Rare Disease Company Coalition

#OneRareVoice

Equitable access to newborn screening can ensure that all babies have the best chance for a healthy life.

Join us in working towards a modern newborn screening system that provides equal access to a timely diagnosis for all babies.

Understanding newborn screening (NBS)

Newborn screening is a vital and proven public health program that screens approximately four million U.S. newborns each year. In the first 24 to 48 hours of a baby's life, a small blood sample is taken to detect serious, often fatal, genetic conditions that can be treated if diagnosed early.

How newborn screening works

Newborn screening is a system that involves federal and state governments, public health labs, researchers, and patient advocates. To simplify a very lengthy and complex process, the NBS program at a glance includes:

- Research pilots provide critical data for considering new conditions for federal and state screening panels.
- The Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns (ACHDNC) maintains and updates a list, called the Recommended Uniform Screening Panel (RUSP), detailing which conditions should be screened for by states.
- States determine the conditions to include on their screening panels.
- Public health laboratories conduct screening.
- States provide genetic confirmation and follow-up care including genetic counseling and medical guidance.

Keeping pace with Innovation

Therapeutic innovation & NBS

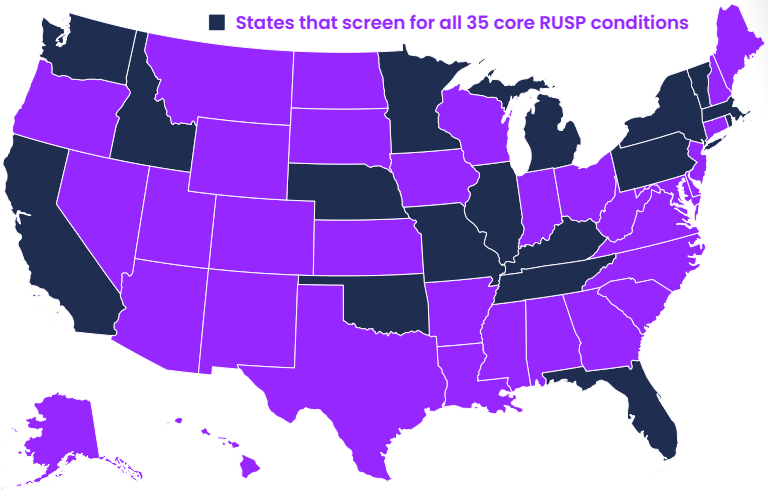
A wave of innovative therapies in development will constitute a disruptive event for which the NBS system is currently unprepared. Modernization of the system will be necessary to ensure that NBS can keep pace with medical innovation.[1]

Current challenges to the NBS System

Limitations of the current system can cause significant delays between the availability of transformative therapies and comprehensive screening, putting children at risk for preventable-injury or death.

- **Pilot and data process:** An effective intervention is necessary to justify NBS, but it is challenging to study new treatments in babies and children without the early diagnosis NBS provides.

1 Bailey DB, Porter KA, Andrews SM, Raspa M, Gwaltney AY, Peay HL. Expert Evaluation of Strategies to Modernize Newborn screening in the United States. *JAMA Netw Open*. 2021;4(12):e2140998. doi:10.1001/jamanetworkopen.2021.40998



The majority of states **do not** screen for all 35 core RUSP conditions

- **Federal reviews:** Adding new conditions to the 35 RUSP is a slow process. In the last 16 years, only 6 new disorders have been added and, the capacity of the ACHDNC to add new conditions is limited to 2 conditions per year.
- **Slow state uptake:** State implementation of new conditions varies widely. In some cases, it has taken a decade for all 50 states to begin screening for conditions on the RUSP.
- **Inequitable access to care:** Major discrepancies exist between the screening panels in each state resulting in life-threatening disparities in access to treatment.
- **Lack of resources:** Funding, staffing and equipment are barriers for state programs and labs to begin screening for new conditions.

Modernizing newborn screening in the U.S.

A recent study conducted by researchers at RTI International, a non-profit research institute, in 2021 calls for systemic change and modernization of the NBS system with a cross-section of forty NBS experts putting forward feasible and effective solutions for change. These recommendations fall into the categories of better data, increased alignment among states and federal agencies, expanded capacity for genetic technologies and increased funding. This will require stakeholder engagement and community consensus to build a modern NBS system that will keep pace with medical innovations and advancements. In the immediate term, several challenges need to be overcome.

Proposed recommendations to support and advance U.S. newborn screening

- **Reauthorize the newborn screening Saves Lives Act and state appropriations:** Ensuring the current system can continue to perform its life-saving work through the NBS reauthorization and clear state appropriations pathways.
- **Increase state funding:** Supplying state labs with the funding and resources necessary to add new conditions through federal appropriations and state support.
- **Improve awareness:** Increasing understanding of the life-saving benefits and complex challenges of the NBS System through improved stakeholder education.
- **Promote equality:** Curtailing the rare disease diagnostic odyssey, which can take on average 5-7 years and up to 8 doctors to receive an accurate rare disease diagnosis, by promoting equal access to NBS for conditions with a therapeutic intervention.

Rare Disease Company Coalition

We are dedicated to being a reliable and trusted resource for our leaders in Congress and the Administration, and a productive partner to rare disease-minded industry, academic and patient groups, by educating on the issues and opportunities that affect rare disease companies and advancing our shared mission to improve the lives of people living with rare diseases.

For more information, please visit www.rarecoalition.com or contact info@rarecoalition.com.
Follow us on Twitter at @RareCoalition.