ACHDNC
Advisory Committee on Heritable Disorders in Newborns and Children

ACHDNC Listening Session Update

Laboratory

November 3, 2023

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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?

- Bare bones approach to get on the list:
  - Condition, newborn screening test, treatment
  - Decreases burden on nominators
  - Will require another step with additional time & resources on the front end to gather information
  - Will not impact timeline for evidence review but may require prioritization of evidence reviews
  - Need to determine leader for the interim step
  - NBSTRN list of candidate conditions (n=34) already gathered this information
  - Need independent, advocacy and federal agency input

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

- Lowering intake for nomination removes barriers to access for advocates
- Need more bi-directional dialogue; most is one-way session
- Ask for all voices
  - Groups known to us come to the table, need to find those we don't know
- Need input from families in general in addition to those with impacted with rare conditions
- How to gather and format input in a more efficient manner
  - Listening, social media, others?

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

- The term nomination package is daunting, sounds like a lot before even tackled
- All information gathered is essential at some point; we will still need it, regardless of how it is assembled
- It is unlikely implementation will go faster even if the entry barrier is removed
- Need a reset - RUSP is not the endpoint, the metric for success is implementation
- **Goal is screening babies**

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How should the Committee consider benefits associated with screening given the different perspectives, such as child, family, clinician, public health system, society?

- Anecdotal stories absent from literature; not easily captured in publications
- Typically see disease discovery and later molecular basis
  - After that unpublishable
  - Little information on actual counts and outcomes
- No data on impact to society and medical system

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How should the Committee consider harms associated with screening given the different perspectives?, such as child, family, clinician, public health system, society?

- Asking questions about newborn screening to the general public may reveal perceived harms unrelated to those which are condition-specific
- False positive results; patients in waiting (overarching)
- Engage broad specialties and nationwide base of clinical providers to hear their perspectives on harms
- Issues regarding harms under study; need to hear results at the conclusion of these projects
- Late onset conditions, hard to know if test can be improved

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How should the Committee balance benefits/harms to come to a decision about net benefit?

- Equal consideration for both; may not balance out naïve to assume we should give more weight to a false positive outcome than a diagnosis
  - Hypothyroidism: high false positive rate; but common / false positive rate deemed “acceptable”
  - Rare conditions: high false positive rate deemed “not acceptable”, need a better test (e.g., 2nd tier)
- Work of the ACHDNC -- programs striving to develop highly specific tests
- Changing dogma that screening is “diagnostic”

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How should the Committee balance benefits/harms to come to a decision about How can the Committee consider the overall burden of potential illness that might be averted?

- Natural history v. early diagnosis: sibling stories
- How to measure emotional burden and suffering in addition to dollars
- Need a good definition of burden

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How can uncertainty regarding screening outcomes be systematically considered given the lack of data, especially about potential harms?

- Did we say definitions?
- Need precise definitions: What should Programs detect as conditions go through evidence review and committee discussion? SMA and SCID
- Counting conditions
- Redefine RUSP conditions – if we look for “everything” we diminish resources and downstream ability to implement screening for new conditions
- What are we looking for? “Classic v. late onset v. carriers”
- Rush to screen, often need to screen for some years to gather additional evidence; thus need a formal look back

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The issues of overall economic costs and opportunity costs are often unclear. How should the Committee consider these unmeasured costs, which are also likely to change over time?

- Costs change over time, need a formal process to review.
- Can only assess costs at the time of review
- Overall costs can be reduced if we establish a specific screening target
- Costs are dependent on geography (rural v. urban; nationwide) and availability of services

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SUMMARY

We must realize that no matter how we change the process, it will never be perfect