Challenges in the Genomic Sequencing and Newborn Screening Disorders Program

Anastasia Wise, PhD
Division of Genomic Medicine
National Human Genome Research Institute
NBS in the Genomic Era

- Examine what genomic sequencing adds to newborn screening (NBS)
- Move towards integrating genomics in health care, anticipate challenges
- Explore ethical, legal, and social implications
- Address clinical validity and utility
- Focus on higher-risk individuals
- Incorporate longitudinal data
Advisor Comments

- ELSI concerns over population selection and study design
- Concern over public perception, public relations, and consent
To explore, in a limited but deliberate manner, opportunities to use genomic information for broadening our understanding of diseases identified in the newborn period.
Research Questions

Must address one or more of the following:

A. For disorders currently screened for in newborns, how can genomic sequencing replicate or augment known newborn screening results?

B. What knowledge about conditions not currently screened for in newborns could genomic sequencing of newborns provide?

C. What additional clinical information could be learned from genomic sequencing relevant to the clinical care of newborns?
Required 3 Components

Genomic Sequencing (C1)

Clinical Research (C2)

Ethical, Legal, and Social Implications (C3)
Good medicine or TMI? What happens if you screen all of a baby's genes?

Maggie Fox, NBC News

Sep. 4, 2013 at 5:29 PM ET

The U.S. government is funding an ambitious set of studies into what happens if you screen the entire genome of newborns. The goal is to find out whether such testing brings better health care or unwanted information overload.
Questions from FDA

- Need for Investigational Device Exemptions for next generation sequencers
- Thinking about device not process (CLIA/CAP)
- Concern over sequence data being returned for clinical use or placed in EMRs

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Any Questions?

THANKS!