

**Laboratory Standards and  
Procedures Subcommittee Meeting**

May 6, 2011

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# Members

## SACHDNC Members

- Bruce Nedrow (Ned) Calonge
- R. Rodney Howell
- Fred Lowry
- Kellie B. Kelm

## 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

# Agenda

- Inderneel Sahai (MA NBS Lab): Refining MS/MS interpretation
- Steven Dowbrowski (Utah): Molecular screening for SMA+
- Jelili Ojodu (APHL): Defining quality measures
- Swapna Abhyanka (NLM): Messaging, ordering and receiving standards and HIT related to NBS labs

# MA MS:MS

- HRSA funded project to improve communication of MS:MS screening results
  - Better communication of risk
  - Improved prediction
- Development of improved markers incorporating multiple analytes (VLCAD example)
- Expanded pilot with MA, NY, WI, and CT

# Molecular Screening for SMA

- Challenges
  - Must account for multiple allelic configurations
  - SMN1 deletions are pathologic but SMN2 status is predictive
- Multiplex PCR based, DNA melting technique
- Can also multiplex with TRECs
- More?

# APHL Quality Measures

- Identify key quality measures currently followed by state screening programs
- Establish quality measures to capture for laboratory systems long term follow up
- Use for national IS purposes
- Very short time line to convene a working group and create a report (July)
- Will involve NLM

# NLM Report

- Update to the LOINC AHIC panel for reporting NBS reporting
- New codes for Hb reporting from Hb workgroup (multiple levels of reporting)
- Focus on new questions and answers for NBS interpretation.
- Requested that committee serve as expert panel
  - Help with developing structure and standard names for new codes for new conditions and tests
  - including diagnostic tests.