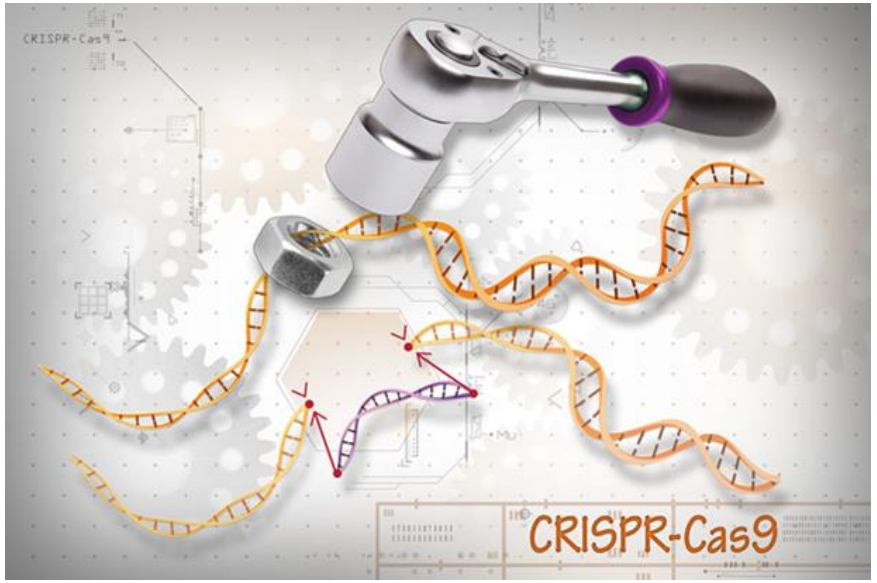
Newborn Screening: Current Status, Future Directions

Don Bailey, Ph.D

NCSL Maternal and Child Health Fellows Meeting

June 4, 2022



What is newborn screening?

- A highly successful state-based program with federal guidance and support
- Tests newborns for serious health problems that are not obvious at birth
- Usually done on dried blood spots
- Treating conditions BEFORE symptoms are obvious leads to better outcomes than treating AFTER symptoms emerge
- Essentially considered a medical emergency

Newborn screening policy in the U.S.

- More than 4,000,000 babies each year
- Now a well-established process for reviewing NBS nominations
- Rigorous review and recommendation by a national committee of experts
- DHHS Secretary determines whether a condition should be on the RUSP (Recommended Uniform Screening Panel)
- States decide whether and whento add the condition to their state panel
- 35 conditions on RUSP
- States are gradually implementing newly recommended conditions

HHS

Advisory Committee on Heritable Disorders in Newborns and Children

Many conditions will not make the RUSP in the near future

- 10 nominated but not initially recommended or sent to evidence review
  - SMA, Niemann-Pick, neonatal hyperbilirubinemia, Krabbe disease, hemoglobin H disease, Fabry disease, 22q11.2 deletion, GAMT, CTX, CMV
  - SMA reviewed and approved 10 years after initial nomination
  - GAMT re-nominated and recommended 6 years after initial nomination
  - Krabbe recently re-nominated 12 years after initial nomination
  - MPS-II recommended in 2020 after first nomination
- Major reasons for not recommending
  - No accurate and cost-effective screening test
  - Implementation too complicated or expensive
  - No pilot study data
  - No data on pre-symptomatic treatment efficacy
- Frustrations are evident
  - Advocates think the pace of expansion is too slow
  - States think the pace of expansion is too fast

Pending federal legislation calls for the National Academy of Medicine to prepare a report on "modernization of NBS"

Newborn Screening Saves Lives Reauthorization Act of 2021 (H.R. 482, S. 350)

SEC. 9. NAM REPORT ON THE MODERNIZATION OF NEWBORN SCREENING.

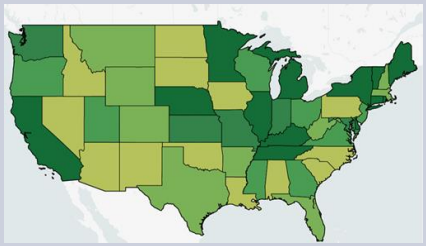
(a) STUDY.—Not later than 60 days after the date of the enactment of this Act, the Secretary of Health and Human Services shall seek to enter into an agreement with the National Academy of Medicine (in this section

WHY???

Four big challenges for newborn screening

How to get the data needed for a successful NBS nomination

Advisory Committee on Heritable Disorders in Newborns and Children

What to do about cross-state variability in screening	
How to incorporate genetic testing or sequencing	
How to prepare for new transformative therapies that must be administered early	

We asked experts about the need for change and possible solutions

JAMA Network | Open

Original Investigation | Pediatrics

Expert Evaluation of Strategies to Modernize Newborn Screening in the United States

Donald B. Bailey Jr, PhD; Katherine Ackerman Porter, MPH; Sara M. Andrews, MPH; Melissa Raspa, PhD; Angela Y. Gwaltney, PhD; Holly L. Peay, PhD

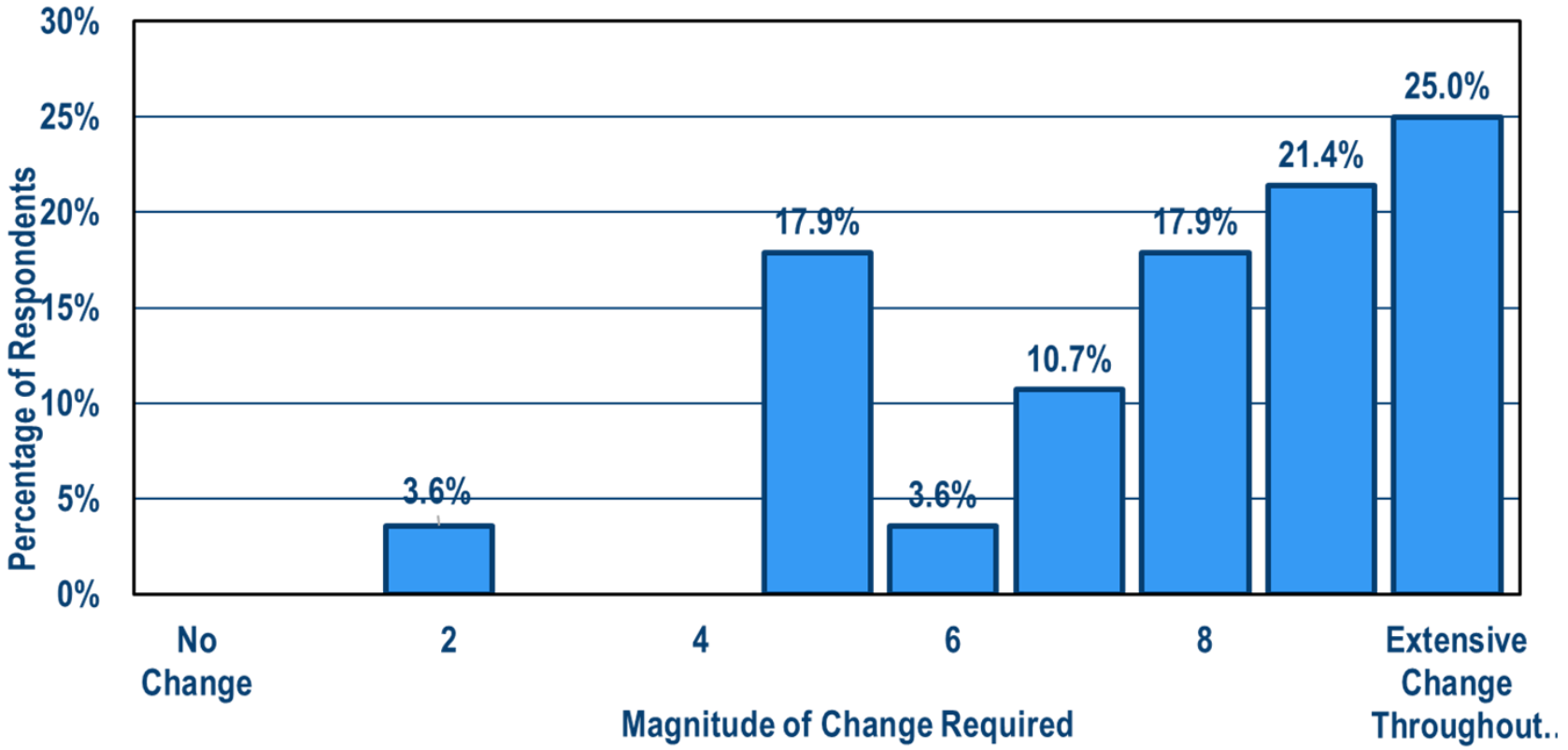
Andrews et al. BMC Pediatrics (2022) 22:90
https://doi.org/10.1186/s12887-021-03035-x

BMC Pediatrics

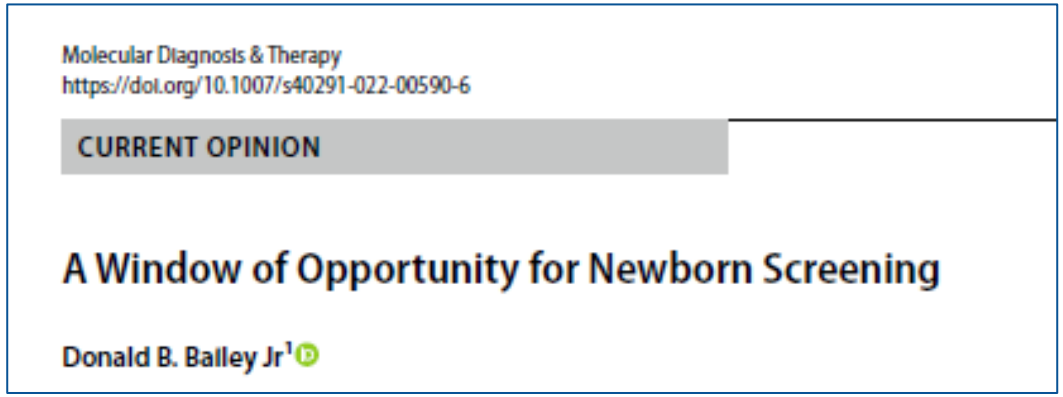
RESEARCH Open Access

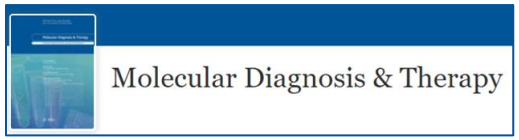
Preparing newborn screening for the future: a collaborative stakeholder engagement exploring challenges and opportunities to modernizing the newborn screening system

Sara M. Andrews†, Katherine Ackerman Porter†, Donald B. Bailey Jr and Holly L. Peay*



Building on this work and other experiences, I recently suggested five potential solutions to consider

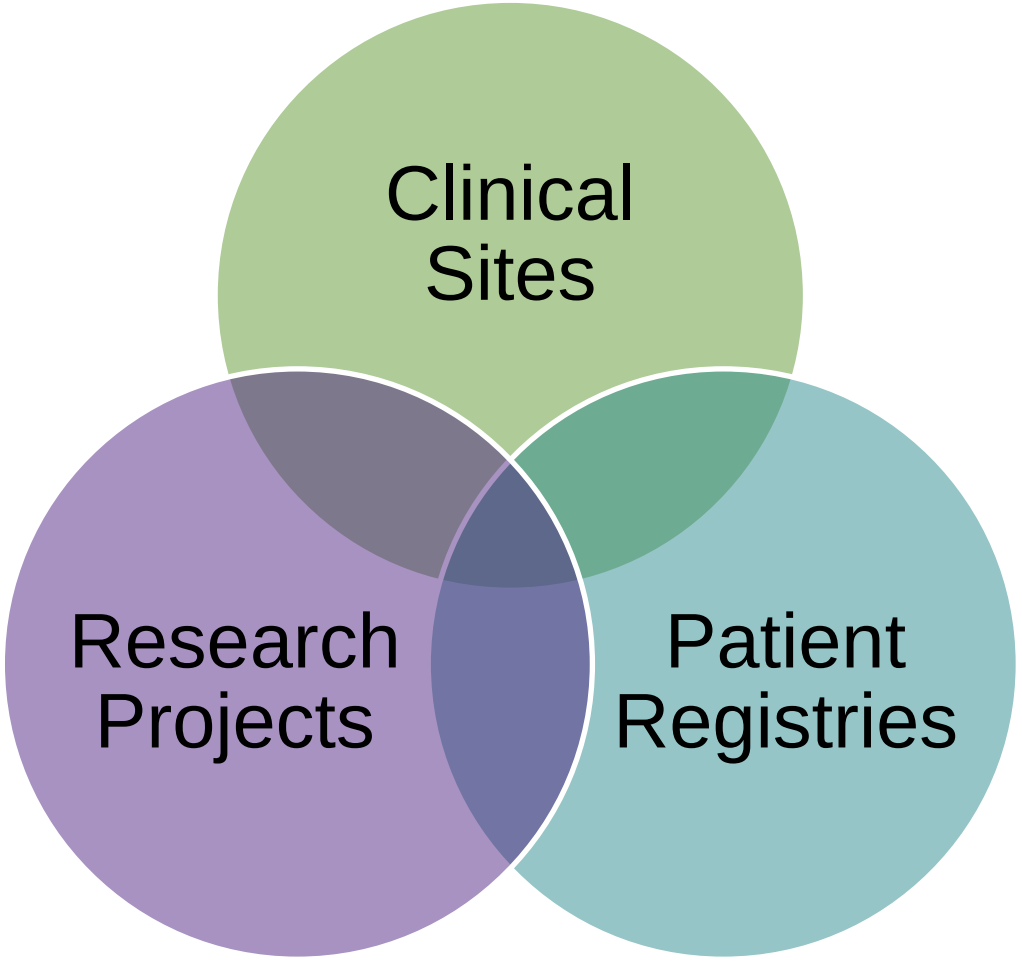




(2022)

- Build systems to integrate existing data
- Establish a national network of NBS research centers
- Create a network of regional NBS laboratories
- Establish a new stream of federal funding to states
- Integrate solutions

Build systems for more rapid integration of extant data



- Relevant data often exists in independent sites
- Merging data from multiple sites could often answer important questions
- States could help by
  - Partnering with research projects and clinical sites in their state
  - Working with other states to harmonize data or compare laboratory practices

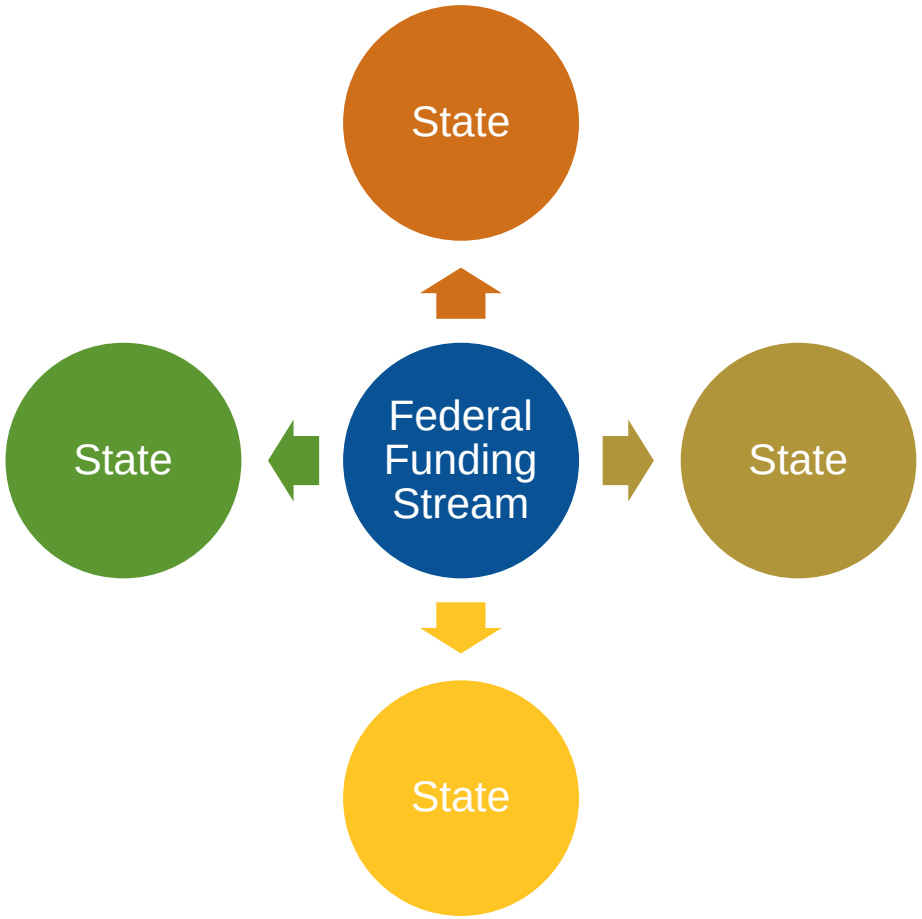
Create a national network of NBS research centers

- Coordinated research
- Common protocols and data
- Research to inform NBS policy
- Examples
  - Natural history studies
  - Clinical trials
  - Public acceptance
  - Alternative surveillance strategies
- States could help by
  - Partnering with the network to support prospective data collection
  - Sharing de-identified blood spots to answer questions about condition prevalence
  - Supporting consented pilot studies

Create a national network of specialized NBS laboratories

- Many state labs do not have ability to rapidly add new conditions
- A network of labs with cutting-edge equipment and expertise could provide a bridge to expedite screening until states are ready
- Network sites could also
  - Evaluate alternative screening methods
  - Partner with research centers on selected projects
  - Promote national harmonization of standards and practices
- States could help by
  - Using the network to accelerate adoption of new conditions
  - Sharing de-identified blood spots to support comparison of different laboratory procedures and cut-off scores
  - Partnering with other states to share data and conduct consented pilot studies

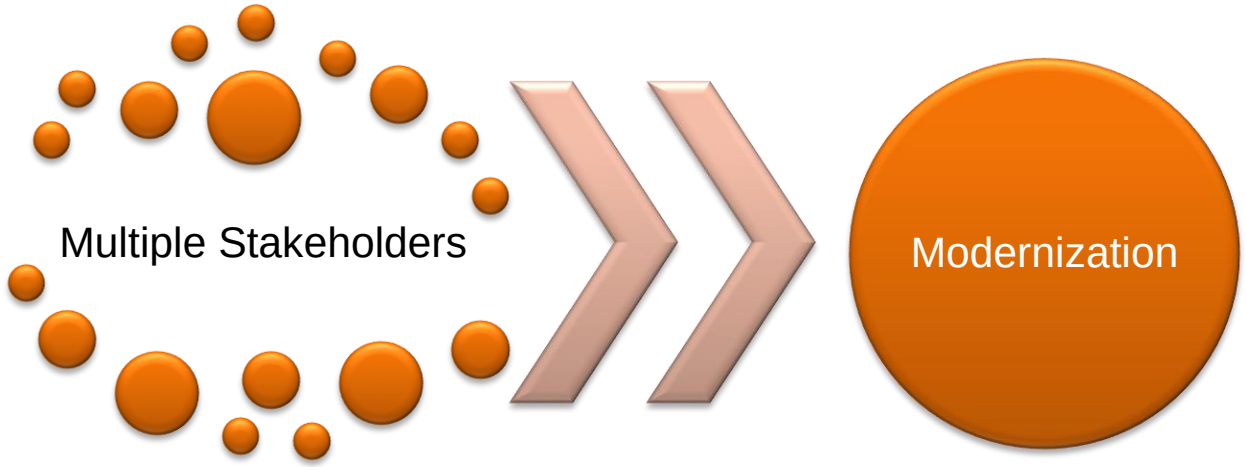
Establish a new stream of federal funding



- Funds to support expansion
- Formula based on # of births/year
- Establish benchmarks for state funding. For example:
  - Ability to add a new condition within 12 months
  - Adoption of nationally recommended cutoffs
  - Achievement of standardized quality indicators
  - Collection of long-term outcome data
  - Standardized data submitted annually
- Could lead to more national harmonization
- States could help by
  - Proactively contributing to policy development
  - Providing information on realistic benchmarks
  - Considering legislation to allow and support participation

Integrate solutions

- National-level leadership is needed
- How can we move from uncoordinated advocacy for a single disease, treatment, or technology to focus on a common set of goals?
- What does modernization success look like?
- States could help by
  - Identifying outcomes for modernization
  - Participating as a stakeholder in the national conversation
  - Supporting local efforts to develop and apply solutions



Who will lead?

What outcomes are we seeking?

Conclusion

- NBS is at a critical point, with a window of opportunity to envision a desired future state and do the hard work needed to achieve it.
- Change is inevitable if NBS as we know it today is to modernize itself to be NBS as we need it tomorrow.
- Incremental improvements over many years are unlikely to be sufficient.
- To advance NBS modernization, changes in research, policy, oversight, resources, and implementation are needed, many of which would require national legislation and visionary leadership.
- But because NBS is a state-based program, states will play an essential role:
  - Actively participating in discussions and contributing to solutions
  - Creating (when needed) new policies, legislation, or funding to complement federal funding
  - Building comprehensive follow-up systems
  - Collaborating with research centers, labs, and other states

Disclosures – Current Funding

Contact information

delivering **the promise of science**
for global good

Don Bailey
dbailey@rti.org
1-919-541-6488
www.earlycheck.org