Evidence-Based Reviews for Newborn Screening

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The Nomination Process

- Step #1: The Nomination Form
- Step #2: Federal administrative review
- Step #3: Review by ACHDGDNC
  A) ACHDGDNC review
  B) External evidence-based review by Evidence Review Group
  C) ACHDGDNC review and decision
Nominations and Review

Nomination Form → Federal Administrative Review → Advisory Committee

Recommendations to the HHS Secretary

Evidence Review Group
Applying the Nomination Form

- Broader dissemination by MCHB
- Definition of several terms
  - Severity
  - Accuracy (test)
  - Available (test)
  - Efficacy (treatment)
  - Urgency (treatment)
  - Risks (of screening and treatment)
  - Sensitivity and specificity floors
- Evidence Review Group will provide proposed definitions to committee and for public comment prior to next meeting
Pilot studies

- Review of nomination form and information by ACHGDGDN may indicate need for more population data prior to evidence review, e.g.,
  - Testing and treating a condition in one State using another State as a control
  - Better evidence of prevalence
  - Screening effectiveness in population application
Issues in Evidence Review

- Rare conditions
  - Lack of randomized trials in many cases
  - Limited information on costs and benefits across all potential outcomes (ie, true and false positives and negatives)

- Access to evidence
  - Published evidence
  - Investigator findings (unpublished)
  - FDA trials database
  - Proprietary data
Paper on Pitfalls in Developing Evidence

- Main author: Alex Kemper, MD, MPH, MS
- Based on October 2006 meeting and work done using Pompe disease as a prototype model
Evidence Review Group at the MGH Center for Child and Adolescent Health Policy

- Core staff
  - Program director (policy, chronic conditions) James Perrin, MD
  - Project director (epidemiology/methods) Diane Romm, PhD, acting
  - Public health (from Mass. Dept of Public Health)
  - Consumer (Trish Mullaley, RN)
  - Cost/benefit analysis (Lisa Prosser, PhD)
  - (General) genetics (Marsha Browning, MD, MPH)
  - Health services research fellow (Ellen Lipstein, MD)
  - Methods and screening (Alex Kemper, MD, MPH, MS)
  - Consultant: Nancy Green, MD
Evidence Review Group
External Support

- Assisted by members of ACHDGDNC and *ad hoc* expertise for specific disorder(s)
  - Clear conflict of interest policy
- External advisory group – broader national representation and review
  - Ned Calonge, MD (Colo State Health Dept)
  - Robert Davis, MD, MPH (Ctr for Hlth Research, Kaiser Southeast)
  - Celia Kaye, MD, PhD (Univ of Colorado)
  - Ed McCabe, MD (UCLA)
Evidence Group Efforts

- Developing draft definitions of key terms for committee to review and public comment; finalize definitions based on comment prior to next meeting
- Final template for evidence reviews determined – available at this meeting
- Evidence group to carry out review(s) for spring meeting, based on Committee review of nominations
Evidence Review Template

- **Background**
  - Condition (prevalence, genetics, natural history, different forms of condition)
  - Rationale for current review (why now?)

- **Methods**
  - Data sources
    - Human only; exclude case reports
    - How obtained and evaluated
  - Decision model and evidence questions
  - Data abstraction (and any new analyses)
  - Focus groups of experts (parents and investigators) as needed to estimate severity and burden
  - Screening and diagnostic testing
  - Treatment (risks, benefits; applicability to what condition groups)
Evidence Review Questions and Outcomes

- Key general questions
  - Natural history, including variations
  - Prevalence
  - Burden and severity
  - Methods of screening and diagnosis
  - Treatment effectiveness and variations (incl. Benefits and risks)
  - Costs of screening and treatment
- Indicate where evidence is absent and what information would be most critical
- Presentation of evidence in summary and table form for AC review
- All decisions by AC – evidence group makes no recommendations