



TRAVERE[™]
THERAPEUTICS

**Newborn Screening and
Rare Disease:
*Opportunities and
Challenges of
Modernization***

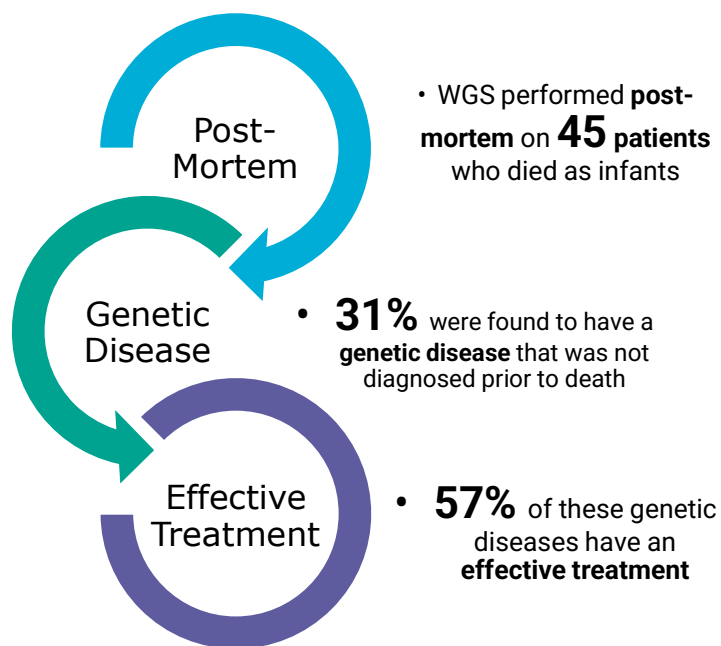
Christopher Porter,
Vice President,
Government Affairs and Policy





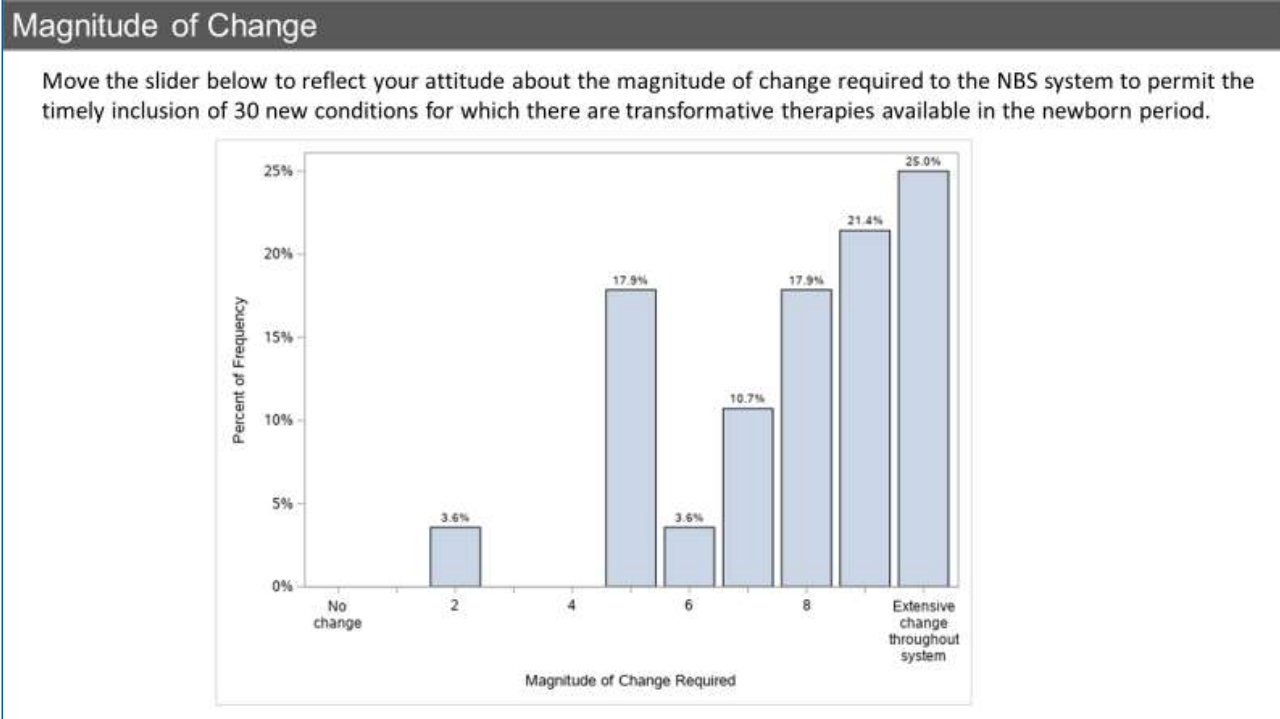
Newborn Screening is Out-of-Step with Today's Science

Need to Incorporate Genetic Screening, Elevate Patient Voice, Eliminate Patchwork "Death-by-Zipcode"



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



Road to Modernize Newborn Screening

2019-2022
Consensus Paper and Roundtables



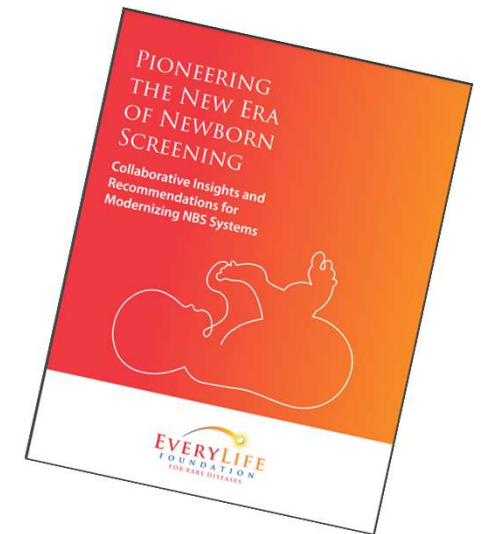
Rady Children's, San Diego



North Carolina

2022-2024
Support Cutting Edge Projects

2023
Set Stage for
Congressional Action





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.**