

# Laboratory Standards and Procedures Subcommittee

9/22/2011

# Members

## SACHDNC Members

Bruce Nedrow (Ned) Calonge  
R. Rodney Howell  
Fred Lowry  
Kellie B. Kelm  
Rebecca Buckley  
Dieter Matern

## Additional Members

Carla Cuthbert  
Amy Brower  
Mei Baker  
Harry Hannon  
Clem McDonald  
Brad Therrell  
Rebecca Goodwin

## Organization Representatives

### **American College of Medical Genetics**

Michael S. Watson

### **Association of Public Health Laboratories**

Jane Getchell

### **Genetic Alliance**

Sharon F. Terry

## HRSA Staff

Sara Copelland

# Subcommittee Charge

- Define and implement a mechanism for the periodic review and assessment of
  - the conditions included in the uniform panel
  - infrastructure services needed for effective and efficient screening of the conditions included in the uniform panel
  - laboratory procedures utilized for effective and efficient testing of the conditions included in the uniform panel.

# Acronyms

ABCDEFGHIJKLMNOPQRSTUVWXYZABCDEFGHIJ  
KLMNOPQRSTUVWXYZABCDEFGHIJKLMNOPQRS  
TUVWXYZABCDEFGHIJKLMNOPQRSTUVWXYZAB  
CDEFGHIJKLMNOPQRSTUVWXYZABCDEFGHIJKL  
MNOPQRSTUVWXYZABCDEFGHIJKLMNOPQRST  
UVWXYZABCDEFGHIJKLMNOPQRSTUVWXYZABC  
DEFGHIJKLMNOPQRSTUVWXYZABCDEFGHIJKLM  
NOPQRSTUVWXYZABCDEFGHIJKLMNOPQRSTU  
VWXYZABCDEFGHIJKLMNOPQRSTUVWXYZABCDE

# Accomplishments

- Second screen project (almost...)
- Health Information Technology (HIT)  
Workgroup: Collaborative NLM IT initiative (LOINC)
- Evaluation of novel molecular technologies
- Monitoring of CDC QA/QC materials for LSD and SCID testing
- Recommended direct comparison of alternative LSD testing technologies

# Committee Changes

- A new chair: Fred Lowry
- A new name: Laboratory and Information Technology Subcommittee
  - Evolution of scope of committee
  - Increasing role of information technology in NBS infrastructure

# Scope of Work

- Newborn screening applications and technology are likely to be all consuming in the near future
- There are other age windows for screening, but too much to deal with currently

# Wide Ranging Possibilities

- Reviewing new enabling/disruptive technologies
- Provide guidance for states making decisions about implement new screening tests
  - Comparative performance metrics
  - Overview of technologies
- Discussion of point of origin vs. traditional NBS screening labs
- *Etc...*



# Subcommittee Goals

- Establish process for regular review and revision of the standard panel
  - Remove disorders
  - Alter status from secondary to primary target
- Recommend specific changes to technology when indicated
  - Tyrosinemia 1
  - Succinylacetone
- Continued activity of HIT workgroup
- Monitoring new technologies