Bringing Genomics to Medicine: Advancing the Research & Policy Agenda

Laura Lyman Rodriguez, Ph.D.
National Human Genome Research Institute

Genetic Alliance: Privacy and Progress Webinar Series
Facilitating Progress in Whole Genome Sequencing
July 9, 2013
Rec 4.1: Funders of research, clinical entities, and the commercial sector should facilitate explicit exchange of information between researchers and clinicians, while maintaining robust data protections, so that WGS data and health data can be shared to advance genomic medicine.

Rec 4.2: Policy makers should promote opportunities for the public to benefit from WGS research and all should promote opportunities to explore alternative models to engage participants in research participation to promote collaborative relationships.
The Relevance of Genomics

Biomedical Researchers

Healthcare Professionals

Patients (and Friends & Relatives of Patients)
Kathryn discovered her Ancestry by DNA™

"I was surprised to learn that I'm as much as 24% Indigenous American. It must come from a closer relative than I thought. I can't wait to share this with my family."

<table>
<thead>
<tr>
<th>European</th>
<th>Indigenous American</th>
<th>Sub-Saharan African</th>
<th>East Asian</th>
</tr>
</thead>
<tbody>
<tr>
<td>67%</td>
<td>24%</td>
<td>9%</td>
<td>0%</td>
</tr>
</tbody>
</table>

Bar Graph of Results

Kathryn Baker
Toledo, OH

Additional Ancestry DNA Tests Available:

- Paternal Lineage (Y-STR) Testing
  This type of DNA testing determines what migration routes your ancient paternal ancestors took.
  Go to page

- Maternal Lineage (mtDNA) Testing
  This type of DNA testing determines what migration routes your ancient maternal ancestors took.
  Go to page
Participants Driving Research Momentum

Live better, together

making healthcare better for everyone through sharing, support, and research

Privacy and access in perfect balance
and access to critical health information.

23andWe Research

We're interested in advancing genetic research. Get involved and learn about yourself while contributing to science.

23andWe isn't just about you. Our research arm, 23andWe, gives customers the opportunity to leverage their data by contributing it to studies of genetics. With enough data, we believe 23andWe can produce revolutionary findings that will benefit us all.

Get involved in a new way of doing research.

Our customers are saying...

"I'm more interested in the researching aspects of it. The fact that you guys are actually asking questions and relating that to ongoing research I think is interesting. That direction is interesting. It just seems like it's a great repository of information." ~ John G.
Moving from Anecdote(s) to Common Practice
Goal: Iterative Dialog and Process

Ethical Norms

Issue
Opportunity
Need
Values

Policy

Research
‘Hot Areas’ in Genomic Medicine

- Cancer Genomics
- Pharmacogenomics
- Rare & Common Genetic Diseases
- Prenatal & Newborn Genomic Analysis
- Clinical Genomics Information Systems
- Genomic Medicine ‘Test Drive’ Programs
<table>
<thead>
<tr>
<th>Program</th>
<th>Goal</th>
</tr>
</thead>
<tbody>
<tr>
<td>eMERGE II</td>
<td>Use biorepositories with EMRs and GWA data to incorporate genomics into clinical research and care, including a pharmacogenomics arm (~9,000 patients)</td>
</tr>
<tr>
<td>CSER &amp; RoR</td>
<td>Explore infrastructure, methods, and issues for integrating genomic sequence into clinical care, including investigations into whether/when/how to return individual research results to ppts</td>
</tr>
<tr>
<td>CRVR</td>
<td>Develop and disseminate consensus information on variants relevant for clinical care</td>
</tr>
<tr>
<td>GMPDP</td>
<td>Develop and disseminate methods for incorporating patients’ genomic findings into their clinical care</td>
</tr>
<tr>
<td>Newborn Sequencing</td>
<td>Explore possible uses of genomic sequence information in the newborn period</td>
</tr>
<tr>
<td>CMG &amp; UDN</td>
<td>Identify genes for rare diseases. Diagnose both rare and new diseases by expanding NIH’s Undiagnosed Diseases Program</td>
</tr>
</tbody>
</table>
Genomic Medicine Working Group

- Convene Genomic Medicine meetings
  - GM programs, Institutions, Laboratories & Payers, Professional Societies, Federal Agencies…International

- Provide guidance to NHGRI in other areas of genomic medicine implementation, such as:
  - Outlining infrastructural needs for adoption of genomic medicine
  - Identifying related efforts for future collaborations
  - Reviewing progress overall in genomic medicine implementation
Key Policy Issues *(selected)*

- Oversight & Governance (& Regulation)
- Participant Autonomy
- Privacy
- Data Sharing
- Healthcare Readiness
- Access
New and Changing Interactions
Need for New Paradigms

“The Research-Treatment Distinction: A Problematic Approach for Determining Which Activities Should Have Ethical Oversight

BY NANCY E. KASS, RUTH R. FADEN, STEVEN N. GOODMAN, PETER PRONOVOST, SEAN TUNIS, AND TOM L. BEAUCHAMP

“...a new ethical foundation needs to be developed that facilitates both care and research likely to benefit patients, and that provides oversight that, rather than being based on a distinction between research and practice, is commensurate with the risk and burden in both realms...”
Updating the Common Rule

Goal: Enhance effectiveness of research oversight by improving protections and reduce burdens, delays, and ambiguity for investigators and subjects

Topics:
- Risk-based framework
- Improve consent forms
- Harmonization
- Improved monitoring & oversight of the system
- Streamlined IRB review
- Extend protections to all research
- Establish data security & information protection

Advanced Notice of Proposed Rule Making (ANPRM)
Read more about the July 22, 2011 ANPRM for changes under consideration to the Common Rule. These changes, the most extensive since the Department of Health, Education, and Welfare published proposed rules for the protection of human subjects involved in research on August 14, 1979, are available for public comment until September 26, 2011.
From patients to partners: participant-centric initiatives in biomedical research

Jane Kaye, Liam Curren, Nick Anderson, Nadja Kanellopoulou, David Lund, Daniel James Shepherd, Patrick L. Taylor, Shara Bywater

Abstract | Advances in computing technology and medical research is increasingly characterised by large numbers of researchers that are reliant on large datasets, as well as a number of challenges for obtaining consent, addressing patient concerns and maintaining public trust. Participant-centric approaches provide the basis for long-term interaction and engagement with patients and provide the basis for long-term interaction and engagement with patients. This article provides an overview of this rapidly moving field by presenting participant-centric approaches, as well as the benefits and challenges that come with them.

Table 1 | Key functions of participant-centred initiatives

<table>
<thead>
<tr>
<th>Function</th>
<th>Characteristics</th>
<th>Examples</th>
</tr>
</thead>
<tbody>
<tr>
<td>Metchmaking</td>
<td>Brings together participants and researchers by either promoting communication or facilitating recruitment</td>
<td>PrivateAccess (USA)</td>
</tr>
<tr>
<td>Direct-to-consumer services</td>
<td>Provides participants with services as well as social-networking capabilities</td>
<td>23andWe (USA)</td>
</tr>
<tr>
<td>Dynamic negotiation</td>
<td>Enables an ongoing discourse and negotiation between researchers and participants</td>
<td>CuraRata and String of Pearls Initiative (Netherlands)</td>
</tr>
<tr>
<td></td>
<td>Enables participants to manage their preferences for personal data sharing while facilitating more accountable research governance</td>
<td>CHRIS — Cooperative Health Research in South Tyrol (Italy)</td>
</tr>
<tr>
<td>Citizen science</td>
<td>Allows participants to provide and to control the samples and data and, in so doing, to have an active involvement in facilitating research</td>
<td>PatientsLikeMe (USA)</td>
</tr>
<tr>
<td></td>
<td>Allows participants to drive the research agenda and to carry out their own research projects</td>
<td>TuAnalyze (USA)</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Genomes Unzipped (UK)</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Genomera (USA)</td>
</tr>
</tbody>
</table>
International partners describe global alliance to enable secure sharing of genomic and clinical data

By Broad Communications, June 4th, 2013

Over 70 leading health care, research, and disease advocacy organizations that together involve colleagues in over 40 countries have taken the first steps to form an international alliance dedicated to enabling secure sharing of genomic and clinical data. The cost of genome sequencing has fallen one-million fold, and more and more people are choosing to make their genetic and clinical data available for research, clinical, and personal use. However, interpreting these data requires an evidence base for biomedicine that is larger than any one party alone can develop, and that adheres to the highest standards of ethics and privacy. These organizations recognize that the public interest will be best served if we work together to develop and promulgate standards (both technical and regulatory) that make it possible to share and interpret this wealth of information in a manner that is both effective and responsible.
Data Sharing Initiatives from the Top

**Action 2: Release of high value data**

- We recognise the following as areas of high value, both for improving our democracies and encouraging innovative re-use of data.

<table>
<thead>
<tr>
<th>Data Category (alphabetical order)</th>
<th>Example datasets</th>
</tr>
</thead>
<tbody>
<tr>
<td>Health</td>
<td>Prescription data, performance data</td>
</tr>
<tr>
<td>Science and Research</td>
<td>Genome data, research and educational activity, experiment results</td>
</tr>
</tbody>
</table>
### Notice on Development of Data Sharing Policy for Sequence and Related Genomic Data

**Notice Number:** NOT-HG-10-006

**Key Dates**
- **Release Date:** October 19, 2009

**Issued by**
- National Human Genome Research Institute (NHGRI); [http://www.genome.gov](http://www.genome.gov)

**Purpose**

The purpose of this Notice is to inform the research community of plans by the National Institutes of Health (NIH) to:

1. Update data sharing policies for NIH supported research, including extramural and intramural projects, involving sequence and related genomic data obtained with advanced sequencing technology (e.g., medical resequencing data, sequence data from non-human species, including microorganisms, transcriptomic and epigenomic data, as well as data needed for interpretation, including associated clinical, other phenotype and metadata, such as supporting study documents and methodologies).

2. Encourage investigators and IRBs to consider the potential for broad sharing of sequence and related genomic data in developing informed consent processes and documents for such studies involving human sequence data and;

3. Communicate the agency’s intent and current underlying considerations related to developing a policy pertaining to the deposition of these large datasets into centralized databases, such as the GenBank/Short Read Archive (SRA) or the Database of Genotypes and Phenotypes (dbGaP), so that they are available as broadly and rapidly as possible to a wide range of scientific investigators.

---

- Extension of the rationale for data sharing policies
- Update expectations for range of genomic data produced today … and tomorrow
- Consider questions of appropriate consent
- Data release and publication policies
ARE WE THERE YET?
Thank you!

laura.rodriguez@nih.gov