

# Laboratory Standards and Procedures Workgroup Meeting

February 11, 2021

Kellie Kelm, Chair

Susan Tanksley, Co-chair

# Laboratory Standards and Procedures Workgroup Members

## ACHDNC MEMBERS

- Mei W. Baker, MD
- Carla Cuthbert, PhD
- Kellie B. Kelm, PhD (chair)
- **Shawn E. McCandless, MD**
- Scott M. Shone, PhD, HCLD(ABB)

## ORGANIZATIONAL REPRESENTATIVES

- **Maximilian Muenke, MD FACMG**  
American College of Medical Genetics & Genomics
- Susan M. Tanksley, PhD (co-chair)  
Association of Public Health Laboratories

## WORKGROUP MEMBERS

- Stanton L. Berberich, PhD
- Michele Caggana, ScD, FACMG
- George Dizikes, PhD
- Rosemary Hage, PhD
- Patricia Hall, PhD, FACMG
- Travis Henry, PhD
- Nathalie Lepage, PhD, FCCMG, FCACB
- **Van Leung-Pineda, PhD, DABCC, FAAC**
- Jelili Ojodu, MPH
- Miriam Schachter, PhD
- Bonita Taffe, PhD
- Holly Winslow

# Laboratory Standards and Procedures Workgroup

## Discussion Questions

- What information would be most helpful from newborn screening laboratories related to the review of conditions on the RUSP? How can we prepare newborn screening laboratories to collect and report this data?
- Expected PPV and NPV should be determined for screening each condition. States report whether they meet the NPV and PPV. Report and evaluate the FPR and FNR. May demonstrate need for 2<sup>nd</sup> tier test.
- Must start with good case definition, then examine what we're screening for and what else we're finding. Note that the case definition could also change over time.
- NewSTEPS could be used to collect the data.

# Laboratory Standards and Procedures Workgroup

## Discussion Questions

- Should there be more in-depth information regarding cost to labs for adding a new condition to the panel. Or is there already enough information provided?
- Cost of the overall *system* (Susan's egg diagram), not just the reagents, employees, instruments, second tier testing, LIMS, etc.
- Use a bucket approach (small/medium/large) – break cost down by states that start at different levels of readiness to bring on a specific new condition

# Laboratory Standards and Procedures Workgroup

## Discussion Questions

- Are there any other considerations for enhancing either the nomination process or review of conditions on the RUSP?
- Better define the new conditions under review (like SMA)
- Evidence review has provided extensive and thorough information, will benefit from the proposed enhancements
- Confusion over what the RUSP is.
  - Many interpret intent is to screen for primary and secondary targets
  - For those with RUSP in state law, then interpret as required to screen for secondary targets
    - Suggestion: education for everyone in the system about the secondary targets. Revise wording/display of RUSP on ACHDNC website