Advisory Committee on Heritable Disorders in Newborns and Children

Cutoffs and Risk Assessment in Newborn Screening

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Catalysts for Discussion

- Committee Discussion
- NBS Advocates
- NBS in the News
- State NBS Programs expressed a need
Issues Raised by Stakeholders

- Missed cases
- Borderline results
  - Communication from NBS program
- Lack of uniformity
  - Laboratory methods
  - Conditions screened
Challenges Identified

- Due to different screening methods and multiple factors that impact screening results, cutoffs and screening algorithms can not be directly compared with one another.
- No consensus on the definition of a borderline result or how to process borderline results.
- Incomplete dataset on missed cases.
Challenges Identified

- Lack of resources to implement QA/QC activities
- Access to data (and staff with expertise to analyze and interpret complex data) that can inform the evaluation of cutoff values and screening algorithms
Addressing Challenges - Work in Progress

- **Association of Public Health Laboratories - Risk Assessment in Newborn Screening Resource Document**
  
  - A resource for states on approaches that can be used to assess risk and the tools available to assist in these efforts
  
  - Near final draft, posting on website, anticipated in June, 2018
Addressing Challenges - Work in Progress

- CDC’s Newborn Screening Quality Assurance Program – Harmonization Activities
  - Reporting de-identified normalized cutoffs to NBS laboratories to help them compare their cutoffs to their peers
  - Development of new proficiency testing and borderline materials for states to use

- NBS Technical Assistance Center and Data Repository
  - Supports state program evaluations
Are There Additional Ways The Committee Can Support States?

- Provide guidance on developing a systematic approach to evaluation of cutoffs and screening algorithms
- Support efforts to improve access to data, laboratorians, epidemiologists or biostatisticians as needed, who can conduct complex analyses on the data available, at the state level
- Provide rationale for development of resources at the state level to implement needed activities
Discussion