

LABORATORY STANDARDS AND PROCEDURES WORKGROUP

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WORKGROUP ROSTER

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Chair: Kellie Kelm

Co-chair: Susan Tanksley

HRSA staff: Ann Ferrero

Agenda

- Welcome & roll call
- Workgroup roster updates
- Discussion on Newborn Screening in Genomic Medicine and Public Health (NSIGHT) presentations
- Timeliness and the impact of molecular testing

NGS/NSIGHT Inspired Discussion

- Discussion of non-newborn/childhood testing – could the committee play a role?
- When is there another time to have all children tested (audience capture) if it's a public health activity?
- Discussion of guidelines for laboratories using old data – e.g. hemoglobinopathies, DNA raw data 20 years from now
- Discard data – liability issue? Cheaper to re-test? Best practices?
- Software compatibility

Wednesday's vote on 1 prospective case

- One proposal for a matrix – nomination process
 - Discussion of frequency of the condition vs. cost of screening
 - It will be harder when conditions are more rare, but we can't lower the bar
- Laboratorians are used to a process, removing subjectivity from the process
- Don't bypass the follow-up piece of the process

Timeliness – impact of molecular testing

- Adding NGS and other second tier tests adds time to testing
- May impact ability to meet the timeliness recommendations
 - Revisit the recommendations as we get real data?
- States sharing molecular resources could help with timeliness, \$ issues with adopting new technology
- If we get to the diagnosis faster, then you've made an improvement in timeliness

Timeliness – preanalytic issues

- Quality of DBS and timeliness of collection and transit still an issue
- Turnover at hospitals still a major issue – new staff is not being trained on collecting a good DBS, etc. Members report a variable level of improvement and maintenance of timeliness in their relationships with hospitals.
- Need a champion in each facility
- Hiring a courier service is still a \$\$ issue and some couriers still aren't timely with shipping
- Need a role of Joint Commission
- Transparent timeliness data available to the public