ACHDNC
Advisory Committee on Heritable Disorders in Newborns and Children

ACHDNC Listening Session Update

Public Health

November 3, 2023

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Overarching Themes

- Overall, there was a consensus that changes to the nomination system will be needed
- Lots of discussion around equity and the decision-making process, including data collection
- Concerns about data collection
  - who does it
  - who pays for it
  - Standardized approach to data collection
- Don't make changes that will delay timelines further
  - Centralized or standardized process may delay timelines
- Be cognizant of potential for unintended consequences of changes

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- Consider the purpose of the "1 case identified by pilot study" to determine why that is needed and whether the information from that could be obtained more efficiently in a different way
  - Case by case basis: what is the purpose of the pilot study and are there other more efficient ways to achieve that goal?
- Balance benefit of early diagnosis vs. risk of "breaking the system" by rapid additions of new conditions
- It may be ok to "limit the boundaries" of NBS to actually allow reasonable decision making - may need to consider follow up separately
- NBS needs to be a continuous learning system that adapts to it's learnings

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Nomination Process

- Various potential nominators don't have the bandwidth to put together the nomination package
- Involving advocacy groups and those with lived experiences is important to have their voice amplified in the nomination package
  - There was mention of having a clearer definition of "lived experience"
  - Anecdotes: balance between time spent and importance of hearing unique life experiences for each condition
  - Bundling conditions: generalizability of family lived experiences
- Implementation in three years is becoming more and more difficult for states
- If there is a rapid influx of new conditions, there is a potential to overwhelm NBS systems

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Evidence-based Review Process

- Important to look at the outcomes that parents and families care about and not just the intervention (i.e. early intervention)
- Many questions about how to measure and weigh the relative benefits and relative harms of true positives and false positives
  - Consider the downstream effects of both TP and FP performance of the screening approach
  - Minimizing false positives is very important
    - Multi-tiered testing approaches

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- Are there ways to try and standardize and score quality/ magnitude of benefit?
- The role of NBS in the context of the larger healthcare system
  - Challenges with follow-up in the healthcare system shouldn’t necessarily be a major factor in adding a condition that otherwise meets the criteria
  - Need to monitor and adapt to trends in the healthcare system
  - We shouldn't expect NBS to fix problems in the larger healthcare system
- Need to follow the evidence of whether early diagnosis impacts outcome

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Uncertainty around what we know

- Uncertainty about conditions and outcome of treatment
  - Natural history of rare diseases is often not well understood
  - NBS often leads to a recognition of milder forms
- Uncertainty about economic impact
- Often you don’t know the answers until you start screening

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Other Thoughts

- Prevalence of variants in conditions could be ascertained from existing sequencing data if it were available
  - That would be very helpful for NBS
- “Next Gen NBS” (credit Mei)
- Need for re-evaluation for conditions on the RUSP
  - Removal would require a high bar of evidence
  - But it could be done