

# Pilot Study Workgroup Recommendation on Required Data Elements



**Joseph A. Bocchini, Jr. MD**  
Professor and Chairman  
Department of Pediatrics  
Louisiana State University Health Sciences  
Center – Shreveport  
Chairperson, ACHDNC

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# Background

- Evidence review process dependent on quality data
- Pilot studies are essential to yield evidence about several aspects of NBS systems
- The Public Health Service Act 42 U.S.C. 217a requires that ACHDNC must vote on nominated condition no later than 9 months after having initiated the external evidence review

# Charge to the Workgroup

- Recognize and support current efforts regarding pilot studies and evaluation
- Identify other resources that could support pilot studies and evaluation
- Identify the information required by the Committee to move a nominated condition into the evidence review process (i.e., define the minimum pilot study data required for a condition to be accepted for evidence review)

# Recommendations

1. Data should be available on the analytical validation of one or more screening modalities proposed for use in population-based screening in newborns.
2. Data should be available on the net benefits of clinical interventions following early detection compared to clinical diagnosis.
3. **Data should be available from pilot studies involving population-based screening of identifiable newborns.**
4. Continued support should be provided for NIH initiatives relevant to pilot studies in newborn screening including NBSTRN, NSIGHT, Pilot Studies grants, Natural History grants, Innovative Therapies grants, and grants supported under the Parent Announcement.
5. Continued support should be provided to CDC for its activities relevant to pilot studies that address technical training and quality materials for state laboratories, assistance to state and other programs in obtaining laboratory equipment, creation and distribution of “Validation Test Packages,” population surveillance, and fostering of “Laboratories of Excellence.”
6. DHHS should support the development of a research network comprised of state-based public health programs, laboratories, and academic or other research centers that would provide a stable, experienced, compliant, efficient, and quality infrastructure for the conduct of population-based pilot studies for newborn screening.

# Recommendation #3: Current

- Data should be available from pilot studies involving population-based screening of identifiable newborns.
  - 3A) The study should be sufficiently large to identify at least one true positive, clinically affected newborn for the condition under consideration, and
  - 3B) The population included in the pilot study, and the screening protocol used, should be similar to the US population and to state NBS programs with respect to known prevalence of the condition, and the timing and approach to screening. The screening modality used in the pilot study should be comparable to the method proposed in the application.

Scott Shone, PhD

# **PILOT STUDY RECOMMENDATION – ONE CASE REQUIREMENT**

# **Recommendation 3: Proposed**

- **3A) The study should evaluate the newborn screening process from collection through diagnosis and identify at least one screen positive newborn with confirmation of presence of the condition under consideration.**