Lab Standards and Procedures Subcommittee Meeting Summary

Dietrich Matern, MD

May 18, 2012

Secretary's Advisory Committee on Heritable Disorders in Newborns and Children
Laboratory Standards and Procedures Subcommittee Meeting
May 2012

Fred Lorey, PhD (Chair)

SACHDNC Members
Dietrich Matern, M.D.
Kellie B. Kelm, Ph.D.

Organization Representatives
American College of Medical Genetics
Michael S. Watson, Ph.D., FACMG
Association of Public Health Laboratories
Jane Getchell, Dr. PH.

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

Agenda:

2:15- 2:20	Welcome from Dr. Lorey
2:20- 3:20	Discussion of priorities and projects upcoming for the Lab Standards Subcommittee
3:20- 4:15	Review of Condition Review Process report- Alex
4:15- 5:00	Updates and issues from NLM

Priority 1:
Review new enabling/disruptive technologies

- **Project #1 - Provide the pros/cons incl. uncertainties of the various (old & new) platforms for the nominated and current conditions on the RUSP in order for states to make informed decisions (e.g., succinylacetone as part of AC/AA analysis).**

- **Project #2 – Provide implementation toolkit to NBS programs for new conditions on the RUSP (e.g. SCID)**
  - **SOPs**
  - **"slide sets" that can be used for the education of advisory boards, administrators, legislatures, etc.**

- **Project #3 – Region 4 MS/MS Data Project summary**
  - **Review data collected and tools developed as part the project;**
  - **Assess project impact on NBS programs using R4 data and tools;**
  - **Review training course that was offered by the project.**

Priority 2:

Provide guidance for state NBS programs in making decisions about implementation, integration, FU, and QA

- **Project #1 – Comparative performance metrics**
  - **Review APHL Quality Indicators metrics**
  - **Review NBS case definitions**
- **Project #2 – Point of care NBS**
  - **What's the landscape? - review/outline the roles of PH programs in POC testing (who is responsible for administration/quality; compare loss to FU for different models; use hearing loss screening as example).**
  - **Is there a perfect model?**
- **Project #3 – Develop a tool for capturing delayed/missed diagnoses**

Priority 3:

Establish process for regular review and revision of the RUSP and recommend specific changes to technology when indicated

- **Project #1 – How to remove disorders**
- **Project #2 – How to move a condition from secondary to primary target**

Provide input to Evidence Review group on lab and technical aspects related to testing for conditions