

Lab Standards and Procedures Subcommittee

May 13, 2010

Committee Members

SACHDNC Members	Additional Members
Ned Calonge	Carla Cuthbert
R. Rodney Howell	Amy Brower
Michael Skeels	Don Chace
Piero Rinaldo	Harry Hannon
Organization Representatives	Clem McDonald
Michael S. Watson	Kellie Kelm
Jane Getchell	
Sharon F. Terry	

Outcomes Study for FAODs

- Georeianne Arnold
- Utilize existing NBS databases to prospectively mine information on disease outcomes
- Allow assessment and utility and appropriateness of data collection within NBS databases
- Validate standards for diagnosis and stratify risk based on NBS results
- No obstacles to moving forward to full committee

Statistical Issues in Screening

- Reem Ghandour
- Review of statistical significance of second tier screening (especially CAH) and other NBS measures
- Balance between sensitivity and specificity
- Repeat vs. second tier screening
- Weigh costs and benefits of adding a sequential screen

NBS Program QA Measures

- Mike Watson (ACMG/NBSC)
- Report on existing QA systems and set stage for standardization of pre and post analytical best practices over whole newborn screening program
- Develop national benchmarks for timing and quality of NBS tests and consequences of not meeting them
- Establish quality measures for each step of process
- Transition from “how many” to “how well”

SCID Testing

- Ken Pass
- Luminex platform
 - CD3 and CD45
 - Correctly identified 11/127 samples
 - Compatible with TREC screening on same sample
- May be multiplexed to include other antigens
 - Endocrine markers
 - Lysosomal storage diseases

Future Focus

- Review of QA information on existing systems/tests
- Discuss role of routine second tier testing in NBS
- Development of network of regional specialization of newborn screening labs with expansion of testing menus
- Follow up on Mayo LSD comparison project