Follow Up and Treatment Workgroup Update

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Advisory Committee on Heritable Disorders in Newborns and Children

2021 Follow-up and Treatment Workgroup

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- Jeffrey P. Brosco, MD, PhD (FUTR Chairperson)
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- Annamarie Saarinen

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  Society for Inherited Metabolic Disorders
- Christopher A. Kus, MD, MPH (FUTR Co-Chair)
  Association of State & Territorial Health Officials
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  Child Neurology Society
- Robert J. Ostrander, MD
  American Academy of Family Practice Physicians
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- Marci Sontag, PhD
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MCHB

- Hannah Kotz
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FUTR Workgroup Charge (Revised September 2011)

Engage in a multi-step process that:

- Identifies barriers to post screening implementation and short- and long-term follow-up, including treatment, relevant to newborn screening results;
- Develops recommendations for overcoming identified barriers in order to improve implementation and short- and long-term follow-up, including treatment, relevant to newborn screening results; and
- Offers guidance on responsibility for post-screening implementation and short- and long-term follow-up, including treatment, relevant to newborn screening results.

“Follow Up” and “Treatment”

- “Follow up”
  - For clinicians, this implies treatment: when you “follow-up” with a patient, you are implying that you will be providing whatever treatment is indicated
  - Many non-clinicians hear “follow-up” as implying only data-gathering
  - Hence the word “treatment” a key part of the workgroup name
- “Long-term”
  - Different meanings for different organizations (5-year, 10-year, lifelong)
  - FUTR workgroup has decided to used the word “**longitudinal**”
    - From one year to “lifespan”

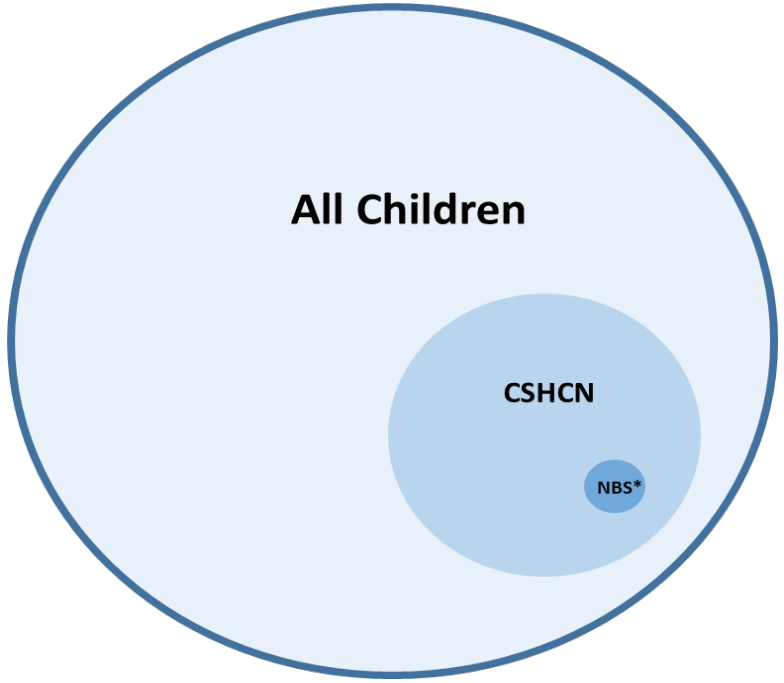
Examples of "Longitudinal Follow-up"

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

Who is the “we”? Some examples.

- 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



ACHDNC – Genetics in Medicine (2008)

Long-term follow-up after diagnosis resulting from newborn screening: Statement of the US Secretary of Health and Human Services' Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children

Alex R. Kemper, MD, MPH[1], Coleen A. Boyle, PhD[2], Javier Acéves, MD[3], Denise Dougherty, PhD[4], James Figge, MD, MBA[5], Jill L. Fisch[6], Alan R. Hinman, MD, MPH[7], Carol L. Greene, MD[8], Christopher A. Kus, MD, MPH[9], Julie Miller, BS[10], Derek Robertson, MBA, JD[11], Brad Therrell, PhD[12], Michele Lloyd-Puryear, MD, PhD[13], Peter C. van Dyck, MD, MPH[13], and R. Rodney Howell, MD[14]

- Central components
  - Care coordination
  - Evidence-based treatment
  - Quality improvement
- Features
  - Quality chronic disease management
  - Condition-specific treatment
  - Care throughout lifespan

ACHDNC – Genetics in Medicine (2011)

What questions should newborn screening long-term follow-up be able to answer? A statement of the US Secretary for Health and Human Services' Advisory Committee on Heritable Disorders in Newborns and Children

Cynthia F. Hinton, PhD, MPH[1], Lisa Feuchtbaum, DrPH, MPH[2], Christopher A. Kus, MD, MPH[3], Alex R. Kemper, MD, MPH[4], Susan A. Berry, MD[5], Jill Levy-Fisch, BA[6], Julie Luedtke, BS[7], Celia Kaye, MD, PhD[8], and Coleen A. Boyle, PhD, MS[1]

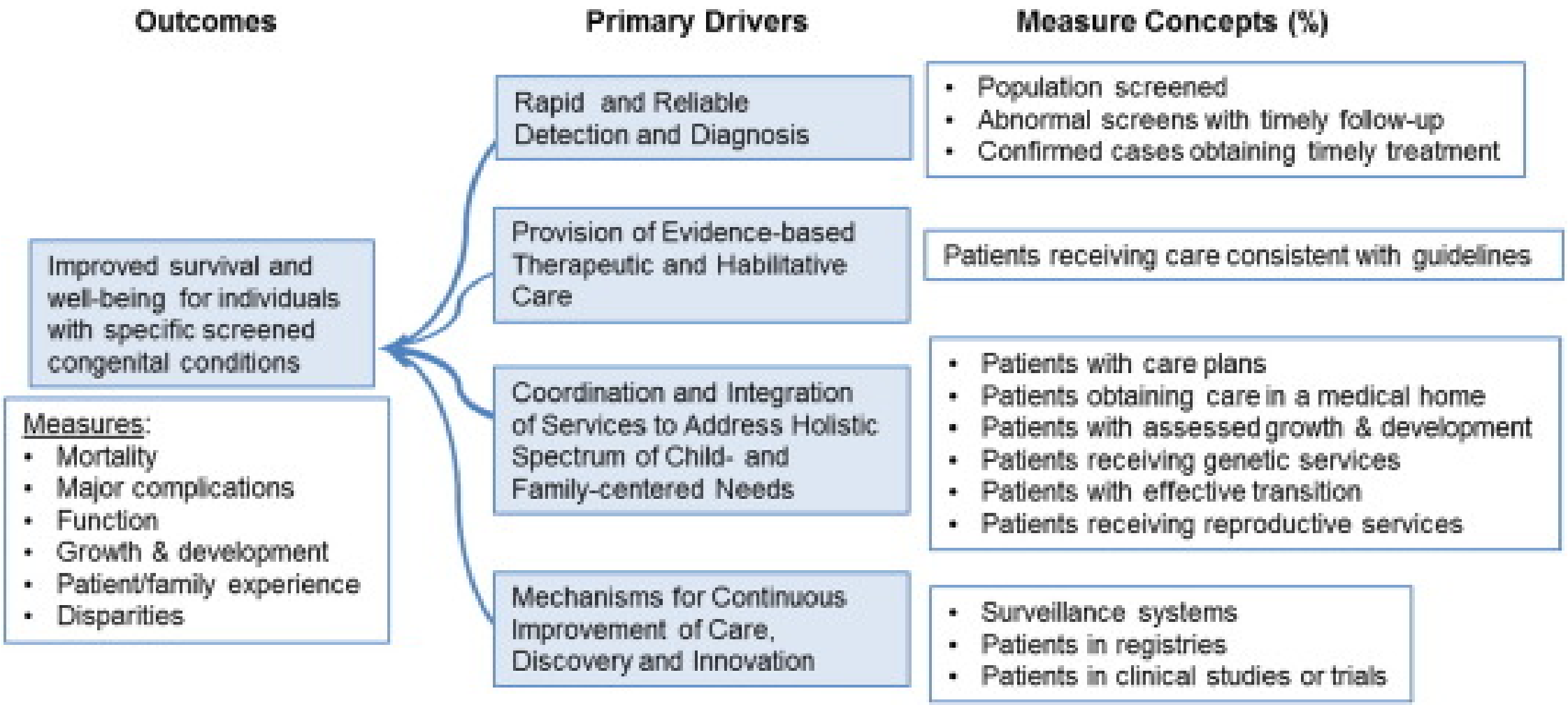
- Central components
  - Care coordination
  - Evidence-based treatment
  - Quality improvement
- Perspectives
  - State and nation
  - Primary/specialty providers
  - Families

ACHDNC – Molecular Gen & Metab (2016)

A framework for assessing outcomes from newborn screening: on the road to measuring its promise☆

Cynthia F. Hinton [a,*], Charles J. Homer [b], Alexis A. Thompson [c], Andrea Williams [d], Kathryn L. Hassell [e], Lisa Feuchtbaum [f], Susan A. Berry [g], Anne Marie Comeau [h], Bradford L. Therrell [i], Amy Brower [j], Katharine B. Harris [k], Christine Brown [l], Jana Monaco [m], Robert J. Ostrander [n], Alan E. Zuckerman [o], Celia Kaye [p], Denise Dougherty [q], Carol Greene [r], Nancy S. Green [s], the Follow-up and Treatment Sub-committee of the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC):

Framework for Assuring Good Outcomes from NBS



Hinton et al, 2016

The Role of Quality Measures to Promote Long-Term Follow-up of Children Identified by Newborn Screening Programs

Presented by the FUTR Workgroup to ACHDNC (February 2018)

- Quality measures are a crucial part of health and health care system
- Many different types of quality measures
- Creating/collecting data for these measures for NBS can be challenging
- Different perspectives needed, esp. patient/family/consumer
- Engage a broad range of stakeholders to
  - Identify a core set of long term follow-up quality measures and data resources
  - Encourage the use of large data collection activities (e.g NSCH) and QI activities (e.g. HEDIS)
  - Health Information Technology (HIT) standards/Clinical Decision Support (CDS) in the EHR

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)
  - Rare Diseases Clinical Research Network
  - 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
- Help define who is responsible for longitudinal follow-up at each stage (“road-map”)
- Financial resources for LTFU is a major gap/ federal – state partnership.
- How best to learn about access to care after diagnosis and describing the barriers, especially using an equity lens

Workgroup Discussion Questions

1. *What type of longitudinal follow-up information should be considered when a condition is added to the RUSP?*
2. *What type of information should be considered in a systematic review of conditions on the RUSP?*
3. *Should the cost of treatment be a factor in both the nomination process and the review of conditions on the RUSP?*

Q. # 1. RUSP Candidate Process (2019)

- When a new condition is considered, *we should be thinking about longitudinal follow-up from the beginning*
- Nomination process could include a "blueprint" for longitudinal follow-up
  - Will identified infants 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

Q. # 2 *What type of information to consider in a systematic review of conditions on the RUSP?*

- 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.

Q. # 3. Should the cost of treatment be a factor in nomination process and/or review of RUSP conditions?

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: 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