ACHDNC
Advisory Committee on Heritable Disorders in Newborns and Children

ACHDNC Listening Session Update

Clinician Group

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November 3, 2023

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Questions about the Nomination Process

Currently, the Committee encourages a multidisciplinary team comprised of researchers, individuals with lived experience, advocacy organizations, states, etc., to develop and submit a nomination package for the ACHDNC to consider specific conditions for inclusion on the RUSP. In practice, this creates a substantial burden on groups that may not have sufficient resources to gather all the required information. Furthermore, in the near future, with advances in genomic sequencing and new treatments for rare conditions, hundreds of conditions may be considered for universal screening.

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1. Should the ACHDNC consider other approaches to the nomination process that reduce the burden on nominators and increase the roles of the ACHDNC and federal agencies to provide needed information?

- Should consider in order to ensure access to conditions without large and/or well supported advocacy group
- Consider proactive identification mechanisms- e.g. monitoring of FDA approvals or other available treatments
- HRSA or ACHDNC member “champion” for nomination packages
- Facilitate pilot programs for conditions that otherwise meet requirement
- Would need to be tied to funding given required expertise and time

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2. If the nomination process changes, how can we ensure that advocates and individuals with lived experience voices are included in the nomination process?

- The regulatory directive for involvement has been valuable for ensuring this is included

- Public comment meaningful but could expand to be discussion session for condition rather than listening session
  - Value to in person meetings

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3. Are there other gaps or concerns regarding the nomination package that you'd like to share?

- Maintain updated list of conditions that could go forward
- Anticipate need for expansion of Evidence Review committee to accommodate multiple
- Tracking data forward with long term follow up would improve future nomination process

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Questions about the Evidence-based Review Process

The criteria for inclusion of a condition on the RUSP is based on (1) published evidence that benefits outweigh harms and (2) the certainty of that evidence available in the peer-reviewed literature. Historically, the ACHDNC has applied these criteria to elements that focus on the benefits and harms to the individual child. Evidence regarding other elements such as benefits to the family or societal considerations such as financial cost or public health opportunity costs have not been considered. Furthermore, when making recommendations, the ACHDNC does not have a way to weigh the benefits to one population of children against the harms to another population of children.

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1. How to consider benefits of screening given the different perspectives, child, family, clinical, PHS, etc?

- Traditionally, the focus of benefit has been the newborn, and treatments, rather than:
  - Reducing the diagnostic odyssey
  - Early knowledge to help in family planning
  - Early diagnosis where Early Intervention is valuable, even w/o clear treatments
- Can/should the Committee consider broadening their scope of the benefits of newborn screening?
  - What do families want to know? Diagnoses, access to treatments…

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2. How to consider harms of screening given the different perspectives? 3. How to balance benefits and harms

- False positives on screening create some harms
- Some RUSP conditions have created patients-in-waiting, whose ambiguous health status creates a different medical odyssey
- Would a standing citizens advisory group provide additional perspective to potential harms of proposed NBS
- Should the Committee consider the harms to those affected when conditions are NOT approved for the RUSP

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4. How can the Committee consider the overall burden of potential illness that might be averted?

- Is the Evidence Review Decision Analysis sufficient?
- We have estimates of the costs of living with disease but we do less well in estimating the costs of early death
- Quality of life should also be a consideration
- Disability adjusted life year analysis could be a way of looking at the impact of NBS

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5. How can uncertainly regarding screening outcomes be systematically considered given lack of data, especially, potential harms? 6. How should costs (economic & opportunity) be measured?

- We need robust long-term follow-up of those identified by NBS
  - Informatics may help
- Could the nominators or HRSA provide some idea of longitudinal follow-up for conditions being proposed
- Could some discussion of long-term follow-up be part of the application package

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What I also heard while listening…

- The Committee should be aware of conditions where there is a treatment and a test that can be administered in newborns
  - What are the criteria for determining if the conditions belong on the RUSP?
  - The Committee title and scope includes “Heritable Disorders…” Should we be thinking of alternative ways besides addition to the RUSP to diagnose certain treatable conditions early
- The Committee should be proactive about the conditions nominated and about long-term follow-up to help understand the impact—good and bad—of NBS

Mandy David rocked as Listening Session moderator. Also, thank you Lisa Song!