Newborn Screening and Rare Disease: Opportunities and Challenges of Modernization

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Newborn Screening is Out-of-Step with Today’s Science
Need to Incorporate Genetic Screening, Elevate Patient Voice, Eliminate Patchwork “Death-by-Zipcode”

- WGS performed post-mortem on 45 patients who died as infants
- 31% were found to have a genetic disease that was not diagnosed prior to death
- 57% of these genetic diseases have an effective treatment
Building the Case for Change

Landmark 2021 study
**Expert Evaluation of Strategies to Modernize Newborn Screening in the United States** demonstrated consensus for change across the entire NBS system

![Magnitude of Change](image)

Move the slider below to reflect your attitude about the magnitude of change required to the NBS system to permit the timely inclusion of 30 new conditions for which there are transformative therapies available in the newborn period.
Road to Modernize Newborn Screening

2019-2022
Consensus Paper and Roundtables

2023
Set Stage for Congressional Action

2022-2024
Support Cutting Edge Projects

Rady Children’s, San Diego

North Carolina

EVERYLIFE FOUNDATION FOR RARE DISEASES

BEGIN NGS
NEWBORN GENOMIC SEQUENCING

EarlyCheck

PIONEERING THE NEW ERA OF NEWBORN SCREENING
Collaborative Insights and Recommendations for Modernizing NHS Systems

TRAVERE THERAPEUTICS
Four Key Themes for Modernization

**THEME 1:** Increase federal leadership, accountability, and transparency within federal newborn screening programs

**THEME 2:** Establish a regional lab network that provides state NBS programs with the opportunity to work together to ensure efficient and faster addition of newborn screening conditions

**THEME 3:** Increase access to population-level data both before and after newborn screening to facilitate the development and adoption of newborn screening conditions to federal and state panels

**THEME 4:** Integrate next-generation, evidence-based neonatal sequencing into newborn screening in a manner that can be broadly implemented in all state newborn screening programs
What Does the Future Look Like?
*Once-in-a-Lifetime Chance to Save Untold Lives, Disability, and Suffering*

- 6-9x more conditions screened
- End “Death-by-Zipcode”
- Increased patient voice in approval process
- Families incorporating long-term genetic information into their health care

*With only 4m births/year, and relentless innovation in diagnostics and treatments, this is one public health challenge we can and must meet.*