Lessons Learned: Newborn Screening (Part One)

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Disclaimer

• These comments do not necessarily reflect the opinion of the Evidence Review Group, the ACHDNC, or the Health Resources and Services Administration
Lessons Learned

“And this is my cousin Dave, who handles the conventional wisdom.”
Sensitivity and specificity are a small part of the overall consideration

1. Epidemiology, clinical detection, and usual care
2. Screening
3. Short-term follow-up and diagnosis
4. Benefits and harms of screening and diagnosis, unrelated to treatment
5. Treatment and long-term follow-up care
6. Outcomes from screen-detected vs. clinically detected cases
Case definitions are central to evaluating overall screening effectiveness

• Case definition should reflect patient-level conditions
  – Not biomarker
  – Not genotype

• Assessment depends on *a priori* determining the goal of screening
Remember that standardizing data collection is critical

• Terminology evolves over time
• Agreement is needed on
  – Nomenclature
  – Approach to data collection
  – How data can be linked for clinical and research purposes
Eventually secondary targets become a primary concern

- Early vs. late-onset
- Carrier status
- Related conditions

Critical for
  - Program operation
  - Evaluation of effectiveness
Epidemiological certainty is ephemeral

- No such thing as natural history
- Case definitions, outcomes, treatments are constantly in flux
- Differences across centers
  - Case definitions
  - Care delivery
Novel partnerships are central to success

- Screening test developers
- Clinician investigators
- Health care providers
  - Specialists
  - Generalists
- Advocacy Groups
- Industry
- The public
- Newborn screening programs and other public health groups
- Payers
- State agencies
- Federal agencies
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• Case definitions are central to evaluating overall screening effectiveness
• Remember that standardizing data collection is critical
• Eventually secondary targets become a primary concern
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• Novel partnerships are central to success
It doesn’t seem right to stop at 6 lessons
Never Underestimate the Challenge of Adherence – Dietary or Otherwise
Outcomes That Are Rare Do Happen
Look to Share Your Lessons – Toot Your Own Horn
Attend APHL’s Newborn Screening and Genetics Testing Symposium
Final Thoughts

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Final Thoughts

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Now, Part Two