SACHDNC Laboratory Standards and Procedures Subcommittee

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CDC Recommendations for Good Laboratory Practices in Biochemical Genetic Testing and NBS for Inherited Metabolic Disorders

- Presenter: Bin Chen, PhD (CDC)
- Intent of recommendations
  - Provide quality management guidance for genetic testing performed for screening, diagnosis, monitoring, and treatment of heritable metabolic disorders
  - Consider BGT and NBS separately when practices differ
  - Clarify CLIA requirements and provide additional good laboratory practice recommendations
  - Complement 2009 CDC guideline for molecular genetic testing

CDC Recommendations for Good Laboratory Practices in Biochemical Genetic Testing and NBS for Inherited Metabolic Disorders

- Intended audiences
  - Laboratory professionals
  - Laboratory surveyors and inspectors
  - Users of laboratory services
  - Standard-setting organizations
  - Professional societies
  - IVD manufacturers
- Expected outcomes
  - Improve quality of laboratory genetic services
  - Improve healthcare outcomes from genetic testing

Lab Subcommittee Discussion

- Report is not ready for the full Committee to vote on for support.
- Would like more information on how this could impact State programs.

Discussion on CLSI Document – Newborn Blood Spot Screening for SCID by Measurement of TREC

- Addresses the detection of SCID by population-based newborn screening using dried blood spot specimens to measure TREC.
- Need volunteers to review draft during the CLSI document development process.

Discussion on NBS Quality Indicators

- Supports Priority B: Provide guidance for State NBS programs in making decisions about lab implementation, integration, follow-up, and quality assurance
- Important to confirm the quality of the data submitted
- Provide feedback to States based on data received
- States could use the new data repository in NewSteps for case management
- Important to discuss with States – What do States get back? How will this data be meaningful to States? What would this be valuable to States.
- Don't duplicate efforts; don't reinvent the wheel
  - Don't want to input same data in various places

NBS Case Definitions

- Supports Priority B: Provide guidance for State NBS programs in making decisions about lab implementation, integration, follow-up, and quality assurance
- Next steps: Several states have volunteered to beta test the case definitions modules for the different disorder categories
- How to get outcome data back to States so they can improve their programs – ACMG looking at this closely

Priority Projects

- Priority A: Review new enabling/innovative technologies
  - Begin with succinylacetone as part of AC/AA analysis
  - Possibly include in MMWR – depends on the information collected
  - Workgroup
    - Lead: Carla Cuthbert (CDC)
    - Dieter Matern
    - Stan Berberich
- Proposed Finish Date: Presentation at May 2013 meeting

Priority Projects cont'd

- Priority B – Provide guidance for state NBS programs in making decisions about lab implementation, integration, follow-up , and QA
  - Project – Comparative performance metrics
    - In progress
  - Project – Slide deck for State Labs when a new condition is added to the RUSP
    - What types of info is needed so State Labs can discuss with CMOs, Legislature, hospitals, etc
      - In progress – begin with SCID
        - Amy Brower, Jane Getchell, Mei Baker

Priority Projects cont'd

- Priority C: Establish process for regular review and revision of the RUSP and recommend specific changes to technology when indicated.
  - Project – Work with Condition Review Group to develop lab requirements for their reviews. **This project is a joint project with all three subcommittees.*

Membership

- Call for self nominations for the Lab Subcommittee.
- Categories of expertise –
  - State Lab (with expertise in molecular)
  - Commercial Labs
  - Clinicians
  - Pathologists

Update – Health Information Technology

- New version of LOINC newborn screening panel is available (www.nlm.nih.gov/newbornscreeningcodes)
- NLM would like feedback
  - Are there new codes needed for second screen tests?
  - New codes needed for confirmatory or diagnostic testing?
  - How are NBS labs reporting mutations found and mutations test for NBS conditions where they do genetic testing.

Questions?