LABORATORY STANDARDS AND PROCEDURES WORKGROUP

February 9, 2017

Kellie Kelm, PhD, chair
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WORKGROUP ROSTER

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Chair: Kellie Kelm
Co-chair: Susan Tanksley
HRSA staff: Ann Ferrero

AGENDA

- Introduction of new members
- A short oral presentation followed by workgroup discussion:
  - Implementing screening for LSDs and X-ALD and difficulties re. timeliness goals
- Other discussion: post-analytical tools

TIMELINESS GOALS

- To achieve the goals of timely diagnosis and treatment of screened conditions and to avoid associated disability, morbidity and mortality, the following time frames should be achieved by NBS programs for the initial newborn screening specimen:
  - Presumptive positive results for time-critical conditions should be communicated immediately to the newborn's healthcare provider but no later than five days of life.
  - Presumptive positive results for all other conditions should be communicated to the newborn's healthcare provider as soon as possible but no later than seven days of life.
  - All NBS tests should be completed within seven days of life.
- In order to achieve the above goals:
  - Initial NBS specimens should be collected in the appropriate time frame for the newborn's condition but no later than 48 hours after birth, and
  - NBS specimens should be received at the laboratory as soon as possible; ideally within 24 hours of collection.

IMPLEMENTING SCREENING FOR LSDs, X-ALD & HOW TO MEET TIMELINESS GOALS

- States have seen improvement in transit time with addition of a courier
- Still much room for improvement in timeliness of hospital submissions
- Many states noted that meeting the timeliness goals for transit time are still challenging

IMPLEMENTING SCREENING FOR LSDs, X-ALD & HOW TO MEET TIMELINESS GOALS

- Screening for LSDs by MS/MS requires an overnight incubation
- Screening for X-ALD uses second tier MS/MS testing to reduce false positives
- Also noted: some states perform second-tier DNA testing to reduce call-outs
- The transit time and screening issues compound and states are having difficulty meeting timeliness goal for reporting

PREANALYTICAL TIMELINESS CONSIDERATIONS

- Transit time = Time from collection to receipt in testing lab
- Logistical issues
  - Timing of collection vs. time of pick-up vs. time of delivery
  - Time of collection should occur between 24 & 48 hrs of life
  - Specimens must dry 3-4 hrs before shipping
  - Specimens drawn too close to courier pick up time can't be shipped until following day so will greatly exceed 24 hr recommendation
  - Large commercial couriers do not pick up 7 days/week (e.g. FedEx, UPS) and have holidays (no pick up and/or delivery)

ANALYTICAL FACTORS AFFECTING TIMELINESS

- Testing methodologies differ in length
  - Long incubations
- Second-tier testing to reduce call-outs
  - Tandem mass spectrometry
    - use of MS/MS as second tier may be faster than DNA
  - DNA-based
    - PCR
    - Sequencing

TIMELINESS GOALS

- Discussion points
  - Despite efforts to improve transit time, one state mentioned they still only get ~50% of specimens within 24 hrs of collection
  - Workgroup could look at more recent timeliness data and assess more recent data and consider whether there could be suggestions for changes to the recommendations presented to the committee.
    - Should we develop a new measure – age of baby when specimen is received?
    - Should there be different timeliness goals for different conditions?
  - Stat labs needed for second-tier?

FUTURE DISCUSSION TOPICS

- X-ALD screening status
- Update on timeliness data from NewSTEPs
- Case Definitions
  - R4S - variable conditions may be recorded with same diagnosis noted

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