# Strengthening Newborn Screening System: RUSP implementation

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# The issue

- The optimization of health impact of newborn screening across the nation benefits from a cohesive approach that assures access to screening, diagnosis and treatment services regardless of where an infant is born
- The implementation of the RUSP varies by state, for a variety of reasons

# Outline for session

- Explore the elements, enabling factors and challenges that impact the implementation of new conditions added to the RUSP by a state's newborn screening program
  - Introduction:  Ned Calonge
  - Diagnosis and treatment:  Erica Wright
  - Laboratory:  Scott Shone
  - Discussion:  Ned Calonge (moderator)

# Introduction

# Overarching elements

- The decision of the ACHDNC to recommend to the Secretary of Health and Human Services that a new condition should be added to the RUSP does not guarantee implementation in every state
  - The Secretary has discretion in accepting the recommendation for the addition
  - The RUSP is not a mandate:

"The addition of MPS II to the Recommended Uniform Screening Panel does not constitute a requirement for states to implement screening and is only a recommendation."

# Implementation stakeholders

- NBS program directors
- PHL directors
- State NBS advisory committees
- State PH leader(s)
- Governor’s office/Governor
- State legislatures/legislators
- Hospitals, providers
- Families and advocates

# Other elements of NBS system

- Confirmatory testing elements
- Treatment elements
- Screening elements

# Example of success: Severe Combined Immune Deficiency (SCID)

- 9/2007: nomination
- 2/2009: evidence review completed and discussed; AC votes no due to evidence gaps to be addressed
  - prospective identification of at least one confirmed case of SCID through a population-based newborn screening program
  - demonstrated willingness and capacity of additional states to implement newborn screening for SCID
  - reproducibility of the screening test and continuance of a false positive rate of less than 0.1 percent
  - creation of a laboratory proficiency testing program through the CDC's National Quality Assurance Program

# SCID

- 1/2010:  with gaps addressed, AC votes to recommend with
  - NIH funding surveillance activities for outcomes
  - HRSA funding education and training materials
  - CDC developing and distributing quality control/quality assurance specimens
- 5/2010: Secretary of HHS adopts the recommendation

# SCID

- Enabling elements:
  - High throughput assay for TREC from blood spots for screening test
  - Flow cytometry for diagnosis
  - CDC-and NIH-funded pilots and other support
  - Effective treatment (bone marrow transplant) available
  - Education materials including decision support materials developed and disseminated
  - Regional efforts for testing, diagnosis and treatment

# SCID

- Barriers to implementation:
  - Lack of cost benefit information
  - Budgetary concerns (cost estimates for technology infrastructure estimated at $500,000–$1 million)
  - Prior commitment to implement other screening tests mandated by State legislation
  - Lack of the widespread availability of experts in immunodeficiency within a State for diagnosis and treatment
  - Lack of an FDA-approved or -cleared assay

# Additional benefits

- The SCID experience provided additional benefits though the specific linking of different testing modalities and connections and collaboration throughout the broader NBS community that created a favorable environment for the much more rapid implementation of SMA screening

# Questions?

# Next steps and discussion

- Clarify expectations regarding the time to implement a new condition in a state, given the complexity of system issues
- Gathering/analyze data on enablers and challenges across the states
  - For example, an anonymous survey on what has supported implementation and what challenges have delayed it
- Evaluate feasibility of creating an implementation toolbox with strategies to approach specific barriers