Longitudinal Follow-up for Infants and Children Identified Through Newborn Screening

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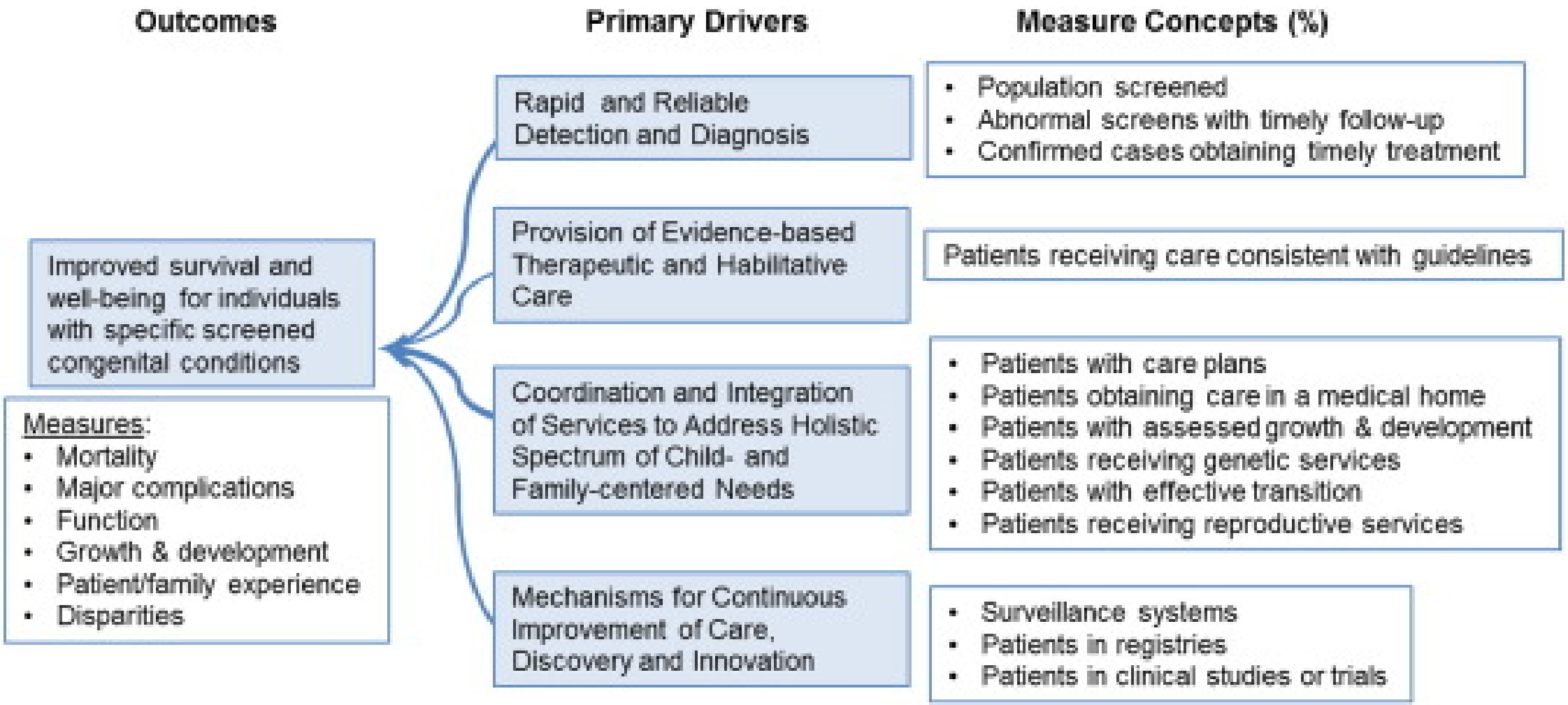
Advisory Committee on Heritable Disorders in Newborns and Children

ACHDNC Follow-up and Treatment Workgroup

- Workgroup Charge (2011): identify barriers, develop recommendations, and offer guidance on responsibility for post-screening implementation and short- and long-term follow-up, including treatment, relevant to newborn screening results
  - "Follow-up" implies treatment for clinicians; maybe not for others
  - "Longitudinal" better than "long-term" and implies "lifespan"
- Publications:
  - 2008 Core components and key features:
    - Care coordination, evidence-based treatment, quality improvement
    - Quality chronic disease management, Condition-specific treatment, care throughout lifespan
  - 2011 Perspectives: State and nation, primary/specialty providers, families
  - 2016 Framework for longitudinal follow-up
  - 2018 Specific quality measures

Framework for Assuring Good Outcomes from NBS



Hinton et al, 2016

Longitudinal Follow-up Starts with Nomination

- Nomination process should include an initial blueprint for longitudinal follow-up – start a conversation/relationship
  - Will infants identified by state NBS program have access to treatment? (e.g. equity, potential barriers)
  - What are the best outcome measures for the particular condition? (e.g. death, quality of life, ability to walk, does not require a ventilator, etc.)
    - Success of NBS: *did we meet the goals, fulfill the promise of NSB?*
  - What will be the (potential) process for obtaining population-level data?
    - e.g., patient registry
- Process should take into account variable resources
  - Nominating group presents a reasonable plan to answer the above questions
  - **Not** a "scored" criteria for adding the condition to the RUSP

Ideas 2019: “Federated System”

- Aug-Sep 2018 – Joe Schneider/Bob Ostrander preliminary proposals
- “Federated System” that *assures that every child identified with a NBS condition receives high-quality, evidence-based, family-centered care*
- Build a national network that can coordinate care and collect data in a standardized way (core outcomes or minimum data set)
  - Examples part of today’s panel discussion: LPDR (NBSTRN), NewSTEPS (APHL), CureSMA
  - Rare Diseases Clinical Research Network (NIH)
  - Region 4 Inborn Errors of Metabolism Information System
- Engage the EHR and AI industry as a current gap that could support more efficient data collection initiatives
- Financial resources for LTFU is a major gap; consider federal – state partnerships
- How best to learn about access to care after diagnosis and describing the barriers, especially using an equity lens?
- Help define who is responsible for follow-up at each stage (“road-map”)

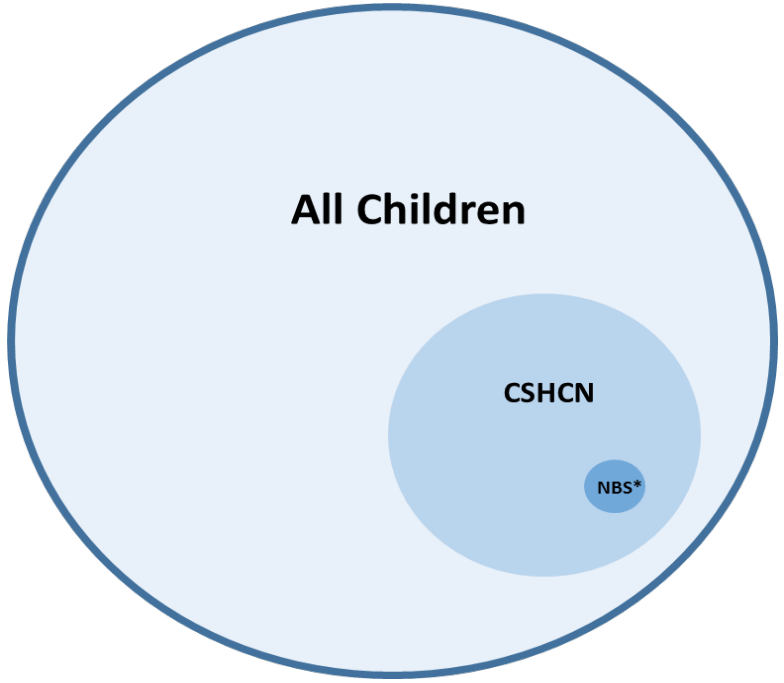
Why a Federated System? Varied goals.

1. Research
   - "What is the outcome of NBS for this condition?" (e.g. early treatment)
2. Quality improvement/assurance/return on investment:
   - "Did this child identified by NBS program get treatment? What was outcome?" (often a "yes/no" answer is sufficient)
   - "What is the impact of the NBS program on a condition(s)?" (population)
3. Clinical care
   - "How is a particular child doing? Getting all necessary treatment? What's the outcome/prognosis?"

- Overlap among all three; could be solved by a universal EHR

Why a Federated System? Varied interested groups.

- MCHB/Medicaid/state department of health
  - Assurance and equity for all children
- State Title V CSHN programs
  - Assurance and equity for CSHCN
- State NBS programs
  - Assurance and equity for "NBS" children
  - What are the limits of responsibility?
- Clinicians/researchers/family members
  - Individual child with an NBS condition
  - Of course, many feel greater responsibility



What Longitudinal Follow-up Information is Most Valuable for the ACHDNC?

- Evidence review models as a way of organizing later systematic review: How accurate was the prediction of benefits/harms? (lessons learned)
- Did everyone benefit from NBS? Equity, population health
- What is the condition? Range of diseases, secondary targets, late-onset, true prevalence, etc.
- Harms as a way to prioritize? "Red flags," how to define harms, health/psychosocial/costs/etc.; significant change in benefit/harm
- Barriers – systematic collection in common categories allows states and others to learn from each other; are barriers condition-specific?
- When/what conditions to review? Two-step process to set priorities.

State NBS: Equity in Diagnosis and Treatment

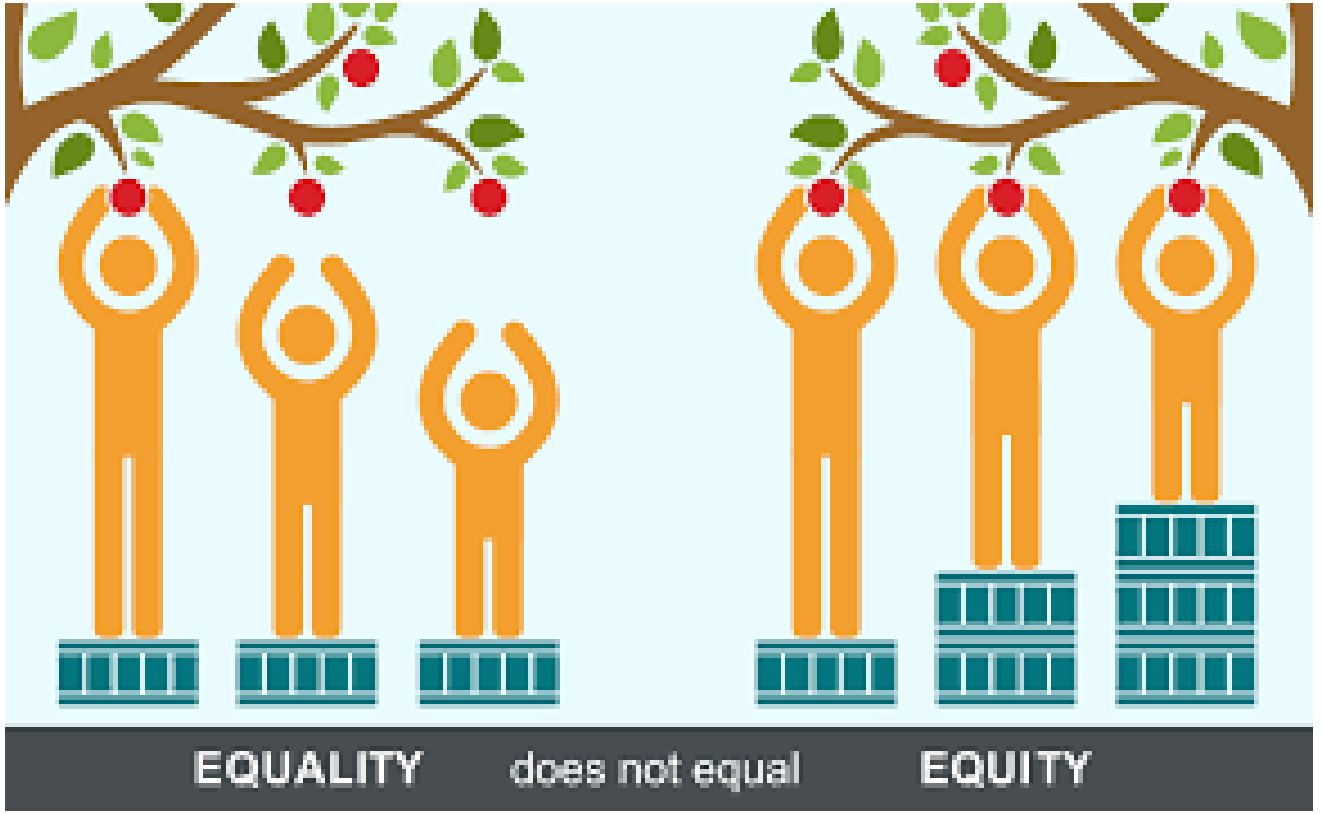
Diagnosis: e.g. racial/ethnic heterogeneity of SCID

Brosco et al, "Universal state newborn screening programs can reduce health disparities," *JAMA Pediatr* 2015

Treatment:

- antibiotics for SCD
- congenital hypothyroid guidelines: sub-optimal cognitive development
- PKU access to specialists and medical foods necessary to protect cognitive development

Kemper et al, "Ensuring the Life-Span Benefits of NSB," *Pediatrics* 2019



EQUITY: Redefine Cost of Treatment as Access to Care

The Definition of 'Access'

The WHO defines it as an interaction of different factors, which include availability, affordability, accessibility, appropriateness, acceptability, and quality.

Availability	A medical device is able to be purchased on the market. Also applies to functional medical devices that are physically available at health care facilities
Affordability	Medical device is a cost-effective option for both the patient and health care facility
Accessibility	Individuals are geographically within reach of health care facilities that house imaging technologies
Appropriateness	A medical device or imaging technology must be scientifically valid, address local need, and be utilized in a manner that a country can afford
Acceptability	Refers to cultural beliefs and individuals' attitudes regarding the use of various medical devices and imaging modalities
Quality	Based on the national regulatory standards that are in place to assure safe and effective use of all health technologies

State NBS Programs in the Age of Equity

"Sometimes you can't have everything in place to start. Re: equity. I think you may have used an example of a situation where only half kids can access treatment at launch of a new condition. Barriers are condition specific . . . OF COURSE children should have access to treatment for identified conditions. But access cannot mean exactly the same thing for every baby/family. The family from Fargo that has to travel to Minneapolis for their child's treatment and follow up appointments with specialists has a very different experience than I do (12 mile drive to U of M). People lose livelihoods over this sort of thing. My husband and I had to thousands out of pocket to stay in the only motel we could stay in during Eve's heart surgeries - even that would be completely impossible for many families. I guess what I am saying is, we can only try to level the playing field. It cannot be an excuse to delay implementing something that can help a portion of babies **until the rest of the investments in infrastructure** and more equitable access can happen. As you know, it often takes STARTING the process to spur equity."

State NBS Programs in the Age of Equity

- “The ‘cost’ of seeking care includes not just medical costs, but costs of transportation, lodging, child care, and loss of earnings from time off work. Lower SES is associated with cost barriers of that type and also psychosocial barriers of lower health literacy and familiarity and lower trust or communication differences with healthcare providers due to differences in social class and race/ethnicity, or language.”
- “Disparities in access are unfortunately the norm in this country.”

State NBS Programs in the Age of Equity

- We cannot expect state NBS programs, researchers, clinicians, family advocacy to solve all the problems of the US health care system, or issues of racism, economic inequality, etc.
- “On the other hand, NBS is a public health program, so that implies a greater obligation to meet the treatment needs of infants and children: ‘don’t tell me that my child has PKU then say you won’t help with the formula/foods.’ So while we can’t expect to solve all the inequity in the US with NBS, we probably need to go a bit further than we do right now.”

ACHDNC Follow-up and Treatment Workgroup

- Workgroup Charge (2011): identify barriers, develop recommendations, and offer guidance on responsibility for post-screening implementation and short- and long-term follow-up, including treatment, relevant to newborn screening results
- Potential next steps:
  - Continue to work with ACHDNC re specific tasks such as
    - Contribute to discussion re nominating process changes, etc.
    - Work with partners to draft a “roadmap” for a federated system
      - What conditions to review and when
      - What information most relevant to ACHDNC
      - Who is responsible for which information
    - Continue to discuss access and equity as an issue for NBS policy/practice