Follow-up and Treatment Workgroup Meeting Summary

Jeffrey P. Brosco MD PhD

ACHDNC Meeting, November 1-2, 2018

Follow-up and Treatment Workgroup

ACHDNC MEMBERS

- Jeffrey P. Brosco, MD, PhD (FUTR Chairperson)
- Susan A. Berry, MD
- Kamila B. Mistry, PhD, MPH
- Annamarie Saarinen

ORGANIZATION REPRESENTATIVES

- Debra Freedenberg, MD, PhD
  American Academy of Pediatrics
- Christopher A. Kus, MD, MPH (FUTR Co-Chair)
  Association of State & Territorial Health Officials
- Jed L. Miller, MD, MPH
  Association of Maternal and Child Health Programs
- **Robert J. Ostrander, MD**
  American Academy of Family Physicians

WORKGROUP MEMBERS

- Sabra A. Anckner, RN, BSN
- Amy Brower, PhD
- Christine S. Brown, MS
- **Kathryn Hassell, MD**
- Nancy Doan Leslie, MD
- **Sylvia Mann, MS, CGC**
- Dawn S. Peck, M.S., CGC
- Margie A. Ream, MD, PhD
- Joseph H. Schneider, MD, MBA, FAAP
- Janet Thomas, MD

FUTR Workgroup Meeting

- Quality Measures report – posted on website
- Medical Foods report – awaiting publication
- NBS Follow-up and Treatment Roadmap
  - Nov 2017 – May 2018 brainstorming
  - Aug-Sep 2018 - Schneider/Ostrander preliminary proposals
  - “Federated System” that assures that every child identified with a NBS condition receives high-quality, evidence-based, family-centered care
    - All children
    - Children with special health care needs
    - Children with a NBS condition
    - Individual conditions (e.g. sickle cell, CF, MCAD, etc.)

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 Aceves, 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]

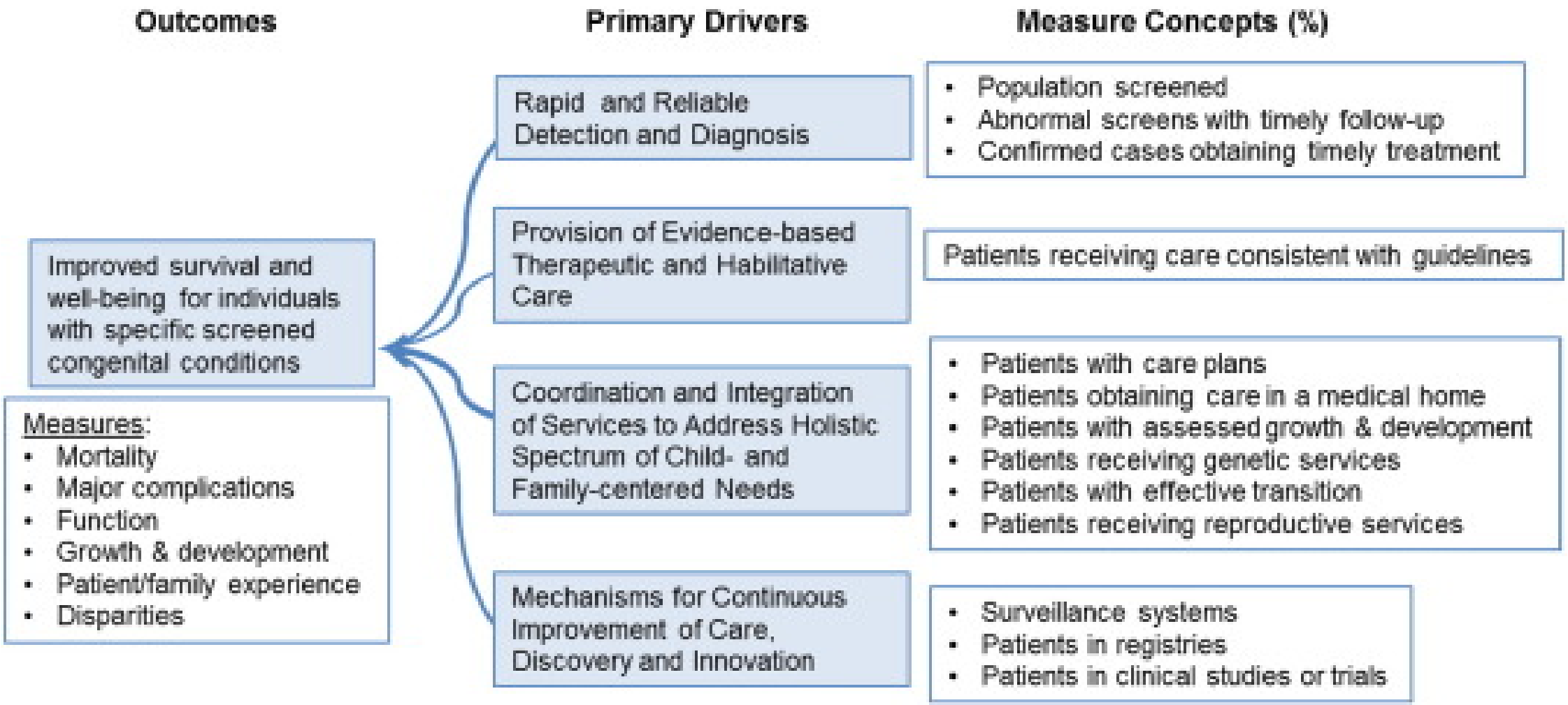
- Central components
  - Care coordination
  - Evidence-based treatment
  - Quality improvement
- Perspectives
  - State and nation
  - Primary/specialty providers
  - Families

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



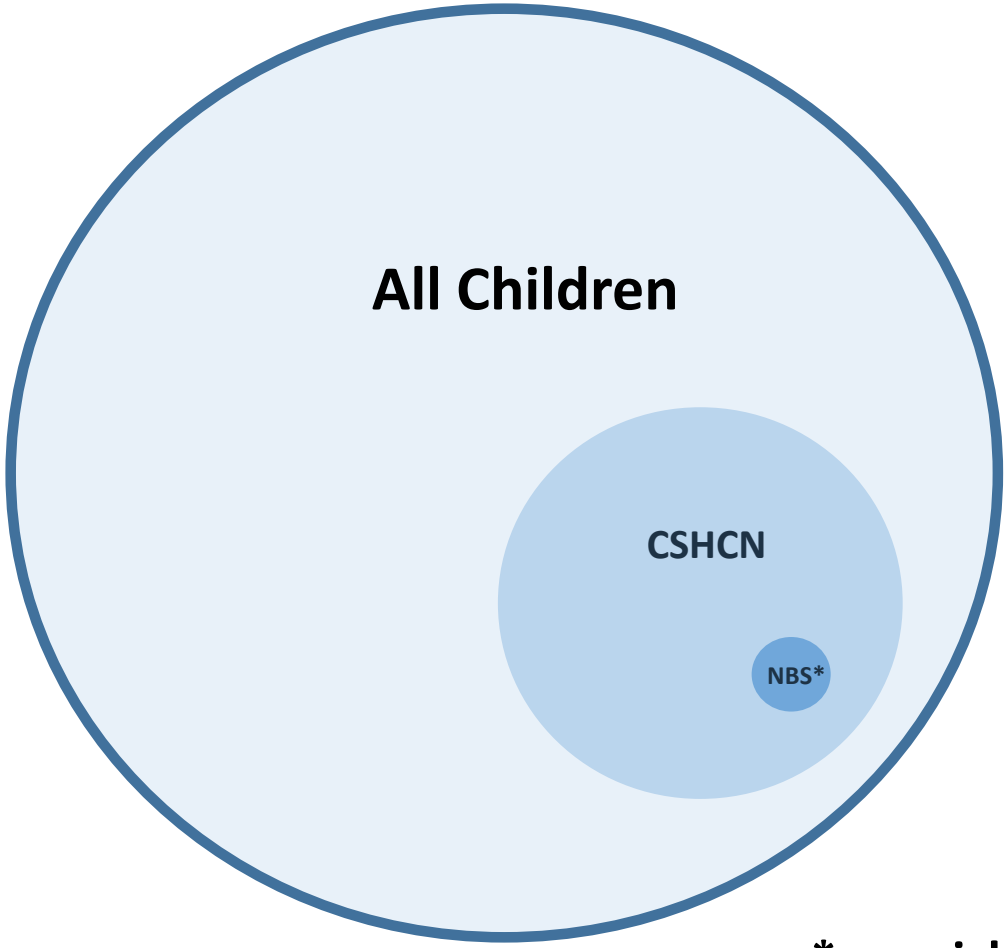
Hinton et al, 2016

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

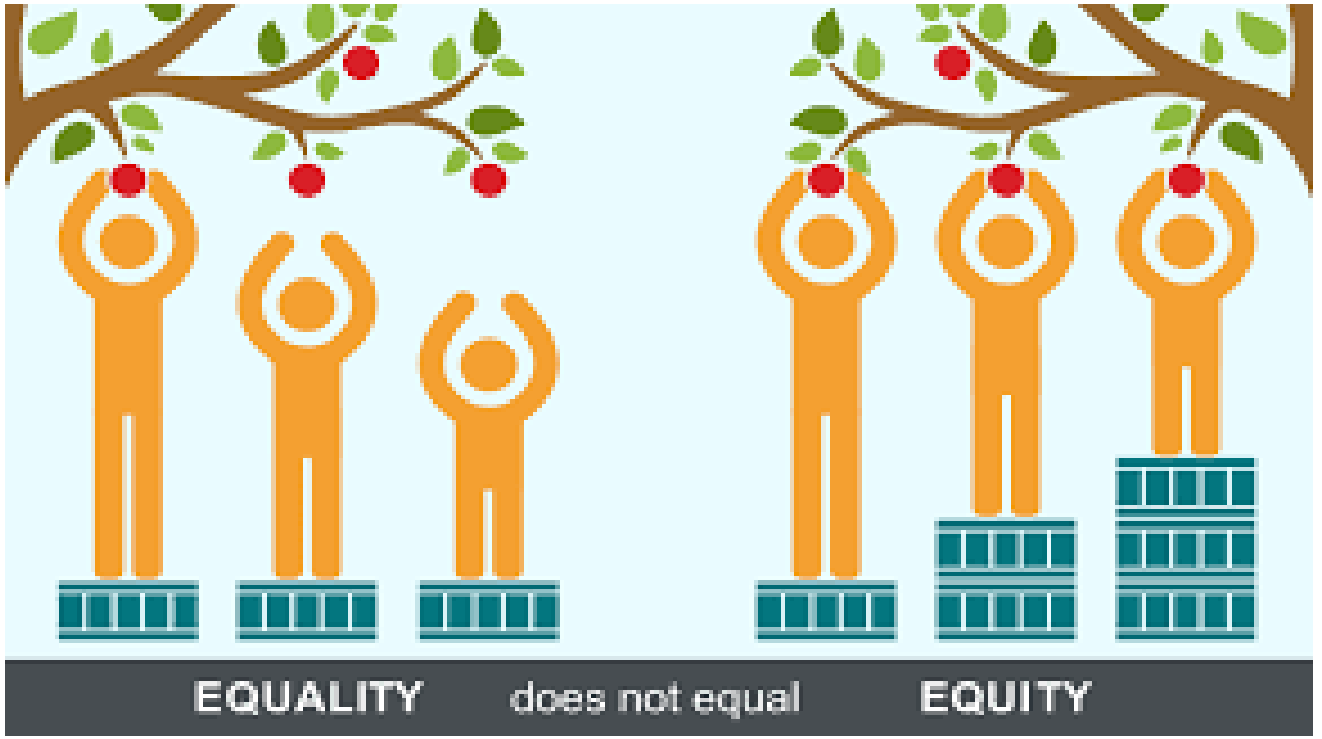
Approaches to Quality Assurance/Improvement



***e.g. sickle cell disease, cystic fibrosis, congenital hypothyroidism, medium chain acyl-CoA dehydrogenase deficiency**

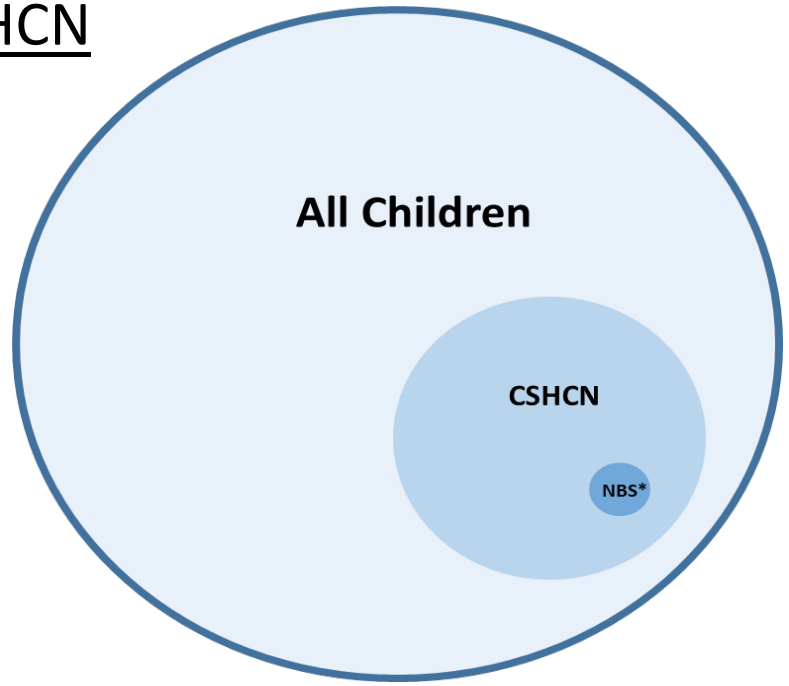
A Rose by any other name . . . “FUTR”

- What does “Follow-up and Treatment” really mean?
  - “Follow up” = assurance/reporting?
  - Does “Treatment” imply “equity”?
  - “Long-term”?
- Who is the “we”?



Who is the “we”? Some examples.

- MCHB/Medicaid/state department of health
  - Assurance and equity (reduce disparities) for all children
- State Title V CSHN programs
  - Assurance and equity (reduce disparities) 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



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.

LTFU Next Steps – Specific Recommendations

- LTFU work recommends that we explore what a coalition proposing a candidate NBS condition for inclusion on the RUSP might do to assure access to long-term follow-up and treatment
  - "Blueprint" addressing key (anticipated) issues for long-term FUTR?
    - E.g. Propose 2-3 condition-specific quality measures?
  - Provide recommendations for Feb 2019 meeting regarding evidence-review
- Continue to explore next steps for federated system
  - What else condition-specific coalitions can do?
    - Patient registries, centers of excellence, NORD
  - What state-level organizations can do?
    - "Birth defects" registries, NewSTEPs pilots
  - What national-level organizations can do?
    - CLSI, HEDIS, EHR