Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children

Laboratory Standards & Procedures Subcommittee

Report 9/18/2007

Routine Second Specimen Study

- Status of State IRB Approvals
  - Retrospective study
    - 1 out of 16 states gained IRB approval
    - Note that retrospective study is category IV exempt per CDC
  - Prospective study
    - 0 out of 16 states gained IRB approval
    - Note that prospective study may not be considered Human research per CDC
  - Plan – continue to work with states to gain approval
    - Important issue to consider for the Committee as we plan future multi-center studies
- Electronic data collection form
  - Almost complete and will use Delaware data to pilot

Update by Dr. Harry Hannon

DNA Training Course

- One week course
- Emphasis on DNA testing using CF as the focus
- Laboratory and Follow-up personnel
- 4 potential sites (WI, TX, MA, NY)
- Sponsored by APHL, CDC, NNSGRC with input from CFF
- Projected start date Feb 2008

Update by Dr. Harry Hannon & Dr. Megan Latshaw

FDA Guidances Impacting NBS

- Devices
  - Discussed guidance released 9/14/2007 (“Guidance for Industry and FDA Staff - Commercially Distributed Analyte Specific Reagents (ASRs): Frequently Asked Questions”) and the potential impact on CF NBS.
  - Specimen collection paper sole source issue
    - CDC to produce generic collection forms for emergency purposes
- Drugs
  - No update

Update by Dr. Ethan Hausman & Dr. Harry Hannon

Newborn Proficiency Testing

- Expanded Panel
  - 12 primary analytes
  - Not all available commercially
  - CDC lab to synthesize
- ACMG/CAP Meeting – PT Challenge Packet
  - CDC changing challenges worldwide from 4 to 3
  - CDC adding 1 challenge unique to US and Canada
    - Mimic state experience weighted towards variant samples
    - Reporting will focus only on positives to facilitate data analysis
    - Data will be publicly available

Update by Dr. Harry Hannon

NBS For CF and a Uniform Mutation Panel

- CF Testing Implementation
  - 2004 recommendation by HHS, CDC, and CFF for NBS
  - Focus on prevention of symptoms rather than treatment
  - Estimate by 2008 90% of babies will be screened for CF
  - Implementation challenges
    - Testing algorithms vary
    - Mutation panels vary
      - Some include non-disease causing mutations

Update by Dr. Phil Farrell

NBS For CF and a Uniform Mutation Panel

- Recommendations
  - Uniform mutation panel
  - Include only CF causing mutations
    - Class IV and V included
    - CFF funded research to define CF causing mutations
  - Include CF causing mutations occurring in minority populations
  - Exclude CFTR polymorphisms that are not disease causing
  - CFF and CDC determine content of uniform panel
  - Include regulatory agencies and industry in efforts

Update by Dr. Phil Farrell

State Practices on Analyzing & Reporting Non-Mandated Conditions

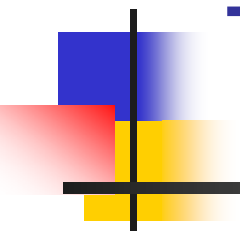
- NNSGRC Survey
  - 47/51 programs answered survey
  - 34 programs report results for non mandated conditions
  - 25 programs report concerns about tyrosinemia-type 1
- Feedback from programs informative for implementation of uniform panel

Update by Coleen Buechner

Prevalence of CH

- Published increase in CH cases based on NNSGRC data (Harris & Pass)
- Dr. Brad Therrell convening working group to review issue
- Update at January 2008 subcommittee meeting

Update by Dr. Marie Mann



Thank you!