

SACGHS Session on Genomic Data Sharing

Charmaine Royal, Ph.D.

Secretary's Advisory Committee on Genetics, Health, and Society

June 16, 2010

Issue Statement

- Genomic data sharing is an important tool for advancing basic science and for making strides in our knowledge of common diseases.
- Collection and broad sharing of individual genomic data facilitates important research but sharing such data, even when de-identified, has ethical implications for informed consent, privacy, and discrimination.

SACGHS Steps Taken to Date

- Dec. 2008: Genomic data sharing was identified as one of seven study priorities.
- Sep. 2009: HHS-ASPE contracted The Lewin Group to develop a report for DHHS on key policy questions and to provide analytic support for SACGHS efforts.
- Oct. 2009: A SACGHS steering group was formed and a session on genomic data sharing organized for Feb. meeting.
- Feb. 2010: The session explored various models of genomic data sharing.

SACGHS Steering Group

SACGHS Members

Charmaine Royal (Chair)

David Dale

Sheila Walcoff

Ad hoc Members

Kevin Fitzgerald

Sylvia Au

Julio Licinio

Ex Officios

Michael Amos, NIST

Douglas P. Olsen, DVA

Laura Lyman Rodriguez,
NIH

Michele A. Lloyd-Puryear,
HRSA

Staff Lead: Symma Finn

Session Goals

- Confirm the central issues in genomic data sharing that are relevant for SACGHS consideration.
- Discuss these central issues and their policy implications.
- Decide next steps.

SACGHS Fact Finding Activities

- Literature Review
 - Focused on the blurring between research and clinical care, genomic literacy and risk communication, and provider and research subject attitudes about genomic research
- Consultations
 - Two Program Directors
 - Government genomic medicine program
 - Consumer disease registry
 - Three Secondary Data Users
 - Ethics Researcher

Literature Review

- Implications of the blurring between research and clinical care for
 - The timing and nature of informed consent
 - The articulation of risks and benefits
 - Provider-patient communication; genomic literacy; clinical utility; understanding complex risk and behavioral change
 - Incidental findings and return of research results
 - Resource allocations, time constraints
- Privacy and security – reasonable expectations
- Group harms; health disparities; public health activities

Consultations – Two Program Directors

- Consumer-control in genomic programs entails formalized mechanisms for patient input into policies, program goals and authorization for uses of data
- Returning research results to participants is generally done through websites and newsletters, patient education conferences and involves non-technical aggregated summaries of findings
- The perception of the potential for a data breach was the greatest challenge encountered by the Registry; successes for both programs are numerous studies and publications (the governmental program had more than 46,000 articles in 7 years)
- Although some data on environmental exposures is included in genomic datasets, family history is not and the ability to link with EHRs or longitudinal data is under development.

Consultations – Three Secondary Data Users

- Genomic data has innumerable potential secondary uses; secondary users are eager to mine what is available.
- There is a lack of clear guidance regarding who is responsible for communication of incidental findings.
- Secondary users experienced some difficulties in the application process, but many difficulties in preparing and reformatting datasets for secondary research use.
- The biggest barriers to data sharing are lack of standards for characterizing phenotypic data and lack of incentives to make data easier for secondary research use.

Consultations – Bioethics Researcher

Previous research by McGuire et al. found participants have a desire for information and control over decisions about sharing their genetic and genomic data.

- Current study looks at unavoidable trade off between privacy protection and advancing research, and the variability in judgments about this trade off.
- Randomized trial of 3 models of informed consent for data sharing and secondary use of data:
 - Traditional informed consent
 - Binary informed consent
 - Tiered informed consent

Consultations – Bioethics Researcher

- There is a gap between reported understanding and actual understanding of study goals, use of samples and who is authorized to access and share data.
- Traditional and binary consent models did not provide as many options to participants as a tiered consent model.
- Participants gave equal importance to privacy protection and advancing scientific research.
- The majority of participants (80%) feel it is important to be involved in sharing decisions.
- 65% of participants want to see all of the data sharing options.

Central Issues in GDS

- Implications of the blurring line between research and clinical practice including public health activities.
- Potential for group harms.
- Reasonable expectations of privacy with current and anticipated information technology capabilities.

Central Issues in GDS – Blurring

- **Adequacy of informed consent** for specimens collected in clinical settings, or during public health activities, that will be used in genomic research, stored and shared with other researchers
- **Provisions for return of research results**, and who is responsible for communicating clinically important findings to research participants
- **Adequacy of existing education** for providers and research participants about the meaning of genomic findings

Central Issues in GDS – Group Harms

- Existing informed consent materials are often limited in addressing the potential for group harms
- Community engagement is an approach that has been used to address this issue
- Guidance is needed to appropriately involve communities

Central Issues in GDS – Group Harms

- Arizona Court of Appeals. Havasupai Tribe v. Arizona Board of Regents (2008)
- Cultural challenges to biotechnology: Native American genetic resources and the concept of cultural harm (Tsosie, 2007)
- Safari Research in Mexico (Seguin et al., 2008)
- Tribe blasts 'exploitation of blood samples (Dalton, 2002)
- The Harvard case of Xu Xiping: exploitation of the people, scientific advance, or genetic theft? (Sleeboom, 2005)

Central Issues in GDS – Privacy and Security

- In research, there is a tension between accepting uncertainty and trusting versus protecting privacy and harboring distrust.
- This tension is a critical point of separation between informed consent (IC) in data sharing and typical research informed consent. Unlike typical IC, the concern in genomic data banking and sharing is not material but entails possible inappropriate use of data.

Questions about the Blurring

- How can we design informed consent documents with the information that patients most want?
- How do we design informed consents that are both meaningful to patients and that allow for sharing of data for future research that cannot be anticipated today?
- Is a one-time consent adequate? How broad can this consent be? Are there circumstances where it is appropriate to waive the requirements for informed consent?
- Who is the best person to obtain participants' informed consent for genomic research in clinical settings?
- How should secondary and incidental findings be handled?

Questions about Group Harms

- What steps can be taken to prevent group harms, e.g., what is lost when genomic researchers don't identify groups in designing and conducting genomic studies and in publications?
- Are there best practices for raising awareness among researchers and clinicians about the potential influence of diverse values and beliefs on research participation?
- Whose responsibility is it to think about or address the potential for group harms?
- Are there additional concerns that need to be addressed related to genomic research and groups?

Questions about Privacy and Security

- Should informed consent policies relating to privacy be reconsidered in light of changing views of privacy in our society and the increase in sharing personal information online?
- Do existing policies for genomic data sharing have provisions for notification of breaches of security?
- Should privacy concerns be de-emphasized as a “risk” and a new perspective promoted that places greater value on the public health and societal benefits of genomic research?

Next Steps

- Should SACGHS continue to pursue this topic?
- If so, what should be the focus of SACGHS's work related to genomic data sharing?
 - The implications of the blurring line between research and clinical practice including public health activities?
 - The potential for group harms?
 - Reasonable expectations of privacy, de-identification and informed consent?
 - International issues such as policy and regulatory differences that affect broad genomic data sharing?
 - Other?