

ACHDNC Listening Session Update

Laboratory

November 3, 2023



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*Advisory Committee
on Heritable Disorders in
Newborns and Children*

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

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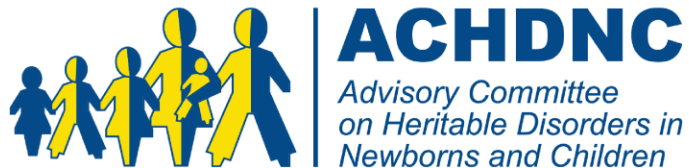
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How can uncertainty regarding screening outcomes be systematically considered given the lack of data, especially about potential harms?

- 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

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



SUMMARY

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