Pilot Studies Work Group

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Pilot Studies Work Group

- **Membership**
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  - Kellie Kelm
  - Dieter Matern
  - Mark McCann
  - Melissa Parisi
  - Scott Shone
  - Tiina Urv
  - Mike Watson

Pilot Studies

- General recognition that evidence review process requires evidence
- Challenges
  - Rare conditions
  - Population based research is complex and expensive
  - Section 12 of the NBSSLRA requires informed consent for use of DBS

Pilot Studies

- Consent issues
  - Section 12 requires parental consent for use of DBS
  - Eliminates the ability to conduct federally funded research that involves adding a new screening test on a pilot basis on an opt-out basis or with a simplified consent process
  - Consent processes reduce uptake substantially

Pilot Studies

- Notice of Proposed Rule Making for human subjects regulations is pending
- Comment period followed by drafting of final rules
- For the time being, “pilot studies” in the context may require either consent or to be conducted through state mandated systems (would not be federally funded research)

Charge to the Committee

- 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)

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Focus

- The question is what data are the minimal necessary to move a nominated condition to the evidence review process.
- NOT what evidence is necessary to approve a condition for the RUSP

ACHDNC Nomination Form

- For a nominated condition to be considered there are 3 core requirements
  1. Validation of the laboratory test
  2. Widely available confirmatory testing with a sensitive and specific diagnostic test
  3. A prospective population based pilot study

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ACHDNC Track Record

- The absence of a “pilot study” has been a consistent fault identified in applications in decisions not to move a condition to evidence review
- Additional clarity on the nature of the “pilot study” is necessary

“Pilot Studies”

- Used in the literature for a variety of types of studies in this domain
  - Test validation studies
  - Testing of anonymous dried blood spots
- The term “pilot study” is non-specific
- Better clarity in the type of study necessary to move a nomination forward

Proposed requirements

- How does a screening test perform on a population-based sample in terms of clinical validity?
- Existing requirement
  - A prospective population based pilot study
- Proposed requirement
  - “A prospective population based evaluation of newborn screening and patient identification”

Proposed Requirement

- Stipulations
  - Newborns screened should be identifiable and their clinical status evaluated to determine the clinical validity of the screening test result
  - At least one affected newborn should be detected through population screening
  - The evaluation need not demonstrate clinical utility as long as other data are submitted to address the utility of screening
  - The screening evaluation should be conducted in an appropriate population, that is, one that adequately represents the US population that will be screening in NBS programs for the condition at hand

Process

- What process should the ACHDNC use to determine if the criteria have been met to move a condition to evidence review?