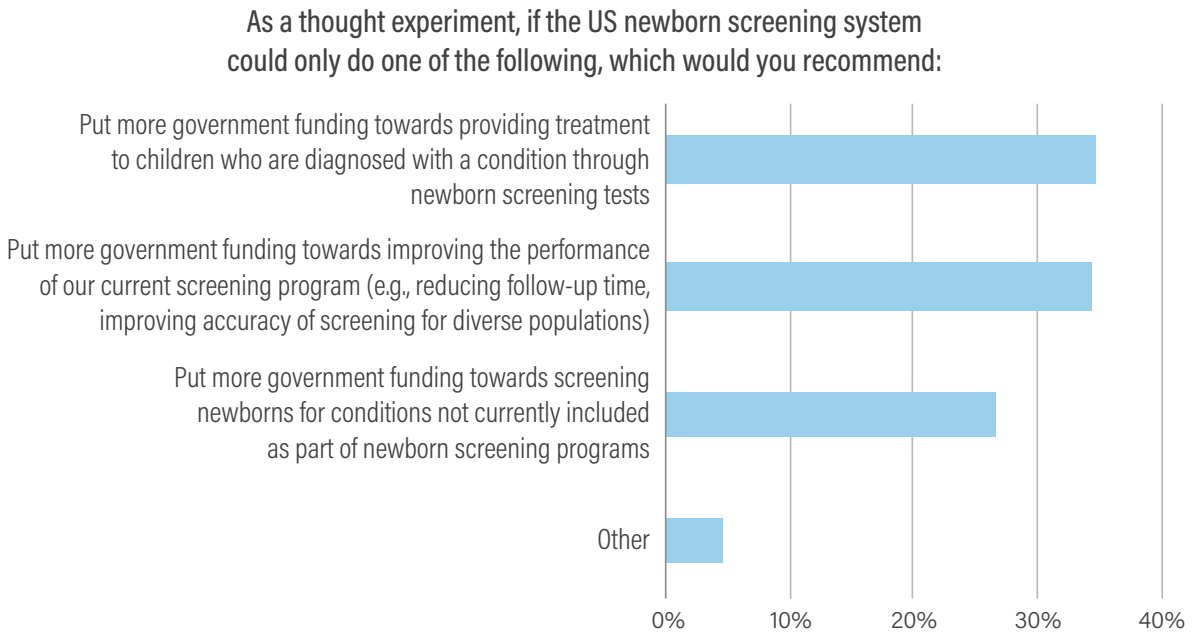



*Figure 13. Questionnaire respondents' answers to a hypothetical question regarding NBS funding. Respondents do not represent a statistical sampling of the American public.*

After the question about funding priorities represented in the table above, questionnaire respondents were asked: *Why did you choose this answer?* A number of respondents described that all of the hypothetical choices are important and it is difficult to prioritize one over the other. "Tough question," said a healthcare provider, as "we need to do all three!"

## Treatment

Of those who recommended allocating funding towards treatment, the most common sentiment was about **access to affordable healthcare**. This view is represented in one parent's response: "What good is knowing conditions if people can't afford treatments?" Many specified the need to ensure **equitable care**, including an NBS professional who said "infants are being left behind. A baby's [sic] socioeconomic status should not determine whether or not they get quality care."

Others pointed to the **goals of NBS** as their reason for prioritizing the treatment funding option. "There is no point in detecting conditions early, if there is no way to treat children," said one parent. Another theme raised was recognition of the costs and impact on family finances. "The financial burden is immediate, unexpected, and difficult," said a parent of a child with a rare disease.

### Current NBS program

Almost all of those who recommended improving the performance of the current NBS program agreed with the sentiment expressed by one NBS follow-up professional respondent, who said, "The **current structure needs to be improved on before expanding**." Other common reasons for prioritizing improvements to the existing system included the need to, in the words of a health researcher, "ensure the current system is equitably working for everyone", and the observation that most programs are overwhelmed by the number of new conditions recently to NBS.

### Adding conditions

Those who recommended funding the addition of new conditions for NBS pointed to the importance of **early detection**, including one parent of a child with a rare disease who said, "My daughter died because her disease was not caught in time to have treatment. NBS would have saved her life." Many respondents underscored that NBS enables **early treatment** which can save suffering, improve outcomes, and reduce healthcare costs over time.

Other respondents highlighted the potential to harness modern **technology** for identifying conditions, the **difficult process of adding conditions** to NBS, and the importance of **state parity** in NBS tests.

## UNCERTAIN SCREENING RESULTS

Input on how to address uncertain screening results in NBS programs came mostly from listening session participants, who were asked a specific question on this topic. Participants were also asked to discuss the issue of uncertain test results: *How should the newborn screening system respond to screening results that are hard to interpret or have unclear clinical consequences?*

Most participants agreed that building a **good follow-up and referral system** around NBS is critical for addressing the fact that many screening tests produce hard to interpret results. This means **increasing access to specialists** to support families navigating unclear or complex screening results—referrals to genetic counseling, disease specialists, and centers of excellence. It also means supporting access to services like telemedicine and expanded insurance. Many participants emphasized the need for systematized **longitudinal follow-up**—that is, long-term monitoring and scheduled follow-ups for parents with ambiguous NBS results.

A number of participants also noted the **need to limit false positives** in NBS results. Many participants raised concerns regarding the impact of false positives, with some also concerned about suggestions to invest in new types of screening that may produce unclear results. Healthcare professionals and NBS professionals were particularly adamant about this, with one participant in the NBS professionals listening session saying that the "responsibility on screening labs is to have few false positives; the responsibility on everyone else is to make sure babies are taken care of with structure."

For some, the solution would be to ensure the criteria for the RUSP precludes the addition of new diseases that do not have reliable screening. One participant from the health industry made the following suggestion: "Do not report on a variant of unknown significance. Only report on what is known in the database. Only report on pathogenic variants. Keep improving diagnostic acumen in this way." The participant also suggested "tiers of testing based on certainty of variants and unknown significance." Others called for more federal funding to implement more testing for new screening methods.

A substantial number of participants at the listening session for rare disease patients, families, and advocacy organizations suggested that ambiguity in screening results should be accepted as a potential outcome of NBS. One participant in the rare disease community session noted, "Parents nowadays are more receptive to receiving ambiguous information." Another at the same session suggested, "If you frame false positives as a tax to pay, then the **general public can tolerate some of the consequences of false positives** in order to diagnose more babies." But a couple of participants at the listening session for healthcare providers also raised the issue that NBS is currently unconsented or done without parents' knowledge, which would have an impact on how parents receive results that are unclear or hard to interpret.

> "Parents nowadays are more receptive to receiving ambiguous information.
>
> —Participant in the rare disease community session

In the listening session convened with the assistance of REACHUP, Inc., mothers participating with limited knowledge about NBS prior to the session had mixed perspectives about uncertain NBS results. One mother expressed **discomfort with expanding tests** if there is more likelihood of false positives, saying, "I like to be in less of the gray area … you might possibly panic about something that doesn't actually have the possibility of really happening." This parent went on to say that if there is a likelihood of false positives, the parent should know that "some kinks are still being worked out." Another mother in this session observed that "nothing is perfect. It's subjective to every parent. Some people want to have the **peace of mind** [by knowing all the likelihoods] and some people just want a clear answer, it just depends on the parent… do they want to live in the gray?"

> "Nothing is perfect. It's subjective to every parent. Some people want to have the peace of mind [by knowing all the likelihoods] and some people just want a clear answer, it just depends on the parent… do they want to live in the gray?
>
> —Parent participant

## BLOOD SPOT RESEARCH

Questionnaire respondents were asked specific questions about what researchers can or should do with residual blood spot samples after an NBS program is complete.

Residual blood spots are dried blood spots that are 'left over' after all screening tests have been completed. These samples are not labeled with the name of the child—this is called deidentified. These samples are used to improve NBS for future infants, including performing regular quality control to make sure the screening tests consistently produce accurate and consistent results. These samples also can be used for other types of research. Regulations for this research varies state to state.

As a thought experiment, questionnaire respondents were asked to share their views on notification and consent for three different research situations. The first case was: *If researchers want to use leftover, deidentified biospecimens from adult patients (e.g., from a blood draw) for medical research to improve patient care.* Respondents were asked to indicate whether they thought the individuals should be notified and asked for their consent. Just under half said they should be notified and asked for consent, while about one-third said neither would be necessary in this situation. One-tenth said the individuals should be notified but not asked for their consent, or something else.

The second question in this series was: *If researchers want to use residual, deidentified newborn bloodspots for research directly related to newborn screening (e.g., develop a new screening test for a condition that could later be included in public health newborn screening programs)*. Over two-fifths of respondents said parents/guardians should be notified and asked for their consent. Over one-third selected: "It is fine to do so without parental/guardian notification or consent." About one-tenth said parents/guardians should be notified but not asked for their consent, or something else.

The last closed-ended question about bloodspots was: *If researchers want to use residual, deidentified newborn bloodspots for other types of medical research unrelated to newborn screening.* Almost two-thirds selected: "Parents/guardians should be notified and asked for their consent." One-fifth said: "It is fine to do so without parental/guardian notification or consent." About one-tenth said parents/guardians should be notified but not asked for their consent, or something else.

These results reflect respondents' somewhat mixed views about the **role of consent** in medical research. More respondents indicated that individuals should be asked for consent for their deidentified bloodspots to be used in research, particularly for medical research unrelated to NBS. With regards to using the residual blood spots directly for researching for new screening tests, respondents were more split, with a larger portion indicating that consent from parents and guardians is 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 strengthen families' experiences with NBS. Other input described the benefits of collaborations among governments, advocacy groups, medical professionals, NBS professionals, industry, universities, researchers, and others.

### Questionnaire

Respondents of the questionnaire recommended greater collaboration to ensure a consistency of care, regardless of a baby's location. Collaboration was also recommended as a means to support data sharing, engage with families as partners in care, strengthen NBS research and testing, and leverage strengths across the NBS system. Improved communication with families about NBS and its results was an especially strong theme in the questionnaire.

### Listening Sessions

In listening session discussions about challenges, many participants noted the need for **collaboration and communication**. One participant from the rare disease families and advocacy groups listening session said, "Most of us are not here by choice. We are here by force. We need those of you who were here by choice to help us." To support families, a number of participants called for better case management and mechanisms to help people navigate the system, tie **community-based resources** to clinical care, and leverage the public health departments' educational and home visitation systems.

> "Most of us are not here by choice. We are here by force. We need those of you who were here by choice to help us.
>
> —Participant from the rare disease families and advocacy groups

**Government partnerships** was recommended as a key solution by some participants. Partnerships between states and advocacy groups could inform both NBS programs and follow-up. Collaboration among federal departments could address silos in the NBS system. States could learn from one other by sharing investigations and best practices.

A participant working in Medicaid emphasized the benefit of **information sharing** for rare disease treatment approvals. "It saves a lot of steps when the right people get the right information," they said. Participants also cited collaboration among and between industry, universities, and other researchers as an area of promise.

## FINAL ADVICE TO THE COMMITTEE

To strengthen current NBS programs, engagement participants offered a range of suggestions to the committee, including advice on how to address health disparities, approach NBS through a regional lens, and review the RUSP. They also offered feedback on parent education, collaboration, innovative technologies, data privacy, the NBS workforce, and continued engagement with families.

### Questionnaire

Questionnaire respondents were asked: *What advice do you have for the National Academies committee examining the US newborn screening system, particularly about how to improve the system for the future?*

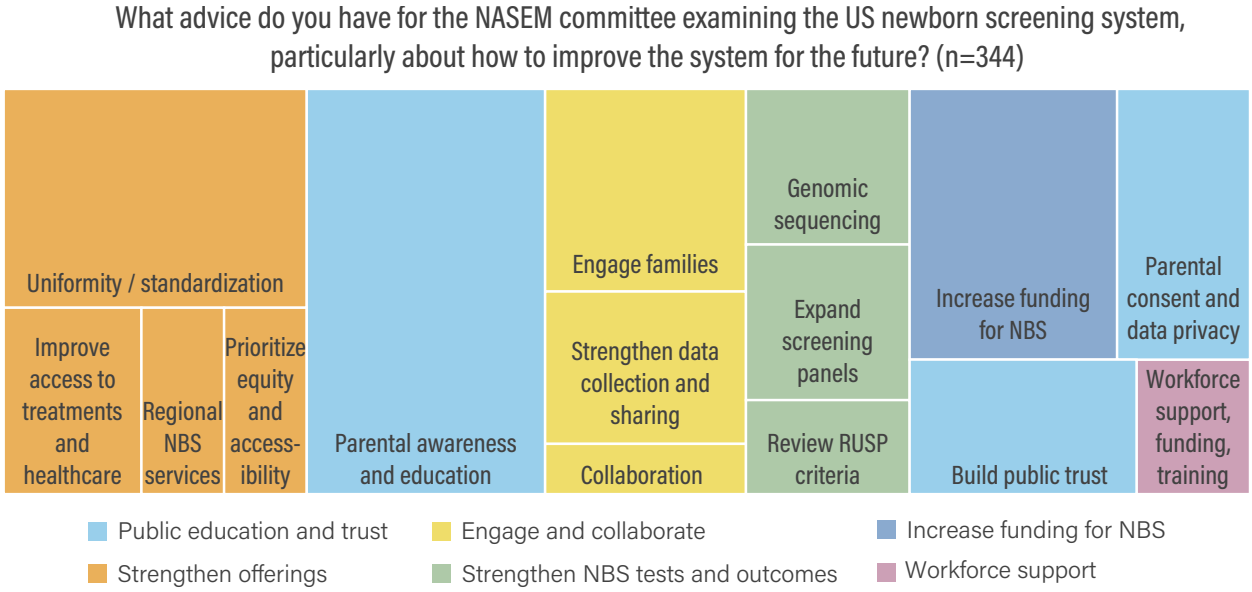



*Figure 14. Questionnaire themes about recommendations for the National Academies NBS committee. The box size corresponds with the frequency of comments on the subject. Respondents do not represent a statistical sampling of the American public.*

### Public education

One-fifth of all responses to this question described the importance of **parent awareness and education** before and after birth. A representative comment came from a parent of a child with a rare disease who said, **"**Better communication to parents who are having newborns as to what the newborn screening is, how it is being administered, and when results should come back are all very important. Also, it is important to have clear communication to those parents receiving a negative result as well." Accessible education materials and prenatal education were also key themes on this topic.

### Data Privacy and Parental Consent

Respondents also recommended that the committee consider the role of **data privacy**. "Program transparency and accountability include communicating how leftover DBS [dried blood spots] are stored and may be used," said a health researcher. A parent said, "Bloodspots contain genetic material that is intensely personal, and parents shouldn't have to give up that fundamental right to privacy in order to have their children screened."

Other respondents underscored the importance of engagement on this topic, including an NBS follow-up professional who said, "Engage with minority and Indigenous communities to ensure their views on data privacy and DBS storage/use are taken into account."

A parent offered the following perspective: "Understand that the majority of end-users of the newborn screening system are not rare disease patients, but instead patients with negative test results. Given that their children did not have a disease identified, their main concerns and considerations may not be related to diagnosis and care, but instead privacy, consent for the use of samples, and why their child may need to be screened in the first place."

Views on **parental consent** for NBS were somewhat mixed. Many respondents wanted to see the informed consent process strengthened for both NBS and research. Most respondents were supportive of the opt-out process due to the serious nature of NBS conditions, while others wanted more choice for parents.

> "Understand that the majority of end-users of the newborn screening system are not rare disease patients, but instead patients with negative test results.
>
> —Parent participant

"I personally don't care at all how leftover blood is used, either from my baby or myself, because my default stance is that we should be improving these processes at all times and I'm game for whatever that takes. But I'm not part of a marginalized population so I recognize I should defer to them, because ultimately, consent is important and people may have reasons why they want to opt out."

See the above sections on bloodspot research and parent education and support for further comments on privacy and consent.

## Public Trust

Many respondents connected **public trust** with parent education. Some respondents were forward-looking in their advice about public trust; "As excitement grows for sequencing the genomes of newborns," said a health researcher, "we will lose the public trust in NBS without assurances that newborn genomes can't be accessed by law enforcement."

Public trust in NBS was also a specific area of inquiry in the questionnaire. All questionnaire respondents were asked: *What factors are most important for strengthening trust in the newborn screening program?* Respondents had the option to choose up to three from the provided list.

Three-quarters of respondents recommended education about the purpose and procedures of NBS. Half of respondents recommended clear communication about the goal of NBS and the meaning of results, as well as greater transparency and accountability in NBS programs.

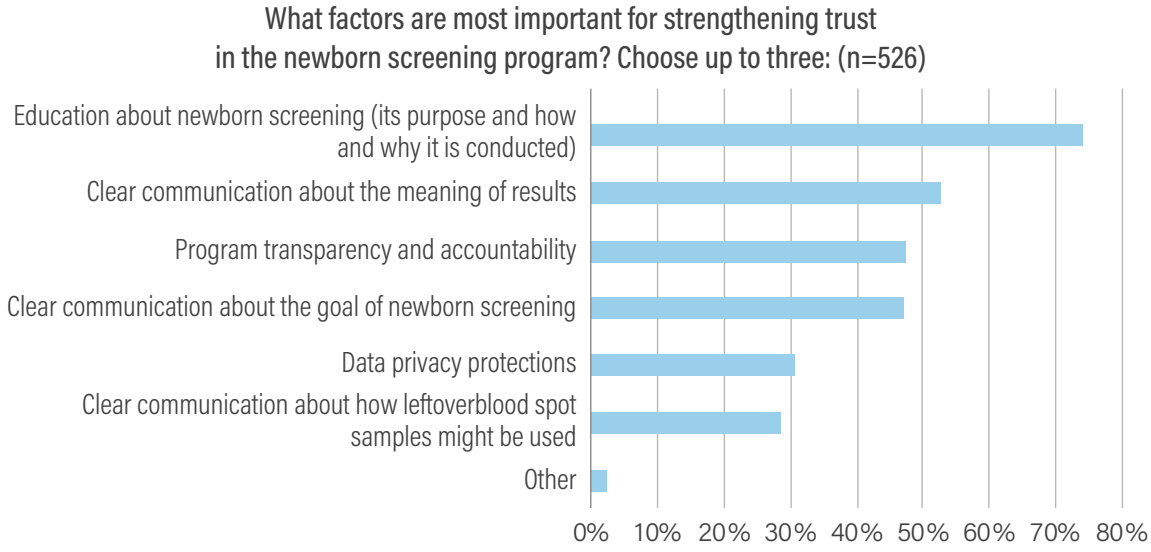



*Figure 15. Questionnaire responses on ways to strengthen public trust in NBS. Respondents do not represent a statistical sampling of the American public.*

## Strengthen Existing NBS Programs

One-quarter of respondents sharing their advice with the committee focused on strengthening today's NBS programs. "Before looking to the future of NBS, consider the shortcomings happening right now and how these must be addressed before we seek to expand NBS programs in the United States," said an NBS follow-up professional.

One-eighth of responses advised that the committee address the **unevenness of NBS from state to state**. "The programs need more uniformity across states," said a public health professional. Many called for a federal standard, including one NBS lab professional, who said, "Propose a federally mandated minimum NBS program that should be followed by all states."

"Make sure the system works for those that are screened. Identification and diagnosis without access to treatment is a failure of the system," advised a parent of a child with a rare disease. Responses about **improving access to treatments** recommended looking at equity through the lens of race, income and geography. A number of respondents called for more funding for NBS and follow-up treatment, as well as more funding for the healthcare system overall.

Some respondents advocated for **regionalizing NBS testing and follow-up**. An NBS lab director described that "state NBS labs are terribly under supported ... Regionalization of labs may be the best solution," as this approach would pool funds and staff.

> Identification and diagnosis without access to treatment is a failure of the system.
>
> —Parent of a child with a rare disease

Other respondents focused their suggestions on **regionalizing access to specialists**, such as a health researcher who said, "I would love to see a system where we think about how to increase access to care for those who are diagnosed. Maybe fund a regional model of care where the few specialists can see patients in several states through clinics, telemedicine, etc."

Lastly, a number of respondents called on the committee to **prioritize equity and accessibility**. Equity meant different things to the questionnaire respondents. Some called for ensuring NBS equity for all Americans regardless of their socio-economic identities. Others emphasized equity through the lens of including a wider range of conditions in NBS screening.

### Modernize NBS testing

Strengthening NBS tests and outcomes was a primary concern for many respondents. Some respondents suggested incorporating **genomic sequencing** into NBS to enhance diagnostic accuracy and early detection. Additionally, some respondents expressed that expanding the screening panels to include a **wider range of diseases** will ensure a more comprehensive approach to newborn health. Some respondents also mentioned that to maintain the effectiveness of the program, it is essential to review and refine the RUSP criteria regularly.

A few respondents also suggested **improving the quality of bloodspot collection**. They said ensuring high standards in this initial step will lead to more reliable test results and better health outcomes for newborns. In their opinion, addressing these key areas will strengthen NBS testing and follow-up, ultimately improving the effectiveness of the program.

### Workforce Support

Most participants said **increasing support, funding, and training for the NBS workforce** is necessary. Some respondents said that enhanced financial resources and training programs will ensure that NBS staff are well-prepared and capable of managing the screening process effectively.

**Improving healthcare providers' access to results** was also mentioned by many respondents. They suggested that streamlined access to screening outcomes will enable timely and accurate diagnoses, enhancing overall patient care.

## Increase Funding

**Increase funding for NBS** was a consistent recommendation across all respondent groups. Two groups had particularly high levels of support for recommending this action: NBS professionals and public health professionals. Some participants mentioned that enhanced financial support for NBS will enable the implementation of advanced technologies, the expansion of disease panels, and overall improvements in the screening process. According to multiple respondents, by addressing these key areas, the NBS program can achieve greater effectiveness and reliability.

## Engage and Collaborate

A strong message in the questionnaire respondents' advice was to **engage families and patients** in planning the future of NBS. Respondents most often recommended that efforts to strengthen NBS should engage parents and families, including both those who have a child with a rare disease and those who received false positive results—an initial positive screening result that subsequently resulted in a negative diagnosis. Other groups suggested for engagement include specialists, pediatricians, minority and Indigenous communities, medical organizations, patient safety organizations, and patient advocacy organizations. "The community needs to be partners in determining how NBS should grow and how it could facilitate future research," said a health researcher.

One healthcare provider underscored the importance of listening to all perspectives, saying, "The NBS system also needs to stop dismissing everyone who disagrees with them as crazy or uninformed."

Respondents called for **collaboration at all levels of the system** in planning the future of NBS. Groups that participants recommended working with include "the most recent HRSA RFI[16] and patient advocacy organizations working to improve the system", general clinical laboratorians and pathologists, other national organizations, state programs, and other nations running NBS programs.

A number of respondents called for **strengthening data collection and sharing** for longitudinal perspectives, with clear guidance for data privacy. "[I] would suggest a funded national workgroup on improving and harmonizing standards for collection, communication, and data exchange on newborn screening," said a healthcare provider. Respondents cited the following benefits of data improvements: better understandings of current practices, improved patient outcomes, and enhanced capacity to be critical about planning for the future.

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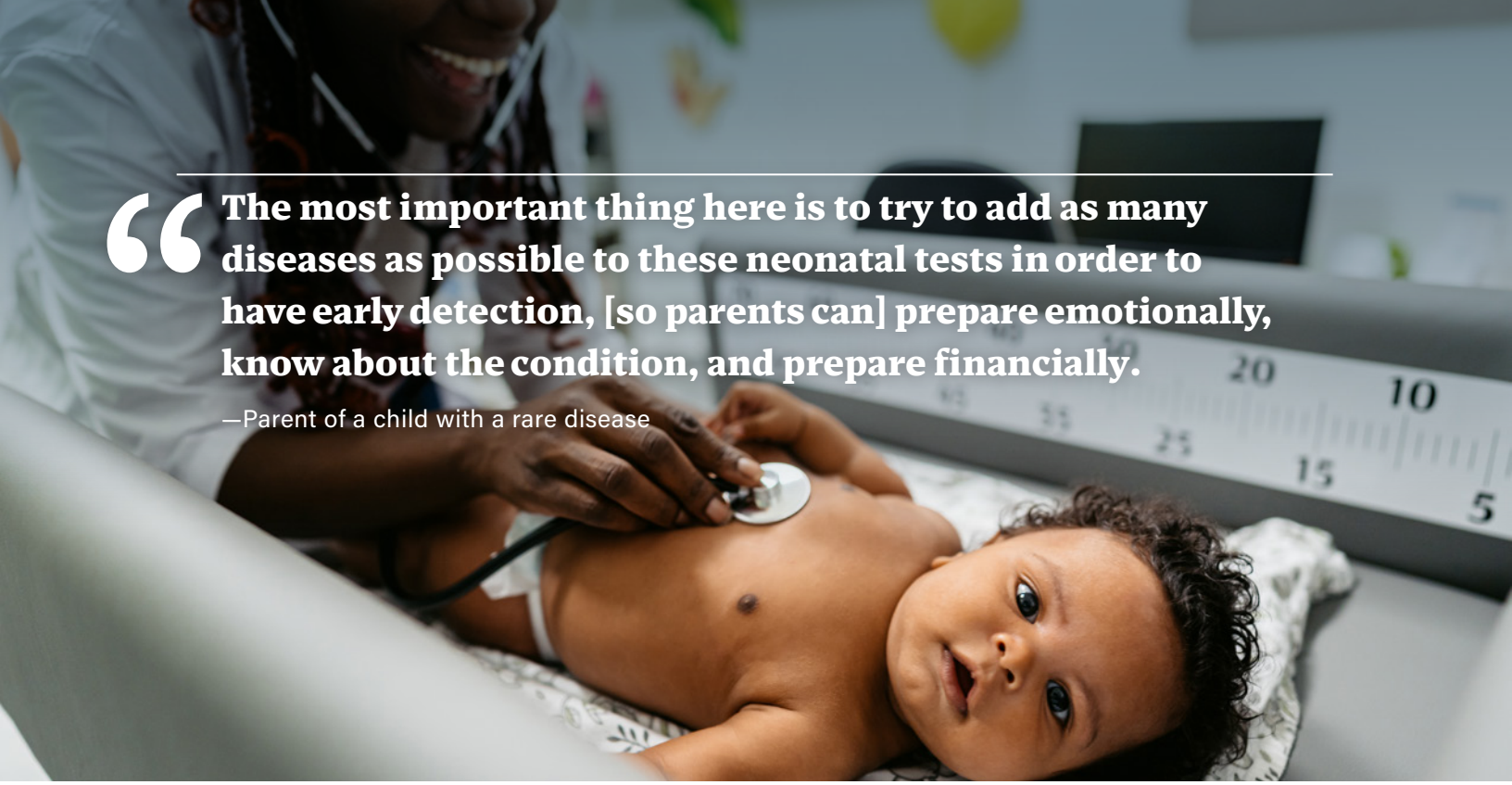
## Listening Sessions

At the end of the listening sessions, participants were asked to briefly share any advice they might have for the committee: *Given what you discussed today, what advice do you have for the National Academies committee charged with strengthening current and future newborn screening systems in the US?*


*Figure 16. Listening session themes about recommendations for the National Academies NBS committee. The box size corresponds with the frequency of comments on the subject. Participants do not represent a statistical sampling of the American public.*



> **The most important thing here is to try to add as many diseases as possible to these neonatal tests in order to have early detection, [so parents can] prepare emotionally, know about the condition, and prepare financially.**
>
> —Parent of a child with a rare disease

As displayed in the chart on the previous page, participants shared a range of advice. Some suggested the idea of fostering greater **collaboration**, such as cross-sector collaboration, and enhancing referral networks around NBS programs. This could involve more collaboration with national organizations and other national programs, as well as offering regional solutions for NBS programs where there are geographic challenges, such as the regionalization of referrals to specialists across state boundaries or the regionalization of NBS laboratories.

A number of participants in the listening session for healthcare providers put forward ideas for substantially remodeling the NBS system, that is, **reimagining NBS**. One NBS professional stated that the committee should "consider the role of NBS as a public health entity and not so much as another diagnostic tool." A participant from industry stated that the committee should "evaluate and significantly change RUSP to USP [Uniform Screening Panel]."

Others were interested in seeing more efforts focused on improving NBS system capacity, such as **addressing staffing and capacity constraints**. One participant suggested during the listening session for NBS professionals that "there should be more national guidance on how NBS programs should be staffed—what type of training staff should have," and that "there should be more specific guidelines as to what is expected of follow-up."

A number of participants were also interested in **modernizing the RUSP**. One participant in the listening session for rare disease patients, families, and advocacy organizations reflected that "in terms of the current NBS system and how to add more conditions, we should get to RUSP 2.0 to have a scope to enable a task group to evaluate new technologies, treatments, diseases that are ready to be screened to add all together, instead of adding one disease at a time." A parent of a child with a rare disease participating in the session convened with the assistance of The Akari Foundation said, "Lo más importante es tratar de añadir todas las enfermedades que sean

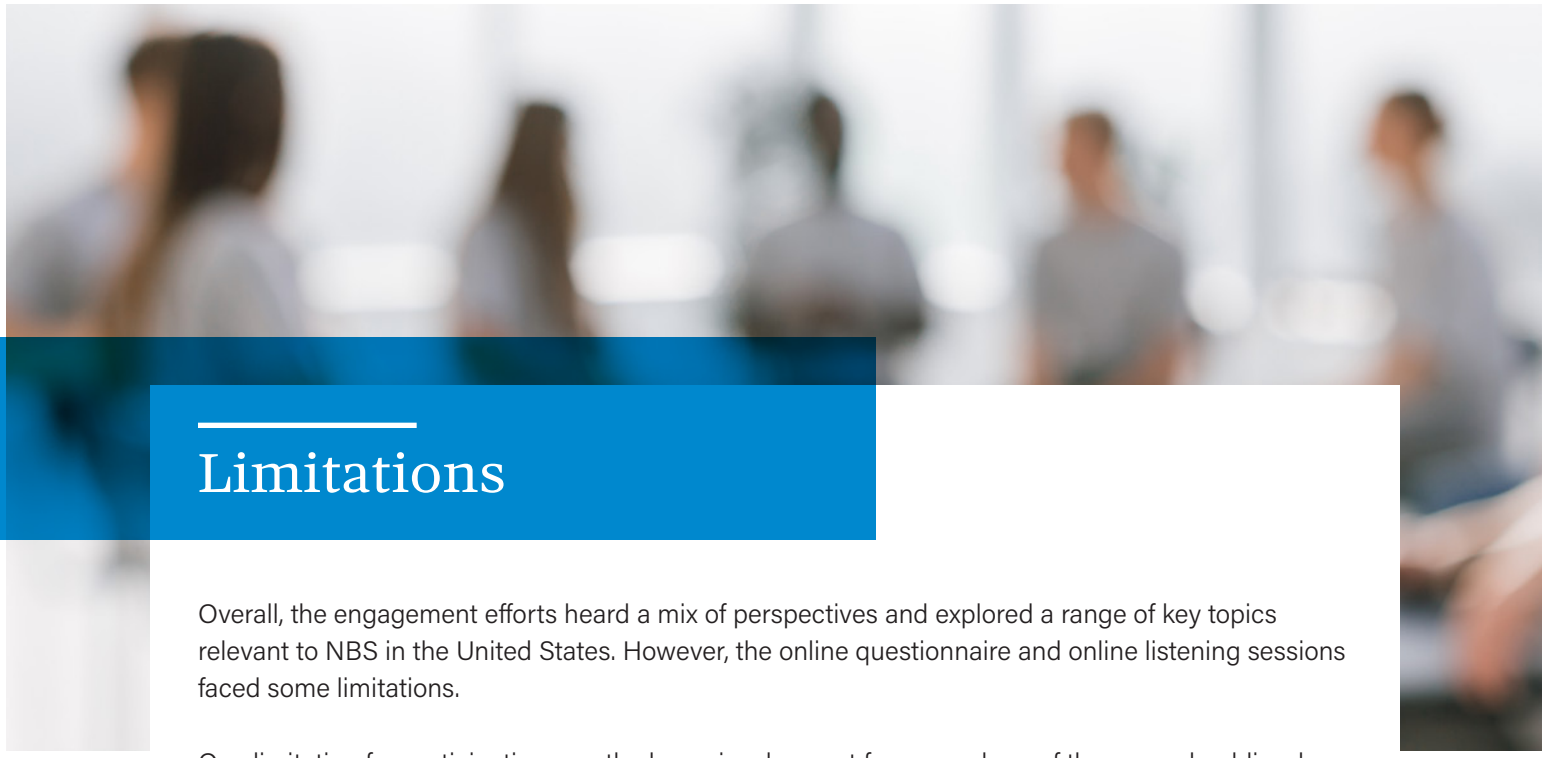
posible a estas pruebas neonatales para poder tener una detención temprana, poder prepararse emocionalmente, conocer de la condición, prepararse económicamente" [The most important thing here is to try to add as many diseases as possible to these neonatal tests in order to have early detection, [so parents can] prepare emotionally, know about the condition, and prepare financially]. Many were also generally interested in **decreasing the time it takes to add diseases** to the RUSP.

The idea of having **NBS programs operate uniformly** across the country was also raised by a number of listening session participants. Some also pointed to the need to embrace and anticipate new innovative technologies.

Others also noted the need for NBS programs to have more access to **government funding**, especially at the federal level, and that it would be important to be mindful of the **need to navigate policymaking** at different levels of government.

One participant at the listening session for NBS professionals, for example, said, "As newborn screening looks toward genetic testing for the future of screening, choosing conditions where a biochemical screen can accompany genetic testing results can help clarify variants of uncertain significance."

Finally, participants at the listening session for healthcare providers particularly emphasized the need to **engage parents more**, either by educating parents on how to navigate NBS programs and the diagnostic journey or simply to **empower parents** with the information to make decisions.



# Limitations

Overall, the engagement efforts heard a mix of perspectives and explored a range of key topics relevant to NBS in the United States. However, the online questionnaire and online listening sessions faced some limitations.

One limitation for participation was the lower involvement from members of the general public who do not have an existing interest in the topic of NBS. This is a somewhat common occurrence in engagement work.

Related to this, participant demographics do not reflect the nation's diversity. 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 also disproportionately from people who primarily identified as parents (general parents 16%; parents of a child with rare disease 14%) and healthcare providers (27%). 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.

## QUESTIONNAIRE

Responses to the questionnaire came disproportionately from people who primarily identified as parents (16% were general parents and 14% were parents of a child with rare disease) and healthcare providers (27%). Other groups participated at rates of 10% or less; these include those who identified as having a rare disease or condition, rare disease advocates who did not primarily identify as parents of children with a rare disease, health industry representatives, privacy advocates, and general public. (See the Who Participated section for details).

The questionnaire was made available online in English and Spanish. People with limited proficiency in these languages or without access to the internet could have faced barriers to participating.

Responding to demographic questions was optional. 12% of respondents chose not to answer these questions. Furthermore, approximately half of respondents did not answer every question, which limited the engagement team's ability to perform subgroup analyses for some questions.

## LISTENING SESSIONS

Each listening session was 2.5 hours and yielded rich and insightful discussions for gathering input. The listening sessions were attended by people coming from a diverse range of experiences in relation to NBS in the United States. However, there was limited attendance from people who identified with the payor and healthcare industry categories. The engagement team also faced difficulty reaching general family groups and organizations who are not proactively organized or engaged in NBS.

In addition, some participants took part in listening sessions that were convened for a different sector than their own. This affected sector-specific aspects of the breakout room discussions as it included input from people with less expertise in said sector.

Lastly, promoting and conducting the engagement activities for this project took place primarily from March through May 2024. This compressed timeframe affected the nature and timing of the project's outreach to identified associations, organizations, and other groups. Specifically, it impacted how these groups received information about the project, how they shared it through their members or networks, and how participants responded and engaged.

# Conclusion

Learning about a range of perspectives of various people affected by NBS across the country is a critical step for assessing what is needed to strengthen NBS in the United States. Through their engagement, questionnaire respondents and listening session participants will deepen the committee's understanding both of how NBS programs in the country are saving lives and how they can be strengthened. The ideas represented in this engagement summary offer insights on how NBS can adapt to meet the needs of those personally and professionally impacted by the NBS system.

The participants' 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. As represented in this engagement summary, there are diverse perspectives on most aspects of NBS.

The input reflected in this engagement report will inform the committee's work to examine the current landscape of NBS in the United States, which will be an important contribution for helping modernize NBS and progress towards an equitable public health program from which all infants benefit.

# Appendices

## APPENDIX A: SELECTION OF QUOTATIONS FROM ENGAGEMENT PARTICIPANTS

"A valid NBS test and valid treatment may exist for a disease, [but] it takes the better part of a decade to get it [added to the RUSP], and taxes the advocacy organizations who can least afford to spend their valuable time and resources on this."

—Parent of a child with a rare disease

"New conditions are added to the RUSP regularly and are expected to be added to each NBS program in a timely manner. Tremendous resources and expertise including laboratory technology, clinical care, informatics, data analytics, and project management are required to add new conditions to programs that have limited resources and daily responsibilities of screening and following thousands of newborns."

—Newborn screening lab professional

"The process of adding conditions to NBS programs is way too slow and reliant on old criteria that are no longer valid our era of increasingly precise and cost-effective genomic screening tools. That's why federal guidance through the RUSP is, in my view, excessively conservative. This conservatism trickles down to state labs, which often follow the guidance and don't have the resources to add much else through pilot programs."

—Parent of a child with a rare disease

"While infants are generally screened universally, the system for follow-up and treatment for screen positive infants mirrors our healthcare system. [Some] Infants are lost to follow-up or don't receive treatment in a timely manner."

—Healthcare provider

"There isn't uniformity from state to state, meaning sometimes families find out about their child's condition based on luck of residence alone. Even federally recommended diseases don't have to be added in every state. Luck or happenstance shouldn't be a factor in determining if a child has a rare disease not immediately apparent at birth."

—Parent of a child with a rare disease

"Expanding a system that is under-resourced and therefore inefficient and under-performing, fails more people while continuing to fall short of current goals. Improvements to the current system, achieving better efficiency would free up resources for new efforts"

—Public health professional

"In a post-Covid America, it appears that the more parents hear about newborn screening, the more they may distrust and seek to opt out of it, despite its demonstrated benefits for newborns as a whole. Better, more nuanced, and earlier parent education may help to enhance parental and community trust -- as would more robust privacy protections for NBS samples/data on the back end."

—Privacy advocate

"As we move forward, maybe the question that needs to be asked is, 'Is identification and treatment of this condition urgent enough that parental consent should be waived?' And if that answer is 'No,' then the condition may not belong with NBS but could [be] offered to parents at a later date outside of the NBS program."

—NBS laboratory professional

"The current system provides not enough information and agency to parents. Informed consent should be required, and it should not be an opt out system."

—Parent

"The most important need of the rare disease community is more effective treatments for more diseases... Development of new treatment or more effective treatment of any rare disease depends on identifying patients early enough, ideally pre-symptomatic, so that the treatment benefits can be better demonstrated. Newborn screening can be the spark of treatment development that in turn fuels the further expansion of newborn screening...It's a chicken and egg dilemma that constrains both newborn screening and treatment development. We can only break out of it if we find a way to tackle both at the same time."

—Parent of a child with a rare disease

"Although these bloodspots have amazing potential for equitable public health research, the failure to protect bloodspots from misuse in criminal investigations and the lack of transparency in storing and using these bloodspots severely damages public trust."

—Health researcher

"A non-consented/mandated public health program has a big responsibility to promote trust and to not inadvertently cause harm by being distracted by special interests outside of the scope of newborn screening."

—Healthcare provider

"We currently cannot assess whether [the proposed benefits of screening] were realized or not because we do not have the structures and processes to assess performance."

—NBS lab professional

"We focus a lot on screening but there's no data or national system to count the number of kids who are screened positive. We need a national system to track patients and follow-up."

—Medical geneticist

"Although I appreciate the desire to press forward and help more children, we absolutely need to make sure that the current system is equitably working for everyone. Otherwise, we will continue to leave children behind. Progress rarely appreciates thoughtful reassessments, so we need to prioritize ensuring that public health interventions work for the entire population before moving forward and continuing to drive health disparities. Those who do not receive the full benefits of this program will never trust public health officials or healthcare providers to truly have their best interests at heart because they have evidence to the contrary."

—Health Researcher

"Clear communication, openness, and transparency would help build trust. There is lack of understanding about DNA and the government. The system needs to be reexamined in light of the advances made in science over the years since it first went into effect."

—Parent of a child with a rare disease

"A national public outreach campaign about newborn screening would be extremely helpful to achieve many of the factors listed above and could help improve trust, transparency and educate the public and providers all at the same time."

—Public health professional

"The general public has poor understanding of NBS. There's a lack of trust by many about any government programs. Add to this the apprehension about genetic information being shared. Lack of education about NBS leaves state and federal programs vulnerable to funding loss or worse. At a minimum, there should be more education for expectant parents (grandparents) to prepare them for potentially receive out of range results."

—Healthcare provider

"Having an opt-out process causes a lack of informed consent with newborn screening. Too many families report never having heard about the newborn screen until their child has an abnormal result or otherwise requires a repeat NBS. The lack of informed consent can lead to difficulty in coordinating follow-up testing with families, as the results are returning during the postpartum period when parents are often sleep-deprived and/or focused on just keeping their child alive. I think the news of an abnormal/critical newborn screen can often feel blindsiding to families and, in some scenarios, may feel like too much to try to understand/comprehend without warning. This can also cause mistrust with the healthcare system. I also think there can be a lack of provider knowledge about the newborn screen at the pediatrician level, which also leads to issues with family follow up, including false reassurance and/or over-medicalization of children with likely false positive newborn screens."

—NBS Follow-up professional

"As technology advances, it's easy to want to advance newborn screening as well. However, it requires thoughtful consideration about whether these advances truly advance newborn screening. Are we doing things for research purposes or to actually help babies and their families?"

—Newborn screening follow-up professional

"Science and technology have advanced at a rapid pace and we can diagnose conditions much more easily, quickly, and painlessly. There are also more treatment options available. It is important to effectively utilize these advances for the betterment of our children/our future, to give them the best opportunity possible for continued good health."

—Rare disease advocate

"Remain mindful of the goal of newborn screening. don't try to make newborn screening something it is not- can different types of screening or opt-in testing be more appropriate for some disorders? A non-consented/mandated public health program has a big responsibility to promote trust and to not inadvertently cause harm by being distracted by special interests outside of the scope of newborn screening."

—Health care provider

"The biggest current challenges to NBS are attempts to broaden the scope of testing to include parents desires or interests, to add disorders with untested treatments, which serves to further undermine trust in the system and increase harms (to families and infants). If one of the "benefits" of screening is for parents to have a choice whether to treat their infant, then that condition is appropriate for an opt-in NBS test but not for an opt-out or mandatory test. The current paradigm appears to be that industry starts supporting "advocacy" groups during drug development so that the company has as large a group of consumers as possible as soon as their drug is approved, even if the approval is accelerated (thus may not have proven effectiveness). The lack of options besides NBS is truly limiting to making good decisions about adding conditions. Mandatory NBS is at risk because there is no option for more appropriate screening for conditions that don't meet the high bar for current NBS"

—Health researcher

"Public awareness and trust of newborn screening (NBS) are important to ensure successful and universal screening. Parent education can be best done during the prenatal period but it has been difficult to obtain cooperation from obstetricians."

—Newborn screening lab professional

"All babies [are] screened unless parents opt-out: This encourages whole population participation in the screening process and allows for time-critical disorders to be identified before morbidity or mortality."

—NBS lab professional

"Our daughter's newborn screen changed the entire course of our lives. We did not know about this disease, and likely never would have. The support and education we have received since the positive result have been a blessing. It is knowledge that EVERY family deserves."

—Parent of a child with a rare disease

"Lo que funciona es que uno no tiene que pagar por esa prueba, no importa si tienes plan médico privado o plan médico del gobierno, es totalmente gratis [What works is that you don't have to pay for the test, it doesn't matter if you have a private health plan or a government health plan, it is totally free.]"

—Parent of a child with a rare disease

"Every day a that a test is not made available is a day that a child entering this world can be missed and forced to endure unnecessary hardship. Knowledge doesn't take away the condition, it is always there. Knowledge allows informed decision making. Knowledge helps to avoid unnecessary burdens on exploratory pipelines. Knowledge allows for quality of life improvements. Knowledge allows for future family planning decision making. Knowledge allows for memories to me made, for the precious lives of our children to not be wasted searching for answers. Knowledge is everything."

—Parent of a child with a rare disease

"Too many families report never having heard about the newborn screen until their child has an abnormal result or otherwise requires a repeat NBS. The lack of informed consent can lead to difficulty in coordinating follow-up testing with families, as the results are returning during the postpartum period when parents are often sleep-deprived and/or focused on just keeping their child alive. I think the news of an abnormal/critical newborn screen can often feel blindsiding to families and, in some scenarios, may feel like too much to try to understand/comprehend without warning. This can also cause mistrust with the healthcare system. I also think there can be a lack of provider knowledge about the newborn screen at the pediatrician level, which also leads to issues with family follow up, including false reassurance and/or over-medicalization of children with likely false positive newborn screens."

—Newborn screening follow-up professional

"Understand that the majority of end-users of the newborn screening system are not rare disease patients, but instead patients with negative test results. Given that their children did not have a disease identified, their main concerns and considerations may not be related to diagnosis and care, but instead privacy, consent for the use of samples, and why their child may need to be screened in the first place."

—Public health professional

"For a mandatory, non-consented NBS program (the current status quo in the US) the purpose should be to identify diseases that have a presymptomatic phase and a treatment that is so highly safe and effective that there can be no reasonable decision to not identify and treat. The benefit should be focused on the affected infants to markedly improve outcomes because they are giving up autonomy and agency. Parental benefit is assumed, but should not be a priority or focus to avoid conflict of interest in decision making (eg, a parent's religious beliefs should not trump the infant's opportunity for effective treatment to prevent death or intellectual disability)."

—Health researcher

"We pride ourselves on saying screening is universal but sadly access to timely, high quality treatment is not. States are pressured by advocates and others to add what is on the RUSP when they really do not have the infrastructure in state or locally to treat the individuals identified."

—Newborn screening follow-up professional

"We have major concerns about the public's trust in storage of genomic data and we've already seen a spike in refusals in the last two years. We will need to figure out how to ensure that this data is protected carefully and how to convey this to the public so that they have confidence in us."

—Newborn screening lab professional

"Please emphasize the importance of data privacy protections for strengthening trust in newborn screening systems. As excitement grows for sequencing the genomes of newborns, we will lose the public trust in NBS without assurances that newborn genomes can't be accessed by law enforcement."

—Health researcher

Research and family advocacy have created a clear and often used runway for researching and adding new conditions as long as the treatment is pharmaceutical and has the financing of that industry. But for disorders without industry backing (aka those with cheap over-the-counter treatments) this is not the case of course. And the input of relevant (aka treating) specialists on whether a disorder is appropriate candidate for screening is demonstrably not taken into account

—Healthcare provider

"I think the process for adding new conditions prioritizes disease advocates and their families. Lab staff don't have the manpower and money to get all these new instruments, to validate, create, and test all of these conditions, at least not where I work. Things in the lab have become overwhelming because it feels like any condition is getting added to the RUSP. We need to focus on adding currently treatable conditions, that have immediate impacts after birth. I think there needs to be a way to slow down the massive influx of tests being added to the RUSP recently, because we are struggling to keep up."

—Newborn screening lab professional

"The next evolution in newborn screening is going to be sequencing. Using sequencing technology. We've been bound or limited by tandem mass spectrometry, which is only limited inborn errors of metabolism…Most conditions are not inborn errors of metabolism that affect infants and children…. There's therapies for these conditions now. And there's a way to identify [infants] before they become symptomatic … I can give you numerous examples of children, where their lives were saved by newborn screening, the new newborn screening of sequencing."

—Health industry representative

# APPENDIX B: QUESTIONNAIRE

The National Academies of Sciences, Engineering, and Medicine (National Academies) Committee on Newborn Screening: Current Landscape and Future Directions invites you to participate in a questionnaire on current and future approaches to newborn screening in the United States.

This effort is sponsored by the U.S. Department of Health and Human Services with additional support from the Chan Zuckerberg Initiative. Learn more about the project.

The purpose of the questionnaire is to get input from people and organizations who are interested in, impacted by, or involved in newborn screening. This includes families, the rare disease community, NBS lab and follow-up professionals, payors, clinicians, the general public, and others.

Results of this questionnaire and other engagement activities taking place in Spring 2024 will be considered by the National Academies committee as it gathers information to develop its report. This report is expected to be released in spring 2025.

## Responding to this Questionnaire

This questionnaire should take approximately 10-15 minutes to complete and will remain open through May 21, 2024. Alchemer will save your responses as you complete the survey so you can return to complete the questionnaire by accessing the link provided to you.

Your participation in this questionnaire is voluntary. You may skip any question that you do not want to answer. You will be asked questions about newborn screening programs. In addition, we will collect certain demographic information to ensure the questionnaire results represent a diversity of perspectives. Your responses will be maintained securely and deleted six months after completion of the study. The National Academies will receive only aggregated data from Alchemer and not your individual responses.

If you have any questions about this questionnaire, please contact NewbornScreening@nas.edu

By clicking below you are confirming that you are at least 18 years of age and agree to participate in this questionnaire.

## About you

1. **Please select *all* of the following that describes you:** [multiple selections].

   a) Parent

   b) Parent of child with a rare disease or condition

   c) Person with a rare disease or condition

   d) Rare disease advocate

e) Health administrator

f) Healthcare provider

g) Health researcher

h) Health industry representative

i) Newborn screening lab professional

j) Newborn screening follow-up professional

k) Public health professional

l) Payor

m) Privacy advocate

n) General public

o) Other _____

2. **Now, which of the following best describes you?** [one choice]

a) Parent

b) Parent of child with a rare disease or condition

c) Person with a rare disease or condition

d) Rare disease advocate

e) Healthcare provider

f) Health researcher

g) Health industry representative

h) Newborn screening lab professional

i) Newborn screening follow-up professional

j) Public health professional

k) Payor

l) Privacy advocate

m) General public

n) Other _____

3. [for those that select health industry in Q1] **Which bests describes your health industry:**

a) Screening technology

b) Diagnostics

c) Therapeutics

d) Information technology

e) Other:

4. [for those that select in Q1 Parent of child with a rare disease or condition, Person with a rare disease or condition, Rare disease advocate] **Which rare disease(s) or conditions(s) are you impacted by or engaged with:** [open ended]

5. [for those that select healthcare provider in Q1] **What healthcare role best describes you?**

   a) Birth worker, midwife, doula

   b) Genetic counselor

   c) Nurse

   d) Advance Practice Provider (e.g., Physician Assistant or Nurse Practitioner)

   e) OB-GYN

   f) General pediatrician

   g) Pediatric subspecialist

   h) Family physician

   i) Other type of physician: (please specify)

   j) Other healthcare role:

6. **Where do you reside?**

   a) *List of all US States and Territories*

   b) Outside of the U.S.

[If "Outside of the U.S." is selected] **6b. Please specify your location outside of the U.S.**

[drop down menu]

## About newborn screening

Each state and territory in the United States has its own newborn screening program run by the state public health department. These programs make sure babies are screened for certain conditions before they leave the hospital. The goal is to help identify treatable conditions before a baby develops problems. Finding and treating these conditions early in babies' lives can prevent brain damage, physical disabilities, costly medical care, and death.

Newborn screening involves a series of tests: tests done on blood spots, hearing tests, and congenital heart tests. These tests occur in the hospital 24-48 hours after birth. **This questionnaire will focus only on the bloodspot tests**.

The federal government provides advice to states on a list of conditions to consider including in routine, public health newborn screening programs. Guided by this federal list (called the "recommended uniform screening panel"), each state chooses to screen for diseases and conditions based on the unique needs of their populations and other factors including legislative requirements, budget, workforce availability, and technological resources. The conditions being screened for tend to:

- Urgently need treatment as early in life as possible;
- Likely produce serious health effects if untreated; and
- Have an available medical treatment.

Within the newborn screening program itself, public health professionals play key roles in connecting babies identified through newborn screening with diagnostic testing, parent education, and follow-up care. Newborn screening programs also work with partners in a larger system that supports the identification, follow-up, and treatment of babies with these conditions. These partners include regulatory agencies, clinical care, advocacy groups, research, patients, and parents, among others.

**We are interested in your opinions and perspectives on newborn screening.** There is no right or wrong answer to each question. We invite you to share your responses below.

7. **Had you heard about newborn screening before taking this questionnaire?**

   a) Yes

   b) No

   c) I'm not sure

8. [for those that select parent or rare disease parent in Q1] **Did your child receive newborn screening?**

   a) Yes

   b) No

   c) I'm not sure

## Your views about the US newborn screening program

9. **From your perspective, what should be the main purpose of newborn screening?**
   [open answer]

10. **Which parts of today's newborn screening system are most effective at achieving this purpose? Choose up to three:**

    a) Parent education

    b) Bloodspot collection (collecting drops of blood from the infant for screening)

c) Laboratory screening facilities and/or staff

d) Follow-up on screening test results with families

e) Research to develop new screening tests

f) State administration of newborn screening programs

g) Federal guidance on conditions to be screened for

h) All babies are screened unless families choose not to participate (opt-out)

i) Process for adding new conditions to newborn screening programs

j) Other

k) None [skips to question 12]

11. **Looking at what you chose in the question above, how do these part(s) support the newborn screening system to be effective?** [open answer]

12. **What are the main challenges in today's newborn screening system? Choose up to three:** [randomize choices]

    a) Parent education

    b) Collection of blood from the infant to screen for disorders (bloodspot collection)

    c) Laboratory screening facilities and staff

    d) Follow-up on test results with families

    e) Research to develop new screening tests

    f) State administration of newborn screening programs

    g) Federal guidance on conditions to be screened for

    h) All babies are screened unless families choose not to participate (opt-out)

    i) Process for adding new conditions to newborn screening programs

    j) Other

    k) None [skips to question 14]

13. **Looking at what you chose in the question directly above, why are these part(s) a challenge for the newborn screening system?**
    [open answer]

## Community needs

Newborn screening programs have to balance their mandate to screen all the infants born in the US (approximately 3.6 million a year) with the individual needs of each baby.

14. **Which three of the following elements are most important to offer every infant and their parent/guardian?**

    a) Parent education about newborn screening

    b) Timely collection of bloodspots

    c) Timely return of screening test results

    d) Screening for all urgent conditions that have a medical treatment

    e) Screening for all serious conditions whether or not they have medical treatment

    f) Timely follow-up on screening results, for additional testing to confirm a diagnosis

    g) Connection to pediatricians and specialist care, if applicable

    h) Clear communication around uncertain results

    i) Access to affordable treatment

    j) Other

15. [for rare disease community] **What is your rare disease community's most important need regarding being able to identify and treat people affected by rare disease, and how do you think newborn screening could contribute to meeting this need?**
    [open text]

[FOR respondents that select: Payor in Q1]

16. **If newborn screening included a larger number of diseases, the number of identified babies seeking follow-up diagnostic testing or subsequent follow-up care will increase. How would you recommend newborn screening programs balance the costs and benefits involved?**
    [open text]

17. **Building on the question above, what information would be most important to your decision-making about medical coverage for conditions added to newborn screening?**
    [open text]

## Use of Residual Dried Blood Spots

Residual blood spots are dried blood spots that are 'left over' after all screening tests have been completed. These samples are not labeled with the name of the child – this is called deidentified. These samples are used to improve newborn screening for future infants, including performing regular quality assurance to make sure the screening tests consistently produce accurate and consistent results. These samples also can be used for other types of research. Regulations for this research varies state by state.

*As a thought experiment, please share your views based on the following situations:*

18. **If researchers want to use leftover, deidentified biospecimens from adult patients (e.g., from a blood draw) for medical research to improve patient care:**
[select one]

    a) It is fine to do so without notification or consent of the individual

    b) The individuals should be notified but not asked for their consent

    c) The individuals should be notified *and* asked for their consent

    d) Something else: _____

19. **If researchers want to use residual, deidentified newborn bloodspots for research directly related to newborn screening (e.g., develop a new screening test for a condition that could later be included in public health newborn screening programs):**
[select one]

    a) It is fine to do so without parental/guardian notification or consent

    b) Parents/guardians should be notified but not asked for their consent

    c) Parents/guardians should be notified *and* asked for their consent

    d) Something else: _____

20. **If researchers want to use residual, deidentified newborn bloodspots for other types of medical research unrelated to newborn screening:**
[select one]

    a) It is fine to do so without parental/guardian notification or consent

    b) Parents/guardians should be notified but not asked for their consent

    c) Parents/guardians should be notified *and* asked for their consent

    d) Something else: _____

## Trade Offs

In planning the future of newborn screening, there is a lot to consider. The following questions ask for your perspective on balancing different priorities.

21. **As a thought experiment, if the US newborn screening system could only do one of the following, which would you recommend:**

    a) Put more government funding towards screening newborns for conditions not currently included as part of newborn screening programs

    b) Put more government funding towards providing treatment to children who are diagnosed with a condition because of newborn screening

    c) Put more government funding towards improving the performance of our current screening program (e.g., reducing follow-up time, improving accuracy of screening for diverse populations)

    d) Other:

22. **Why did you choose this answer?**
    [open text]

23. **The federal government recommends (but does not require) the conditions that should be included in newborn screening. States and territories choose which conditions to include in their program to address the needs of their populations and comply with other factors including legislative requirements, budget, workforce availability, and technological resources. Which statement bests reflects your perspective?**
    [select one]

    a) Every newborn in the United States and territories should receive the same newborn screening tests

    b) Each state/territory should be able to choose the tests to include in their newborn screening

    c) Other:

24. **Why did you choose this answer?**
    [open text]

[FOR All respondents]

## Planning for the future

25. **What factors are most important for strengthening trust in the newborn screening program?** Choose up to three:

    a) Education about newborn screening (its purpose and how and why it is conducted)

    b) Clear communication about the goal of newborn screening

    c) Clear communication about the meaning of results

    d) Clear communication about how leftover blood spot samples might be used

    e) Data privacy protections

    f) Program transparency and accountability

    g) Other

26. **What advice do you have for the National Academies committee examining the US newborn screening system, particularly about how to improve it for the future?**

[open text]

27. **Did you feel you had enough information to answer the questions in this survey?**

    a) Yes

    b) No

    c) I'm not sure

## About you

The following questions help us understand who we are hearing from. Results from this section of the questionnaire will also inform our outreach efforts, so we can hear from diverse voices across the newborn screening experience and areas of expertise.

You can skip any question. Your responses will be kept anonymous.

28. **How would you describe the area where you currently live?**

    a) Urban

    b) Mostly urban

    c) Mostly rural

    d) Rural

    e) I prefer not to answer

29. **What is your gender identity?**

    a) Woman

    b) Man

    c) Non-binary person

    d) I prefer to self-describe _____

    e) I prefer not to answer

30. **Do you have lived experience as a trans person (meaning your gender identity does not align with your sex assigned at birth)?**

    a) Yes

    b) No

    c) I prefer not to answer

31. **Are you of Hispanic, Latino/a/e/x, or of Spanish origin?**

    a) Yes

    b) No

    c) I prefer not to answer

32. **Are you of Middle Eastern or North African origin?**

    a) Yes

    b) No

    c) I prefer not to answer

33. **What is your racial background? (Select all that apply)**

    a) American Indian or Alaska Native or Indigenous or Native American

    b) Asian or Asian American

    c) Black or African American

    d) Native Hawaiian or Other Pacific Islander

    e) White

    f) None of the above describe me – I identify as ____

    g) I prefer not to answer

34. **What is the highest level of education that you have completed?**

a) Some high school or less

b) High school diploma or GED

c) Some college, but no degree

d) Associates or technical degree

e) Bachelor's degree

f) Graduate or professional degree (MA, MS, MBA, PHD, JD, MD, DDS, etc.)

g) I prefer not to answer

## APPENDIX C: QUESTIONNAIRE RESPONDENT DEMOGRAPHICS

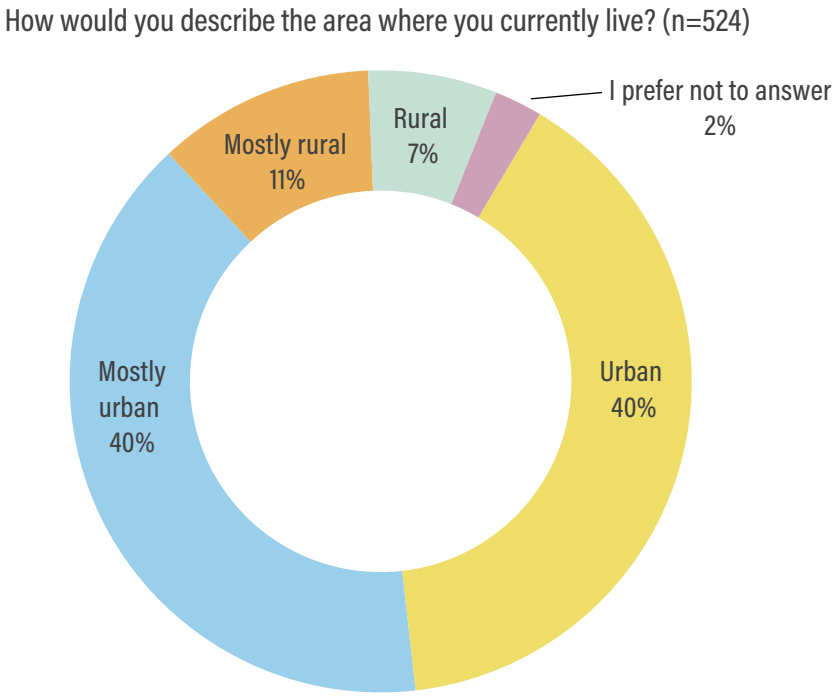



*Figure 15. Questionnaire respondents' area of residence based on self-description.*

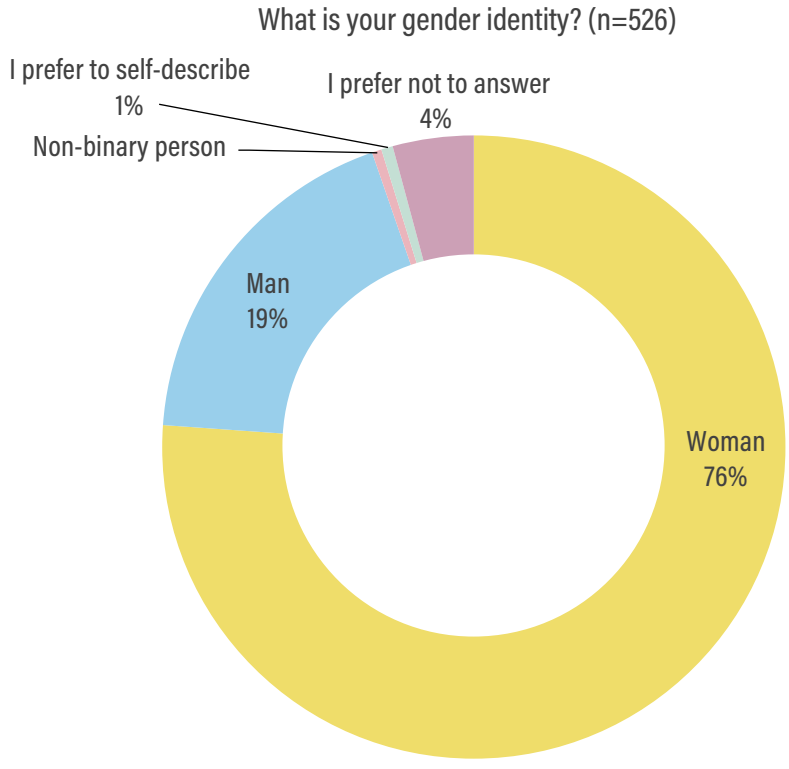



*Figure 16. Questionnaire respondents' gender identity.*

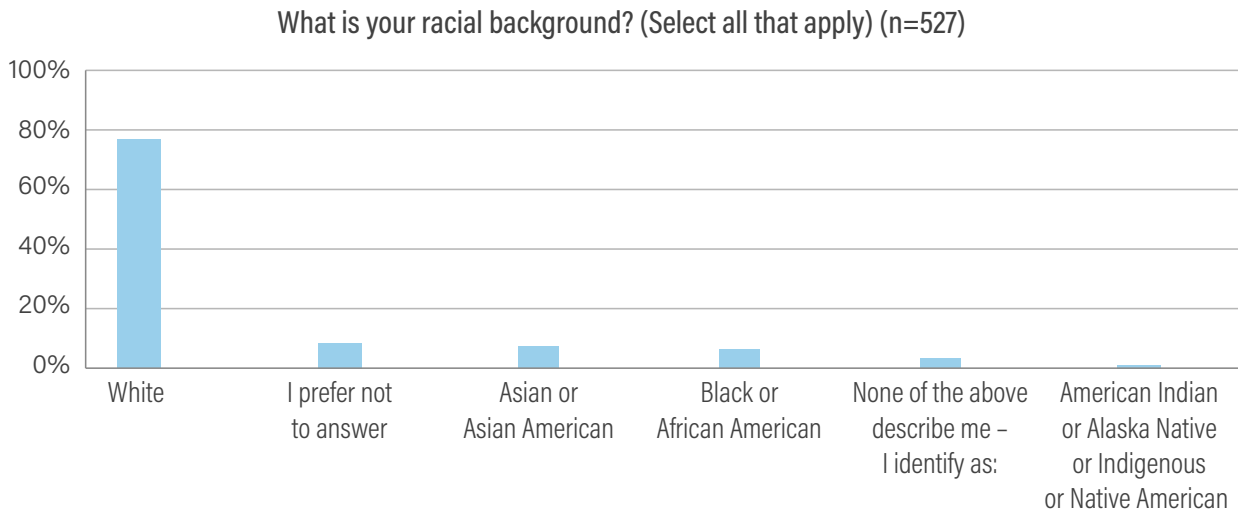



*Figure 17. Questionnaire respondents' racial background.*

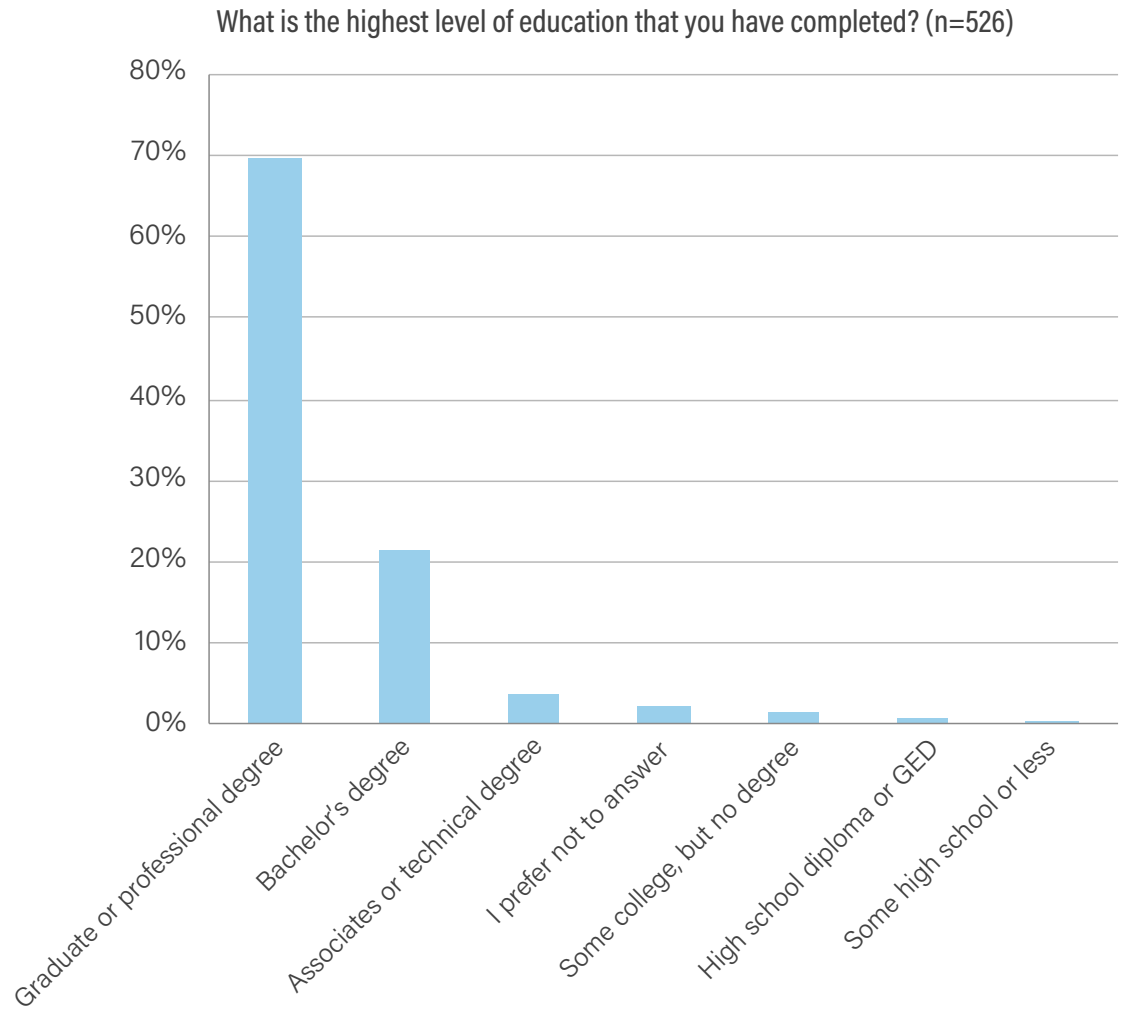



*Figure 18. Questionnaire respondents' highest level of education.*

# APPENDIX D: QUESTIONNAIRE INVITATION

Newborn screening (NBS) programs nationwide identify babies at risk of serious, but treatable conditions, that aren't otherwise found at birth. They enable doctors to diagnose conditions quickly and start treatment as soon as possible. Recently, Congress directed the Dept. of Health and Human Services' Office on Women's Health to commission a study with the National Academies to identify key state and federal actions that could help to modernize newborn screening programs. Learn more about the project.

**The National Academies would like to hear from you:** *How can we strengthen today's newborn screening programs? What changes would you like to see in the future?*

Please share your ideas about NBS programs in this questionnaire

The purpose of the questionnaire is to hear from people and organizations interested, impacted, or involved in newborn screening, and learn their ideas about current and future approaches to NBS in the United States.

Results of this questionnaire and other engagement efforts this spring will be carefully considered by the committee as it develops recommendations for Congress. For details on data collection, please see the questionnaire landing page.

This questionnaire should take 10-15 minutes to complete and will remain open through May 21, 2024 (EDT). [LINK]

Please share this questionnaire with others using the text and link below.

Thank you for completing the questionnaire

## APPENDIX E: LISTENING SESSION PARTICIPANT FEEDBACK

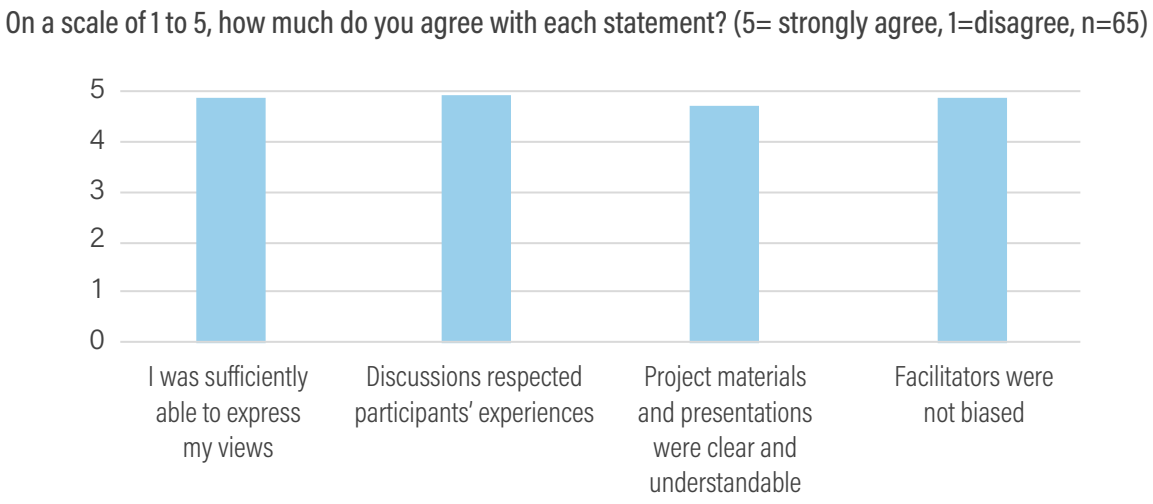



*Figure 19. Listening session exit poll results.*

# APPENDIX F: LISTENING SESSION INVITATION

NBS programs touch almost every family in America, identifying babies at risk of serious but treatable conditions. This committee will report to Congress on both short-term options to strengthen existing NBS programs as well as a vision for the next 5-15 years. Learn more about the project.

**The National Academies would like to hear from you:** *How can we strengthen today's newborn screening programs? What changes would you like to see in the future?*

Please join us for an online listening session in early May 2024. Each 2.5-hour listening session will emphasize hearing from participants, through both plenary and small group discussions.

- Families (general).
  Sunday May 5: 1pm Eastern / 10am Pacific
- Rare disease patients, families and advocacy organizations.
  Saturday May 11: 1pm Eastern / 10am Pacific
- Newborn screening professionals.
  Wed May 1: noon Eastern / 9am Pacific
- Health administrators, payors, and healthcare industry.
  Monday May 6: noon Eastern / 9am Pacific
- Healthcare providers.
  Monday May 13: noon Eastern / 9am Pacific

Click here to register for the listening session that best fits your perspective. Please note that registration is limited and a waiting list will be established as needed.

Input received through the listening session and the committee's questionnaire [LINK] will be carefully considered by the committee as it develops recommendations for Congress.

Download our "What to Expect: NBS Listening Sessions" document for important information about your participation including: format, technology requirements, accessibility needs, participation guidelines, a waiting list, data collection and reporting, and how we will protect your privacy.

For more information about the project, questions, or if you can't attend the session for your sector, please contact [contact person].

# APPENDIX G: LISTENING SESSION AGENDA

**Committee on Newborn Screening: Current Landscape and Future Directions**
Listening Session Agenda
**Healthcare Providers** | Monday, 13 May, 2024
12:00pm-2:30pm Eastern Time | On Zoom

## Purpose

This session is being held by the National Academies committee on Newborn Screening: Current Landscape and Future Directions to learn from people with lived and professional experience with newborn screening, with a focus on bloodspot testing.

This session will explore both how we can strengthen current newborn screening programs in the short-term, and what is needed for the future of this program.

## Agenda

**12:00** **Welcome & introductions**

**Presentation: Newborn screening: current landscape and future directions**
National Academies of Sciences, Engineering, and Medicine
*In your experience, what is working well about newborn screening in the U.S. today?*

**Breakout discussion: Challenges and inequity in newborn screening**
*What challenges do you see – or experience – with newborn screening? Do you have any ideas on how to respond to these challenges?*
*What disparities do you see in newborn screening? How might newborn screening help address long-standing health inequities?*

**Plenary: Share back**

**Break** (10 minutes)

**Breakout discussion: Adding conditions to screening & uncertain results**
*What do you think should guide whether a disease is added to the newborn screening program?*
*How should the newborn screening system respond to screening results that are hard to interpret, or have unclear clinical consequences?*

**Plenary: Reflection**
*What advice do you have for the National Academies committee charged with strengthening current and future newborn screening systems in the US?*

**Next steps & evaluation**

**2:30** **Adjourn**

## Results

Input received from these listening sessions and the online questionnaire will be analyzed by Susanna Haas Lyons Consulting and reported publicly later this year. The results will be provided to the study committee to carefully consider as they develop a report on their work, which will be published in 2025.

The committee will recommend both short-term options to strengthen existing newborn screening programs as well as a vision for the next 5-15 years.

**How to Prepare**

We want to hear your perspectives about newborn screening. Please bring your ideas to the questions listed on the first page of this agenda.

Please plan to join for the full session. These 2.5-hour interactive discussions will primarily be held in small group discussion so you can share your views and hear from others. Facilitators and notetakers will support your discussions.

## Technology Requirements

We recommend that you use a computer with speakers or headphones for the best experience of this event. Some interactivity and accessibility features are not available when using a smartphone or tablet.

**Listening Session Participation Guidelines**

- Please plan to participate for the full duration of the listening session.
- Respect the opinions of others. Every participant brings information, points of view, and ideas to contribute.
- We strive to ensure the safety of participants and speakers. There will be zero tolerance for those who promote violence against others on the basis of race, ethnicity, national origin, sexual orientation, gender identity, religious affiliation, or different ability.
- Respect the privacy of participants—do not share what is said in your listening session with other people.
- Share opportunities for airtime equally. If you've asked a question or shared a comment, ensure that new voices are heard before you contribute again.
- Practice self-care: if you need to get up or take a break, please feel free.

# APPENDIX H: LISTENING SESSION: WHAT TO EXPECT

## Overview

The National Academies of Science, Engineering, and Medicine (National Academies) Committee on Newborn Screening: Current Landscape and Future Directions is hosting online listening sessions to understand various perspectives about America's newborn screening programs. In addition to these listening sessions, the committee is also learning from interested and impacted people through a questionnaire, and comments submitted through the project website. Learn more here.

This effort is sponsored by the Department of Health and Human Services Office on Women's Health in response to a Congressional request. Supplemental funding was provided by the Chan Zuckerberg Initiative to enable enhanced community engagement.

Five listening sessions are being held in spring 2024, for the following groups:

- **Newborn screening professionals**. Wed May 1: noon Eastern / 9am Pacific. For lab directors and staff, follow-up directors and staff, and others involved in newborn screening testing.
- **Rare disease patients, families and advocacy organizations**. Sunday May 5: 1pm Eastern / 10am Pacific. For individuals and families impacted by rare diseases, as well as people involved in organizations that advocate for people with rare diseases.
- **Health administrators, payors, and healthcare industry**. Monday May 6: noon Eastern / 9am Pacific. For those who administer state newborn screening programs, organizations that pay for healthcare services, and businesses that develop tests, devices, procedures, therapies, etc. for newborn screening and associated results.
- **Families (general)**. Saturday May 11: 1pm Eastern / 10am Pacific. For families whose babies did, or didn't, participate in newborn screening.
- **Healthcare providers**. Monday May 13: noon Eastern / 9am Pacific. For clinical care providers involved in newborn screening including pediatricians, neonatologists, disease specialists, and nurses.

## Listening Session Format

Please plan to join for the full session. These 2.5-hour interactive discussions will invite you to share your views on key topics such as:

- Strengths of today's newborn screening program in America
- Changes to newborn screening programs that might better address health inequities
- How the program can better respond to results of newborn screening that are uncertain
- Advice for the National Academies committee, particularly about how to strengthen future screening programs

The majority of the listening session will be held in small group discussion so you can share your views and hear from others. Facilitators and notetakers will support your discussions.

## Technology Requirements

To engage with this online event, you will need a computer (laptop or desktop), tablet, or smartphone, with speakers or headphones.

We recommend that you use a computer for the best experience of this event. Some interactivity and accessibility features are not available when using a smartphone or tablet.

## Accessibility

The focus groups will be held online, with closed captioning available. Discussion materials will be sent in advance.

If there is anything we can do to better accommodate your participation, please contact [contact]. Advance notice is necessary to arrange for some accessibility needs.

## Waiting List

If your desired session is full, please join the waiting list, and we will notify you if a registration space becomes available.

## Participation Guidelines

Please plan to participate for the full duration of the listening session.

- We strive to ensure the safety of participants and speakers. There will be zero tolerance for those who promote violence against others on the basis of race, ethnicity, national origin, sexual orientation, gender identity, religious affiliation, or different ability.
- Respect the opinions of others. Every participant brings information, points of view and ideas to contribute.
- Respect the privacy of participants—do not share what is said in your listening session with other people.
- Share opportunities for airtime equally. If you've asked a question or shared a comment, ensure that new voices are heard before you contribute again.
- Practice self-care: if you need to get up or take a break, please feel free.

Your participation in this focus group activity is voluntary, and you may withdraw your acceptance at any time via an email to [contact]. Additionally, if, for any reason, you want to leave the focus group before the scheduled finish time, please use the chat function to send an individual Zoom message to [contact].

## Data Collection and Privacy

You will be asked at the beginning of the listening session to consent to an audio recording of the session. Your participation is voluntary, and you can decline to comment on any topic that is put to the group for discussion. The audio recording and a transcript prepared by National Academies staff will be stored on password-protected Academies servers until the end of the calendar year. Those who will have access to this meeting recording and transcript will include National Academies staff and the engagement consultant, Susanna Haas Lyons Consulting. Your identity will be kept confidential, and you will not be identified or quoted without your express permission in any publicly available report.

Your responses will be included in a publicly available thematic summary and analysis of input on newborn screening programs in America, but will not attribute any response to you without your express permission. Susanna Haas Lyons Consulting will conduct this analysis and draft this report and it will be shared with the National Academies committee examining Newborn Screening: Current Landscape and Future Directions and made publicly available.

SUSANNA
HAAS LYONS