

Frontiers of Population
Genomics
ELSI Concerns/Challenges

Barbara A. Koenig, Ph.D.
Professor of Medicine
Mayo Clinic

Evaluation of Genomic Applications in Practice & Prevention



Perspective

- Implications of data bases in 5, 10, 50 years (predictions difficult)

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- Science dynamic; ELSI stale

Overarching Social Concerns

- Passage of effective anti-discrimination legislation
 - GINA remains stalled
- Implementation of appropriate oversight to assure
 - Analytic validity
 - Clinical validity
 - Clinical utility

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- Background
 - Shifts in research funding
 - Growth of consumer-driven health care
 - Ascendancy of market approaches

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 - Ascendancy of market approaches
- **Absolute privacy protection appears to honor choices of “consumers”**
 - **Fear: Denial of long-term research to document genomic associations and demonstrate clinical utility will have the irony of providing consumers “choice” among unproven genomic products**

The “Identifiability” Problem

- Simple relationship between privacy & research utility of DNA collections
 - as research value increases
 - possibility of de-identification decreases

“Racial” Categorization

- How should we account for population sub-structure in population-based studies?
 - A lingering concern
 - Harms are outside the scope of current human subjects protection regime

Concerns of Special Populations

- Disorders/conditions with behavioral components
 - Addiction

“Adequacy” of Informed Consent

- Should we question traditional framing?
- How much “work” can (and should) informed consent do?

Community Engagement

- Various techniques have been tested
 - Town hall meetings (open or targeted)
 - Deliberative polling (online)
 - Community engagement
- Optimal approaches undefined
 - Approach as citizens?
 - Focus on micro-communities?

Routinization of Biospecimen Collections

- Widespread penetration of “Biobanks”
- Breakdown of research/care/public health distinction
- Benefits of “passive research” to populations

Transformation of the Research/Health Care Boundary

- The fiction of de-identification & return of results
- Obligations to disclose results
 - Will public expectations change?
 - Should we reconsider our stance?

Ethical Issues across the Translational Research Spectrum

- Consumer-based approaches
- Planning for catastrophe

Questions for Discussion

- How to move beyond consent?
- What methods of community engagement shall we employ?
 - How to assure authenticity
- Normative/descriptive divide
 - Should opinion rule?
- Priority setting in population genomics

Questions for Discussion

- Genes predicting good health and disease resistance (Manolio)
- “Normalcy”