Laboratory Standards and Procedures Workgroup

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ACHDNC
Advisory Committee on Heritable Disorders in Newborns and Children

Laboratory Standards and Procedures Workgroup Members

ACHDNC MEMBERS

- Carla Cuthbert, PhD
  Centers for Disease Control & Prevention
- Kellie B. Kelm, PhD (Chair)
  Food and Drug Administration
- Shawn E. McCandless, MD

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- Maximilian Muenke, MD, FACMG
  American College of Medical Genetics & Genomics
- Scott Shone, PhD, HCLD(ABB)
  Association of State & Territorial Health Officials
- Susan Tanksley, PhD (Co-chair)
  Association of Public Health Laboratories

WORKGROUP MEMBERS

- Stanton Berberich, PhD
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- George Dizikes, PhD
- Rosemary Hage, PhD
- Patricia Hall, PhD, FACMG
- Nathalie Lepage, PhD, FCCMG, FCACB
- Van Leung-Pineda, PhD, DABCC, FAAC
- Jelili Ojodu, MPH
- Miriam Schachter, PhD
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- Holly Winslow

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- Kim Morrison, MS

LS&P Workgroup Discussion Questions

- What are successes and challenges around NBS laboratories in implementing conditions added to the RUSP?
- What issues/factors contribute to the variability of implementation status of conditions added to the RUSP across the states?
- What are potential solutions or resources around laboratory standards and procedures that can address these issues/factors?

LS&P Workgroup Discussion Questions

- What are successes and challenges around NBS laboratories in implementing conditions added to the RUSP?
  - Authority to screen
    - Legislation ← time for these processes not captured in the survey
    - Administrative rules
    - Agency/advisory committee
  - Competing program initiatives or public health priorities
  - Limitation in how much more DBS there is for future tests

LS&P Workgroup Discussion Questions

- What are successes and challenges around NBS laboratories in implementing conditions added to the RUSP?
  - Funding and costs associated with implementation
  - Lack of success in applications to obtain funding
  - Lack of staff or space
  - Access to + establishing relationships with specialists/treatment in state, genetic counseling
  - Waiting for contract laboratory to get the screening test
  - Whether tests can be multiplexed with current NBS tests

LS&P Workgroup Discussion Questions

- What issues/factors contribute to the variability of implementation status of conditions added to the RUSP across the states?
  - Technical expertise for test development, validation
  - Adding analyte to FDA cleared test → LDT
  - Condition not meeting state's criteria for screening
  - Lack of clear cost-benefit (a need for some states)
  - High number of false positives (need to bring on second tier test)
  - How states assess certainty of screening – certainty can come from factors such as results from a large pilot study

LS&P Workgroup Discussion Questions

- What are potential solutions or resources around laboratory standards and procedures that can address these issues/factors?
  - Ability to multiplex with existing conditions on panel
    - Not always the optimal choice for each analyte
  - Having infrastructure/expertise in place (instruments/ people)
  - Having well-defined protocols
  - Technical team from CDC that can help labs implement/ troubleshoot new tests
  - Having an FDA cleared kit

LS&P Workgroup Discussion Questions

- What are potential solutions or resources around laboratory standards and procedures that can address these issues/factors?
  - A champion / project manager in the program that can usher the addition of the test from beginning to end
  - Funding associated with implementation (e.g. seed funding)
    - Entity that could help states put together more robust grant applications (e.g. grant writing training)
  - Strong relationships, communication and expertise from staff, medical professionals and partners
  - Tests using other specimen types (urine, saliva)

LS&P Workgroup Discussion Questions

- What are potential solutions or resources around laboratory standards and procedures that can address these issues/factors?
  - Convening stakeholders to provide input on factors to consider for new tests, e.g., acceptable false positive rate relative to true positive rate
  - CDC developing tools for states using NGS in screening protocol
  - Improving sensitivity of tests, so a smaller punch might be used
  - Different way to classify a state that assesses a condition according to their criteria and decides not to add it to their panel at that time
  - Communicating and sharing insight from NBS programs who are screening
  - Staff attending national trainings