

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

ACHDNC MEMBERS

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- **Kyle B. Brothers, MD, PhD**
- Kamila B. Mistry, PhD, MPH
- **Annamarie Saarinen**

ORGANIZATION REPRESENTATIVES

- **Gerard T. Berry, MD**
Society for Inherited Metabolic Disorders
- Christopher A. Kus, MD, MPH (FUTR Co-Chair)
Association of State & Territorial Health Officials
- Jennifer M. Kwon, MD, MPH, FAAN
Child Neurology Society
- Robert J. Ostrander, MD
American Academy of Family Practice Physicians
- Jed L. Miller, MD, MPH
Association of Maternal and Child Health Programs

WORKGROUP MEMBERS

- Sabra A. Anckner, RN, BSN
- Tracey Bishop
- Amy Brower, PhD
- Luca Brunelli, MD, PhD
- Christine S. Brown, MS
- Debra Freedenberg, MD, PhD
- Lawrence Merritt, II, MD
- Dawn S. Peck, M.S., CGC
- Margie A. Ream, MD, PhD
- Elna Saah, MD
- Joseph H. Schneider, MD, MBA, FAAP
- Marci Sontag, PhD
- Janet Thomas, MD

MCHB

- Soohyun Kim, MPH, MSN
- Hannah Kotz

FUTR Workgroup Discussion Questions

Follow-up and Treatment Workgroup:

- What are the key workforce related challenges impacting access to short-and long-term follow-up, including treatment, for individuals and families identified with conditions on the Recommended Uniform Screening Panel?
- Are there examples of workforce innovations that have supported access to short- and long-term follow-up care?

Workforce Challenges

- Not enough clinical specialists
 - Dietitians, genetic counselors, social workers (with NBS expertise)
 - Physician specialists (clinical genetics, endocrine, neurology)
- Adding conditions to state NBS screening programs makes this worse
 - Children with NBS conditions start follow-up earlier than if clinically identified
 - Children without clear diagnosis can be numerous (“presumptive positives”)
- Burnout is not uncommon
 - Hard work (one clinician called it “critical care medicine”)
 - Treatment protocols not available or unfamiliar to clinicians
 - Pre-symptomatic: “all I have to offer is worry”
- Assumptions about current models of care

Workforce Innovations: Telehealth

- What's Working
 - Direct patient care relieves burdens for patients/families and addresses geographic barriers (specialists not evenly distributed)
 - Consultation model with primary care clinicians allows both adherence to treatment protocols AND local/personal treatment
 - MCHB supports states to have child psychiatry consultations for PCPs
- Challenges
 - Payment for telehealth is/may decrease as pandemic conditions change
 - Medical-legal issues with a consultation model
 - Clinicians crossing state borders (emergency exemptions ending)
 - Exemptions for specialists who practice in federally declared shortage areas?

Workforce Innovations: Payment

- What's Working
 - The U.S. already devotes enormous resources to health care
 - Value-based payment mechanisms have potential to improve systems of care
 - Bundled payments for specific conditions or global accountable care organizations (ACO)
 - Health care team uses resources to maximize quality
 - Pay for nutrition, social work, mental health, peer support, community health workers
 - **Models for ACOs and bundled payment mechanisms exist; potential ROI**
- Challenges
 - Epidemiology of child health in U.S. (e.g. NBS conditions are rare) make such models economically and logistically more challenging
 - Overall system of care for children and youth with special health care needs
 - **AMCHP mapped how to improve system: <http://cyshcnstandards.amchp.org>**

FUTR Workgroup Discussion Questions

All Workgroups:

- Should the Committee consider the availability of follow-up experts (clinical, follow-up public health staff, laboratory staff, etc.) when reviewing a new condition nominated to the Recommended Uniform Screening Panel?
- How could that information be collected?
- What role could the Committee play in calling attention to identified shortages of follow-up experts?

- Should ACHDNC consider the availability of follow-up clinical experts when reviewing a new condition for the RUSP?
- If availability of treatment presented as a yes/no then mixed opinions
 - NO: Don't screen if you can't treat
 - YES: If a treatment exists, we should be able to find a way to provide it
- Equity: is it ok to add a condition if only some will benefit?
- Better conceptualized as a continuum
 - Variation based on geography, insurance status, race/ethnicity, etc.
 - We are all responsible for improving health equity (NBS and beyond)
- ACDHNC
 - Include a “clinical impact” component (analogous to “public health impact”)
 - Treatment likely not available everywhere when condition nominated
 - Is there a reasonable path to sufficient capacity to treat all children?

- How could clinical information be collected?
What is the role of the ACHDNC re shortages of follow-up experts?
- Nomination package
 - Who are the clinicians involved in diagnosis and treatment?
 - Is there evidence of their availability? (e.g. professional society reports, MCHB studies)
 - What is the proposed plan for reaching all children identified with the condition through state NBS programs? (e.g. can PCP manage this with point-of-care consultation)
- Evidence review
 - Professional society surveys members re readiness to treat?
- Post-RUSP
 - Rapidly available treatment guidelines and training for clinicians
- General
 - Support state NBS programs to minimize referrals to clinicians for presumed positives
 - Highlight need to address needs of children with rare conditions (e.g. NBS) in the move to value-based care