DACHDNC Laboratory Standards and Procedures Subcommittee

Susan M. Tanksley, Ph.D.

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Priorities for Lab Subcommittee

- Review new enabling/disruptive technologies
  - No update at this time
- Provide guidance for state NBS programs in making decisions about lab implementation, integration, follow-up (FU), and quality assurance (QA)
  - Updates on CLSI Guidelines
- Establish process for regular review and revision of the Recommended Uniform Screening Panel (RUSP)
  - No update at this time

Update on Guidance to State Laboratories

- Conditions newly added to RUSP
- Severe Combined Immunodeficiency (SCID)
  - NBSTRN requests input from monthly call participants – on agenda for May conference call
  - Conference calls held the fourth Friday of each month at 1 PM eastern
  - Guidance: lab implementation, FU, QA
- Workgroup
  - Amy Brower
  - Mei Baker
  - Jane Getchell

Update on CLSI Guidelines: NBS06-A

NBS06-A: Newborn Blood Spot Screening for Severe Combined Immunodeficiency by Measurement of T-cell Receptor Excision Circles

- May 2010
  - SCID added to Recommended Uniform Screening Panel
- August 2011
  - CLSI convened DDC in Atlanta
- June 2012
  - DDC voted to adopt the CLSI draft
- Oct-Nov 2012
  - Draft document was open to comments from CLSI delegates (~1100)
- March 2013
  - Draft was finalized and approved by consensus committee
- April 2013
  - CLSI published the guideline

Authors

NBS06-A: Newborn Blood Spot Screening for SCID

W. Harry Hannon, PhD
Roshini Sarah Abraham, PhD, D(ABMLI)
Lisa Kobrynski, MD, MPH
Robert F. Vogt, Jr., PhD
Ona Adair, PhD
Constantino Aznar, PhD
Mei W. Baker, MD, FACMG
Amy M. Brower, PhD
Michele Caggana, ScD, FACMG
Anne Marie Comeau, PhD
William Grossman, MD, PhD
Francis K. Lee, MSc, PhD
Jennifer M. Puck, MD
Jennifer L. Taylor, PhD
Danielle M. Turley, PhD
Mirjam van Der Burg, PhD
Berta Warman, MS
Golriz K. Yazdanpanah, MS
Alice Ylikoski, PhD

NBS06-A: Newborn Blood Spot Screening for SCID

- Provided overview of the document
- Sections include:
  - Terminology
  - Biology of SCID
  - Biological and clinical features of SCID
  - Overview of real-time PCR Assays
  - Implementation of T-cell receptor excision circle assay
  - Follow-up activities, communication, and diagnostic testing
- Copies of the CLSI document will be sent to each state program by CDC

NBS06-A: Newborn Blood Spot Screening for SCID

- **Appendix A: Immunodeficiency Disorders and T cell Receptor Excision Circle (TREC) Values in the Newborn Screening Period**
  - Based on specimen collection at 24-72 hrs (initial screens)
  - Immune deficiencies and other T cell lymphopenias divided into 4 categories
    - **Primary Immunodeficiency Disorders Typically Associated with TREC Values Below the Expected Range in the Newborn Screening Period**
      - **Includes Typical SCID & Complete DiGeorge Syndrome**
    - **Primary Immunodeficiency Disorders Variably Associated with TREC Values Below the Expected Range in the Newborn Screening Period**
      - **Includes leaky SCID, variant SCID and syndromes with T cell impairment**
    - **Primary Combined Immunodeficiency Disorders Unlikely to be Associated with TREC Values Below the Expected Range in the Newborn Screening Period**
      - **Includes ADA deficiency (partial) and other defects such as CD8 deficiency which do not cause severe T cell lymphopenia**
    - **Secondary Disorders Variably Associated with TREC Values Below the Expected Range in the Newborn Screening Period**
      - **Includes prematurity and other secondary t-cell lymphopenias (e.g. chylothorax following cardiac surgery)**

Tentative Agenda Items for Future Meetings

- Implementation of Pompe Screening
  - Pending vote from DACHDNC
- Tyrosinemia Type I Survey Outcomes
- Genomic Sequencing Initiatives
- Comparison of Technologies for LSDs and Other Conditions

Questions?