Data Sharing, Public Health and the Bioeconomy

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Genomics in Public Health

- Traditionally, public health applications of genomics have focused on **rare disease**. Much of the future promise of genomics rests on its application to **common diseases**.

- Genomics plays a role in 9 of the 10 **leading causes of death**, including: Heart disease, Cancer, Stroke, Diabetes, Alzheimer's disease.

- **Initial public health recommendations** are becoming available: evidence-based interventions for women at risk for hereditary breast and ovarian cancer, and individuals at risk for hereditary colorectal cancer.

- **Precision medicine** has the potential to profoundly improve **public health** and the **economy**.
GWAS: A shortcut for linking particular gene variants to specific diseases with as little sequencing as possible

- ~2,000 robust associations with >300 complex diseases
- Considerable skepticism regarding their clinical applicability: modest effect sizes and unclear functional consequences
- However, there are promising examples that will translate into clinical care:
  - **Risk prediction**: T1DM loci
  - **Disease classification**: CRP for MODY
  - **Drug development**: ITPA and ribavirin toxicity
  - **Drug toxicity**: SLCO1B1 variants and statins

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The Path to Precision Medicine

DNA Sequence

- Robust scientific evidence of effectiveness
- Appropriate risk analysis
- Cost / benefit studies
- Low-cost seq technologies and accessible bioinformatics
- Effective communication of results and counseling
- Policies for reimbursement and coverage
- Other sensitive ethical, educational, economical, legal and social issues.

Clinical Practice
Precision medicine will impact health disparities

- It is key to have principles, guidelines and best practices to draft a common framework to successfully develop precision medicine globally.

- Precision medicine strategies are likely to be different in different parts of the world: industrialized, emerging and developing economies.

- Strategies need to take in account cultural history, values, population, resources, and health priorities.
The planning stages: Genomic medicine will contribute to health equity and development in Mexico*

- It would benefit public health, development of policies, tackle local health disparities
- Timely development of GM would contribute to reduce social gaps
- GM cannot be “imported” from other countries with different ancestry
- It would stimulate scientific research on local health problems, strengthen infrastructure, and competitiveness
- Driver for innovation in a knowledge economy

Sept. 26, 2005
Description of the Haplotype Map of the Mexican Populations

Over 65 different ethnic groups

Proyecto: Variabilidad Genómica y Mapa de Haplotipos en la Población Mexicana

Carta de Consentimiento Informatado
A thorough process for community engagement and informed consent was established for Mestizo and Amerindian populations.
Community engagement and personal consent
Federal involvement and interest in genomic medicine
Results of the Mexican Genome Diversity Project

People from Mexico show stunning amount of genetic diversity

SLC16A11 haplotype from one synonymous and 4 missense SNPs associated to T2D

- T2DM in Mexico has a prevalence roughly 2X of US non-Hispanic whites: GWAS 9.2M SNPs in 8,214 Mexicans and other Latin Americans.
- Each copy of the SLC16A11 haplotype is associated with a ~20% increased risk of T2D in Mexicans.

**Haplotype frequencies**

<table>
<thead>
<tr>
<th>1000 Genomes</th>
<th>SIGMA</th>
</tr>
</thead>
<tbody>
<tr>
<td>AFR</td>
<td>EUR</td>
</tr>
<tr>
<td>64%</td>
<td>97%</td>
</tr>
<tr>
<td>36%</td>
<td>&lt;1%</td>
</tr>
<tr>
<td>0%</td>
<td>&lt;2%</td>
</tr>
</tbody>
</table>

Challenges associated with the emergence of genomics in Mexico

**Progress of science and medicine**
- Genomics for science & health
- Knowledge-based economy
- Innovation
- Competitiveness

**Fear of foreign abuse**
- Foreign groups studying locals
- International pharma
- Genographic project
- Indigenous rights movement
- Anthropologists
- Ecological groups

**Law of Genomic Sovereignty**
DNA samples for population studies to leave the country need to have local IRB approval and will be included in a registry kept by the Department of Health
Scientific innovation can be fragile in emerging and developing economies

- When the leadership has scientific and global experience, major significant progress can be achieved in a short period of time
- Science and technology are usually not funding priorities
- Institutions are not robust enough to follow mid- long-term plans but are too malleable to political calendars and personal interests
- Ineffective mechanisms to disseminate new knowledge to the public
- Public policies way behind the advancement of science

Trends indicating that genomics innovation will contribute to the economy

HGP – Return on investment
[ROI=$141]
The virtuous circle of innovation: Integrating Genomics into the Bioeconomy

CONNECTING ideas and people across sectors to find new uses and applications for genomics

SUSTAINED INVESTMENT in large-scale science and technology to fuel innovation

TRANSLATING discoveries into applications to maximize impact across all sectors

The Organization for Economic Cooperation and Development: 34 country members

OECD + strategic partners >80% of world’s GDP

64% of world’s population
Bioeconomy 2030: Harnessing the latent value in biological systems

- **Bioeconomy** - set of economic activities related to the invention, development, production and use of biological products and processes.

- **Biotechnology** could be responsible for 2.7% of GDP in OECD countries, this number excluding the potential of biofuels.

- **Genomics** shows impact in the whole value chain and important potential to enable economic growth and development.

Areas where genomics can contribute to meet grand challenges
1. **Guidelines for Human Biobanks and Genetic Research Databases.** Assists in the development of policies and provides guidance for private & public sector HBGRDs.

2. **Guidelines on Quality Assurance in Molecular Genetic Testing.** Offers principles and best practices for the QA of MGT offered in a clinical context.

3. **Guidelines on Licensing of Health Care Genetics.** Offers principles and best practices for the licensing of intellectual property rights that relate to genetic inventions used for the purpose of human health care.

4. **Pharmacogenetics: Opportunities and Challenges for Health Innovation.** Examines challenges facing pharmacogenetics across the health innovation cycle and into the clinic, and reviews its impact on both pharmaceutical R&D and clinical care.

5. **Biomarkers and Targeted Therapies.** Strategies to improve the development and use of biomarkers in health care.


Countries with national blueprints and other policies to promote the bioeconomy

USA (2012)  EU (2012)  Belgium  Canada  Denmark  Finland  Germany  Netherlands  South Africa  India  Brazil  Malaysia

Countries with important bioeconomy policies without a formal blueprint

France  Italy  UK  Japan  S. Korea

Work in the area of genetics and genomics undertaken by the OECD

- Describes drivers and criteria shaping the medical applications of genomic biotechnology in different national settings, and the barriers to its implementation both, nationally and internationally.
GA4GH: A federated ecosystem for sharing genomic and clinical data

- **Timely and pertinent global strategy** to boost our knowledge about mechanisms of disease, development of diagnostics and risk predictions, and discovery of new leads for innovation.

- **Innovation since its foundation**: provides basic principles and core elements for responsible data sharing and is founded on the **1948 Universal Declaration of Human Rights**.

- GA4GH enables responsible and effective sharing of genomic and clinical data in a way that is as simple as using the web: Transforming silos of genome data collection into **perfectly connected independent systems** around the globe.

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*GA4GH, A federated ecosystem for sharing genomic, clinical data. Science 352, 1278–1280 (2016).*
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