

# LABORATORY STANDARDS AND PROCEDURES SUBCOMMITTEE

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February 12, 2016

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# SUBCOMMITTEE ROSTER

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# Original Laboratory Standards & Procedures Subcommittee Charge

- Define and implement a mechanism for the periodic review and assessment of:
  - The conditions included in the uniform panel
  - Infrastructure services needed for effective and efficient screening of the conditions included in the uniform panel
  - Laboratory procedures utilized for effective and efficient testing of the conditions included in the uniform panel.

# Clarification of Subcommittee Charge

Define and implement a mechanism for the periodic review and assessment of

1. The conditions included in the uniform panel
2. **Laboratory procedures** utilized for effective and efficient testing of the conditions included in the uniform panel.
3. **Infrastructure** and **services** needed for effective and efficient screening of the conditions included in the uniform panel

# Potential Projects

1. Define and implement a mechanism for the periodic review and assessment of the conditions included in the uniform panel

- Develop a process to periodically review data for conditions on the RUSP.
  - Ongoing process (NewSTEPs data)
  - One or few conditions at a time
  - Could lead to new projects if issues noted

# Potential Projects II

2. Define and implement a mechanism for the periodic review and assessment of laboratory procedures utilized for effective and efficient testing of the conditions included in the uniform panel.

- Evaluate current methods to determine if improvements are needed to enhance sensitivity or specificity
  - Is there a better analyte to identify a particular condition (e.g. Tyr vs. SUAC and T4 vs. TSH)?
  - Assess utility of additional data (e.g., gestational age, birth weight) to determine callouts
  - Second-tier testing to improve specificity
  - Use of percentile cut-offs vs. floating cut-offs vs. multiple of the mean

# Potential Projects III

2. Define and implement a mechanism for the periodic review and assessment of laboratory procedures utilized for effective and efficient testing of the conditions included in the uniform panel.

- One screen vs. two screens....
  - What are the pros and cons of both models?
  - What are we screening for? Need uniform case definitions
  - Can babies identified on second screen with first screen normal be identified by single screen model with targeted rescreening?
  - How do we establish a study design to assess this issue?

# Potential Projects IV

2. Define and implement a mechanism for the periodic review and assessment of laboratory procedures utilized for effective and efficient testing of the conditions included in the uniform panel.

- Explore the role of next generation sequencing in newborn screening
  - Screening is currently based on phenotypic data. How do we accumulate the data to identify correlation between phenotypic & genotypic data?
  - Are there conditions for which sequencing is the only screening method?
  - What do you gain/lose from NGS?
  - Which data do you report?
    - What do you do with variants of unknown significance?
    - When do you report carrier status? Are there particular conditions where reporting carrier status is important?
- What new infrastructure needs to be built for NGS?



# Potential Projects V

3. Define and implement a mechanism for the periodic review and assessment of infrastructure and services needed for effective and efficient screening of the conditions included in the uniform panel.

- A portion of the timeliness initiatives fits here:
  - Review data related to testing (Timeliness 1.0)
  - What are the implications of earlier specimen collection (<24 hrs)?
    - California recently published study on early specimen collections (12 hrs)
  - What are the unforeseen consequences and costs of timeliness?

# Potential Projects Compiled

1. Assess conditions	2. Laboratory procedures	3. Infrastructure and services
Review screening data	Evaluate current methods to determine if improvements are needed to enhance sensitivity or specificity	Assessment of data for Timeliness 1.0 recommendations
	One screen vs. Two screen	
	NGS in newborn screening	

# Subcommittee question

1. Define and implement a mechanism for the periodic review and assessment of the conditions included in the uniform panel

- What is the mechanism to consider:
  - Moving conditions off the panel?
    - SCAD
  - Promoting conditions from secondary panel to core panel?
    - Arginemia
- Which subcommittee should address point of care issues?