

Building a Research Infrastructure to Support Newborn Screening Preparedness

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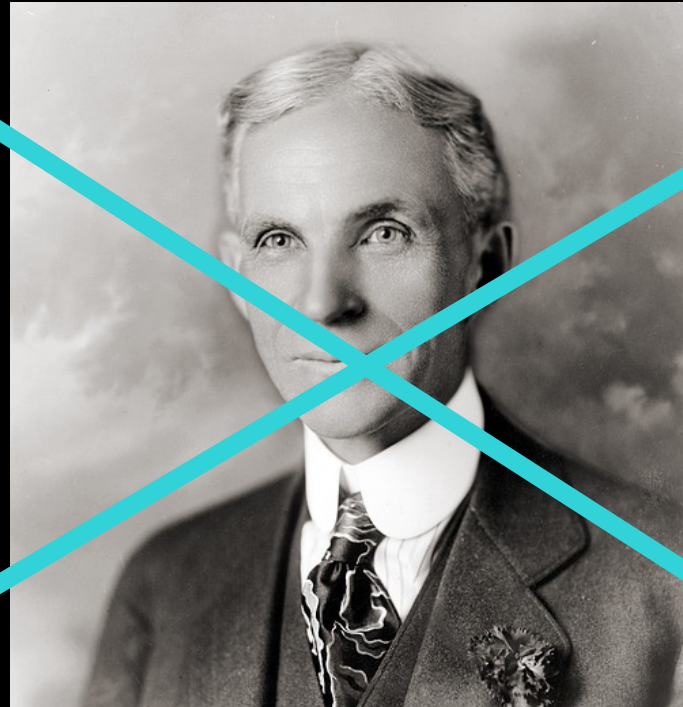


Disclaimers

- No Conflicts of Interest to Report.
- The perspectives of this presentation represents the presenter's view only.
- The findings and conclusions in this presentation are those of the authors.



On January 29, 1886, a patent for his “vehicle powered by a gas engine” was submitted by



Henry Ford

The Founder of the Ford Motor Company

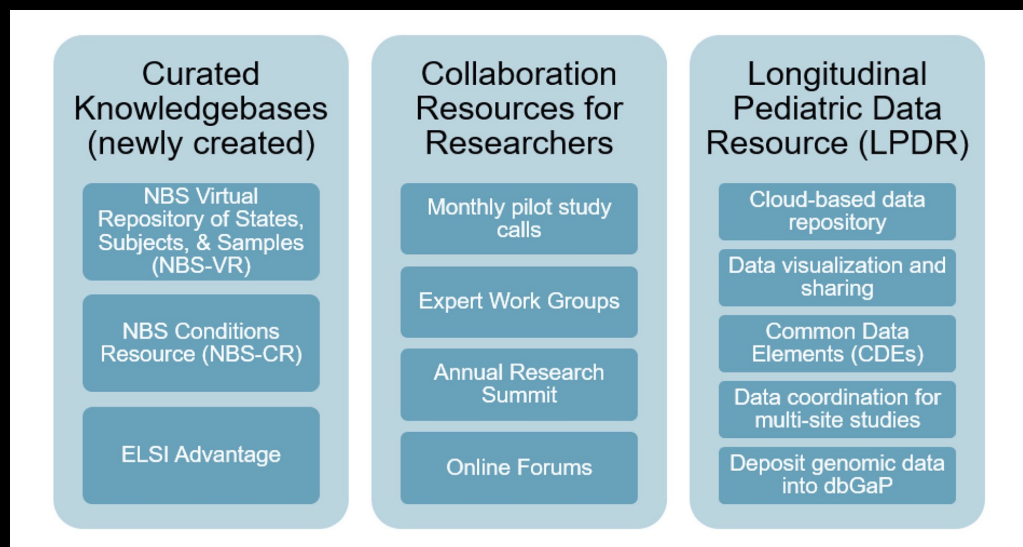
Karl Benz – the German inventor of the first car





Henry Ford made a dream **'affordable, accessible, and efficient'**.

Newborn Screening Translational Research Network (NBSTRN) Year 2008 – 2024



- Website and Social Media
- Tools and Resources
- Community of Stakeholders
- Dissemination
- RUSP nomination
- In person and Virtual Summit
- Work Groups



References on NBSTRN tools and resources:

- Minear, M. A., Phillips, M. N., Kau, A., & Parisi, M. A. (2022). Newborn screening research sponsored by the NIH: From diagnostic paradigms to precision therapeutics. *American journal of medical genetics. Part C, Seminars in medical genetics*, 190(2), 138–152.
- Chan, K., Hu, Z., Bush, L. W., Cope, H., Holm, I. A., Kingsmore, S. F., Wilhelm, K., Scharfe, C., & Brower, A. (2023). NBSTRN Tools to Advance Newborn Screening Research and Support Newborn Screening Stakeholders. *International journal of neonatal screening*, 9(4), 63.



Sequencing

New Conditions

AI

Machine Learning

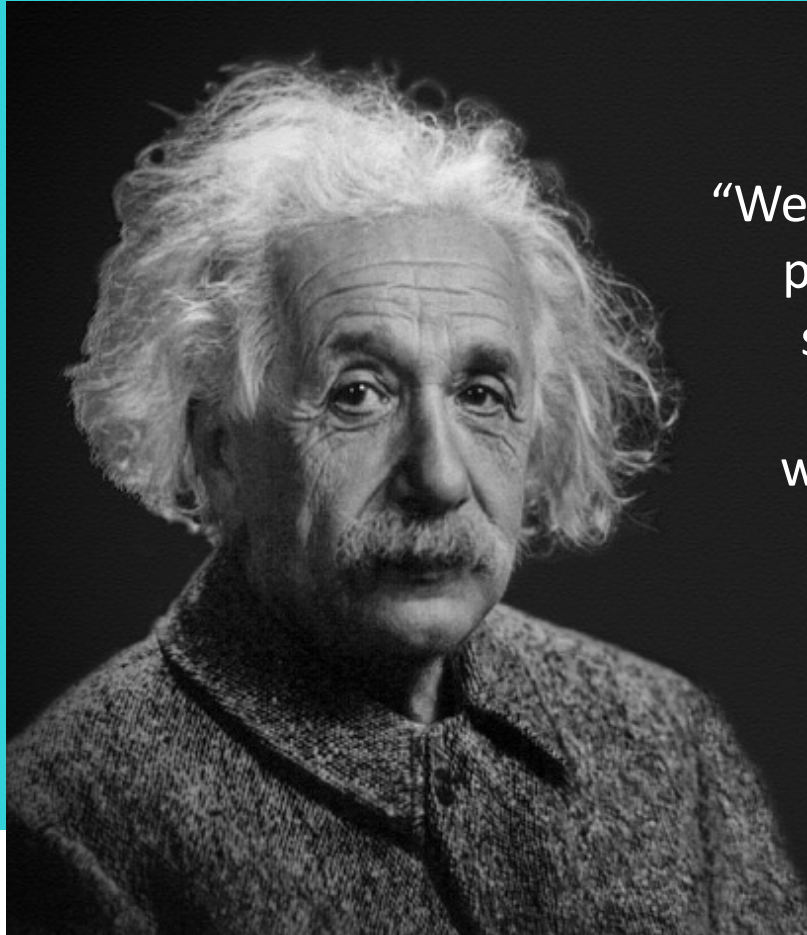
Healthcare Cost

Big Data

Road to RUSP

**New Treatment
(gene therapy)**

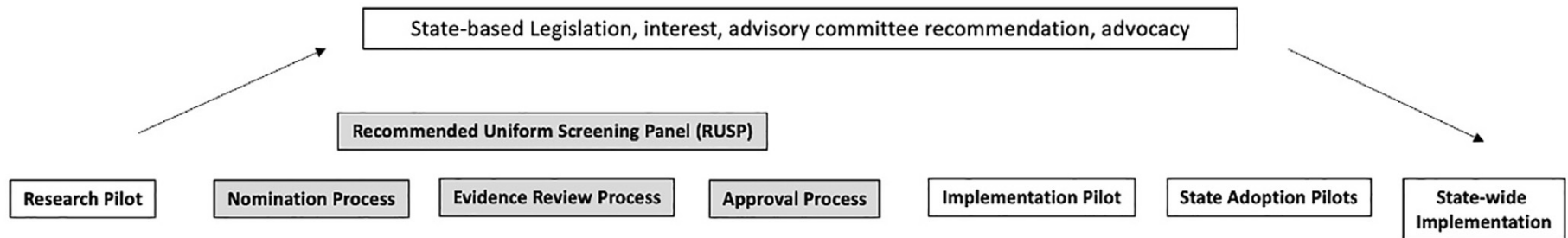
Perfect Storm = Perfect Opportunity



“We cannot solve our
problems with the
same thinking we
used when
we created them.”

-Albert Einstein

Pathway of Candidate Conditions



Chan, K., Brower, A., & Williams, M. S. (2022). Population-based screening of newborns: Findings from the newborn screening expansion study (part two). *Frontiers in genetics*, 13, 867354.

Research, Technological, and Infrastructure needs and solutions



A Review of Research Challenges

**Newborn
Screening
Preparedness**



- Lengthy Process in Research Pilots.
- System Capacity Constraints (RUSP nomination, state implementations).
- No Standard in Data Collection to Inform Policy and Implementation.
- Insufficient or Untimeliness of Resources Allocation.
- Capacity Building and Workforce Development in the NBS and Follow-up.
- RUSP Additions Expand the Definition of NBS.
- Miscommunication and Mistrust.
- Enhance Readiness for Artificial Intelligence and Machine Learning in NBS.

**Strategic
Solutions**



Andrews, S. M., Porter, K. A., Bailey, D. B., Jr, & Peay, H. L. (2022). Preparing newborn screening for the future: a collaborative stakeholder engagement exploring challenges and opportunities to modernizing the newborn screening system. *BMC pediatrics*, 22(1), 90.

Brower, A., Chan, K., Williams, M., Berry, S., Currier, R., Rinaldo, P., Caggana, M., Gaviglio, A., Wilcox, W., Steiner, R., Holm, I. A., Taylor, J., Orsini, J. J., Brunelli, L., Adelberg, J., Bodamer, O., Viall, S., Scharfe, C., Wasserstein, M., Chen, J. Y., ... Watson, M. (2022). Population-Based Screening of Newborns: Findings From the NBS Expansion Study (Part One). *Frontiers in genetics*, 13, 867337.

The Mission of National Preparedness

“....help people and communities to be more prepared by developing the capabilities needed to **prevent, protect** against, **respond** to, **recover** from, and **mitigate** against all threats and hazards. Whether we face risks related to earthquakes, cyberattacks or chemical spills, our goal is shared: **safety and resilience**.”

- Federal Emergency Management Agency



<https://www.fema.gov>

Newborn Screening Preparedness



Proposed Definition - Newborn Screening Preparedness

The condition of readiness with **processes, policies and platforms** in place for:

- **prevention** of death or disability of the affected child.
- **protection** of individual patient, families and society (such as ELSI).
- **risk mitigation** for harm or adverse effects.
- timely public health and clinical **response**.
- societal **recovery** to the expansion (cost) of population-based screening.

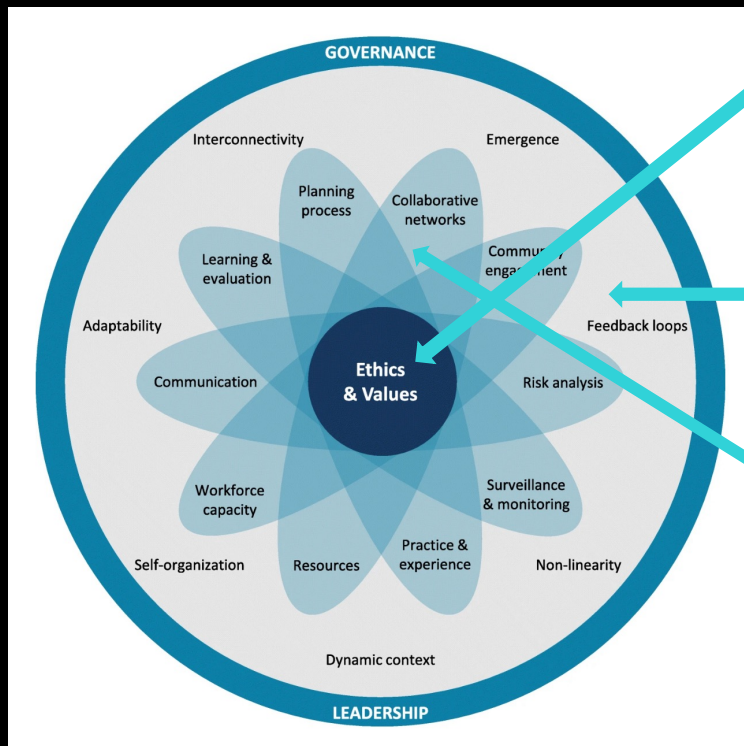
Goals – To promote safety and resilience of the NBS System.

Emergency Preparedness for Resilience Framework



Khan, Y., O'Sullivan, T., Brown, A., Tracey, S., Gibson, J., Génereux, M., Henry, B., & Schwartz, B. (2018). Public health emergency preparedness: a framework to promote resilience. *BMC public health*, 18(1), 1344.

Emergency Preparedness for Resilience Framework



- **Agree on the Ethics and Values based on Altruism**
- **Create “Emergency” Plans with Adaptive Models based on System Thinking**
 - Centers of Expertise (Stakeholders).
 - Asset Mapping of Networks with Technological Tools, Data Collection, and Resources.
 - Communication Channels for Fast Dissemination.
- **Multi-Site Coordinated Automation (Models, AI and Machine Learning) to Facilitate NBS Research, Pilot Research, and Clinical Medicine side-by-side.**
 - From Genome to Treatment (GTRx) at Rady’s Children
 - ED3N (CDC), ClinVar, ClinGen
 - Google DeepMind AlphaMissense

Khan, Y., O’Sullivan, T., Brown, A., Tracey, S., Gibson, J., G  n  reux, M., Henry, B., & Schwartz, B. (2018). Public health emergency preparedness: a framework to promote resilience. *BMC public health*, 18(1), 1344.

How to Build Research Infrastructure for NBS Preparedness in the next 5-15 years?

1. Strategic Planning with Stakeholders.
2. Apply the Concept of Design Thinking in NBS Research.
3. Asset Mapping of Tools, Resources, and Expertise.

↑ NBS Preparedness → ↓ NBS Challenges → ↓ NBS Solutions (Fixing)

Increasing resources and funding to support NBS Preparedness with a Resilience Framework may lead to less NBS Challenges, and thus reduce the need to fix the NBS System Problems.

BeginNGS Consortium



The newborn, parents and caregivers will always be at the center of every decision made by the consortium and include input from the patient's care team.

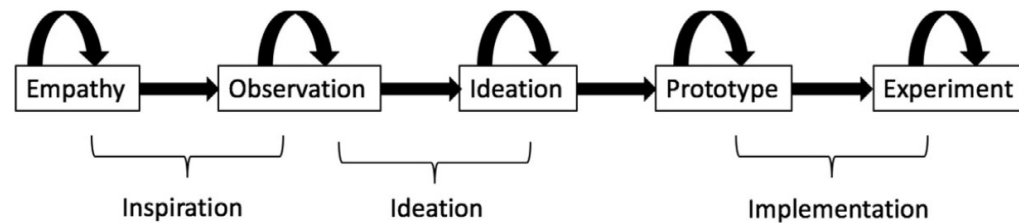
1. NBS Research Consortium

Key Stakeholders Engagement

- Parents, Caregivers, Health Professionals
- Life Science Research
- Diagnostics and Labs
- Health Systems
- Public and Private Payers
- Advocacy Organizations
- Federal and State Government
- Regulatory Bodies (FDA)
- Nonprofit and Research Organizations

Source: Kingsmore, S. F., & BeginNGS Consortium (2022). Dispatches from Biotech beginning BeginNGS: Rapid newborn genome sequencing to end the diagnostic and therapeutic odyssey. *American journal of medical genetics. Part C, Seminars in medical genetics*, 190(2), 243–256.

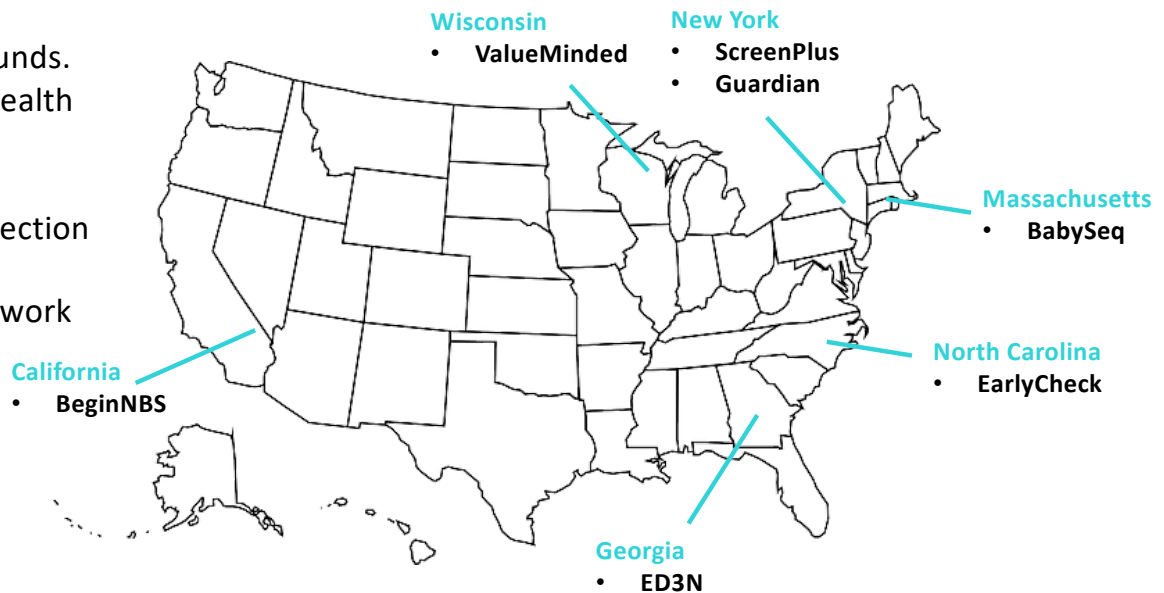
2. Design Thinking for Patient-Centered Platforms



Source: Chan, K. (2018). A design thinking mindset beyond the public health model. *World Medical & Health Policy*, 10(1), 111-119.

3. Assets Mapping

- States awarded with HRSA Propel funds.
- Global Alliance for Genomics and Health (GA4GH) - PhenoPackets
- NBS Genome Sequencing
- Enhancing Data-Driven Disease Detection in Newborns - ED3N (CDC)
- Rare Diseases Clinical Research Network (RDCRN) by NCATS
- Google Deep Mind AI
- Advocacy Groups
- Patients Registry
- NBS dried spot repository





Funding and Resources to Support the Pathway from NBS Research to Clinical Practice as an 'Affordable, Accessible, and Efficient' NBS System.

“Progress is a new season, and
the rule of progress is everything in its
season.”

- Henry Ford



How would you define Newborn Screening Preparedness?

The condition of readiness with **processes, policies and platforms** in place for:

- **prevention** of death or disability of the affected child.
- **protection** of individual patient, families and society (such as ELSI).
- **risk mitigation** for harm or adverse effects.
- timely public health and clinical **response**.
- societal **recovery** to the expansion (cost) of population-based screening.

Share your definition of 'NBS Preparedness' -

- Survey link:

<https://www.surveymonkey.com/r/NBSPreparedness>

- Interested in ValueMinded Consulting Services:

Email: keechanphd@gmail.com



Scan the QR code
for Survey