Personal Genomics
and the Social Sciences and Humanities

(Conclusion of last week’s episode)

FAQ Assignments Due Next Week!

2. The genie is out of the bottle

- Genetic privacy in the Information Age

2. The genie is out of the bottle

- Private Genome Sequencing
  - Private firms have moved ahead of the ethical, legal, and social debates about how the technology should be used, managed or regulated by government.

Private Genome Sequencing

- Direct-to-consumer testing
  - As a result of falling costs, the availability of full human genome sequencing has increased, facilitating the growth of private services like ancestry tracing and “predictive medicine” (a list of genetic variations that may put you at risk for certain diseases).

Private Genome Services: Concerns

- Require lower levels of consumer knowledge and awareness than medical institutions;
- Misinterpretations of test results can lead to stress and misinformed decision making;
- Positive tests can lead to significant medical decisions with wider implications.

23 and Me in the News

- In November 2013, the company was ordered by the Food and Drug Administration (FDA) to halt the marketing and sale of a saliva collection kit and personal genome service test kit, because it was being sold “without market approval.”

- http://www.youtube.com/watch?v=VZXDtTNqDuQ&desktop_uri=%2Fwatch%3Fv%3DVZXDtTNqDuQ&app=desktop

Accuracy?

- Kira Peikoff, "I had my DNA Picture taken, With Varying Results," *New York Times* (30 December 2013).

And so, more concerns…

- No agreed industry standards for evaluating risk factors or reporting language
- The tests these companies used are based on reading segments of DNA (the SNPs Dave talked about) and not the whole genome
- different companies choose different SNPs to read or interpret for the same condition
- the causes of most common diseases remain unknown.

Fetal Testing

"A complete DNA read-out for every newborn will be technically feasible and affordable in less than five years, promising a revolution in healthcare."

"By 2019 it will have become routine to map infants' genes when they are born."

- Illumina CEO Jay Flatley, 2009

Privacy

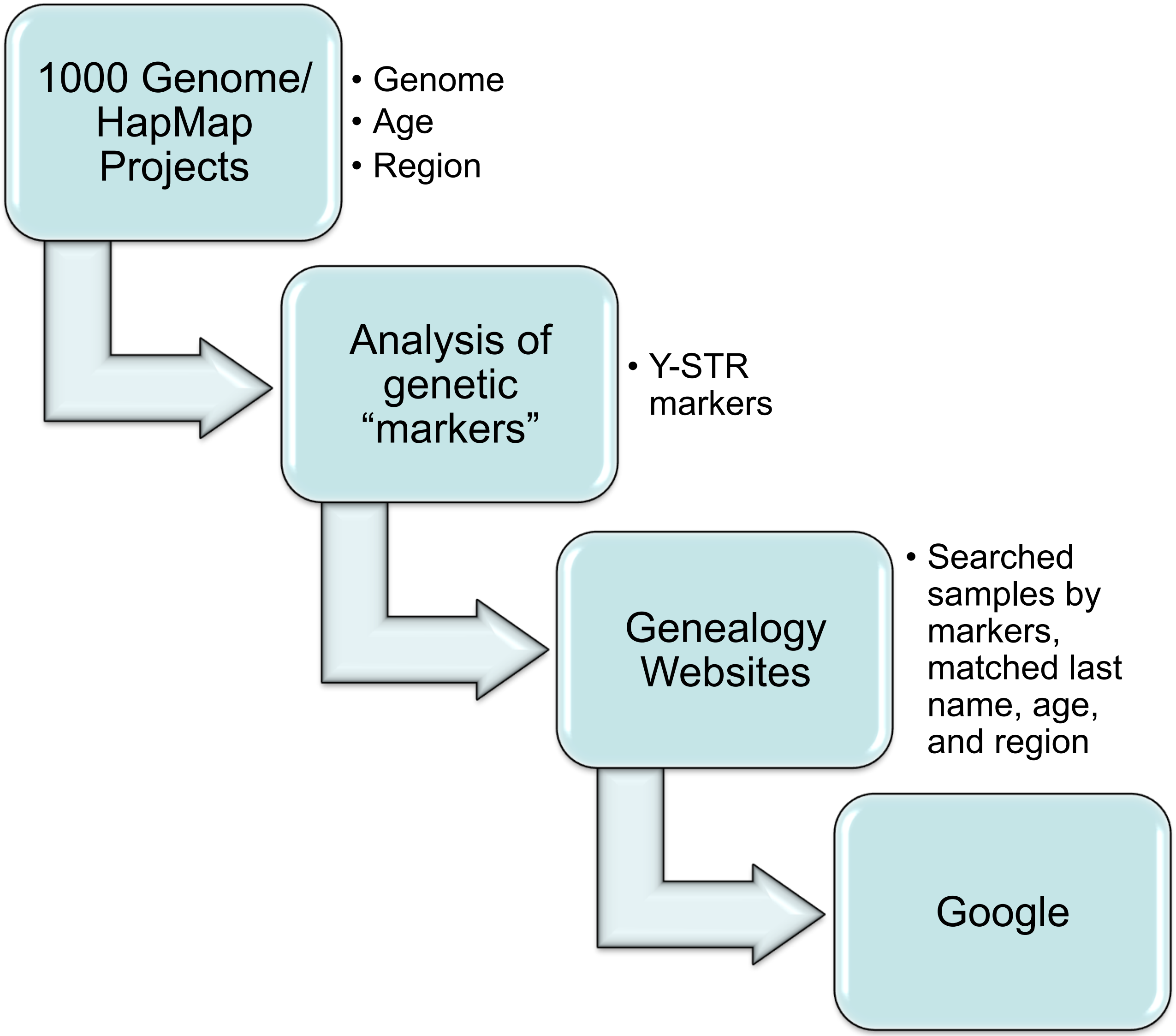
CSI ASIC 200:
From Anonymous Genome to Individual Identity?

A person acquired some anonymous human genomes (only matching info was age and U.S. state)

?

This person was able to match some of those genomes to specific individuals and identify their relatives

Yaniv Erlich's Experiment



1000 Genome/ HapMap Projects

- Genome
- Age
- Region

Analysis of genetic "markers"

- Y-STR markers

Genealogy Websites

- Searched samples by markers, matched last name, age, and region

Google

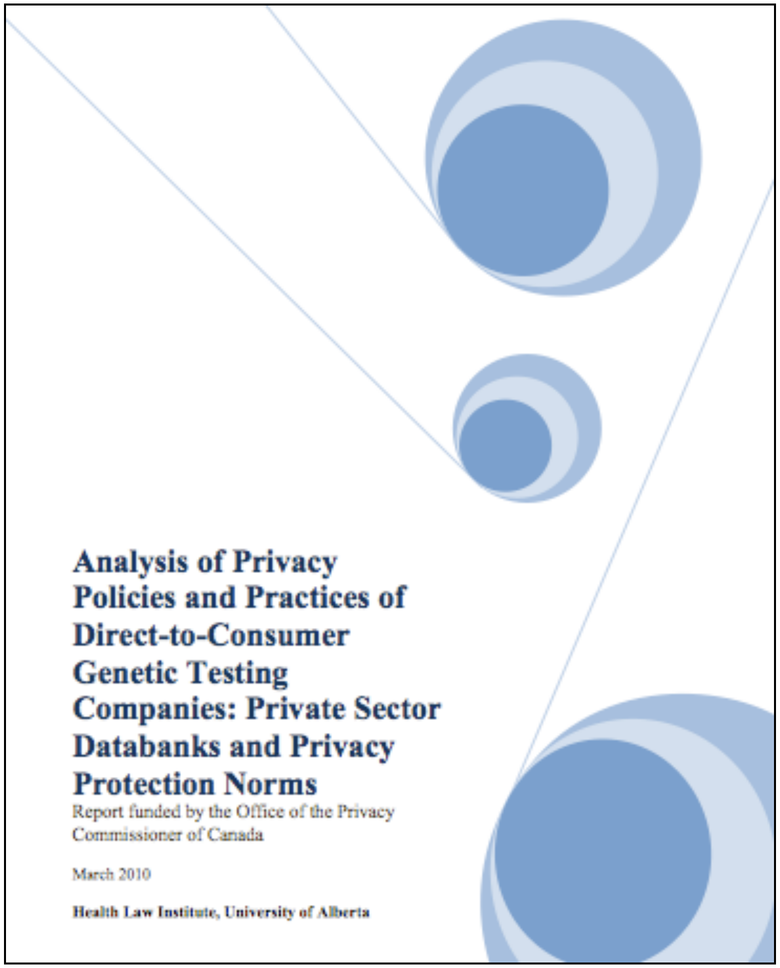
What kind of regulations should be put in place to manage or govern the private sector in human genomics?

In the US…

- 25 states permit direct-to-consumer genetic tests without restriction.
- In the other 25, the most common restrictions are the need for a permit (a state or federal clinical lab certification) and authorization from a physician.

In Canada

There is no regulation of direct-to-consumer genetic testing in Canada under the Health Act or the Food and Drug Act.



Analysis of Privacy Policies and Practices of Direct-to-Consumer Genetic Testing Companies: Private Sector Databanks and Privacy Protection Norms

Report funded by the Office of the Privacy Commissioner of Canada

March 2010

Health Law Institute, University of Alberta

“There is wide variation in the extent to which DTC genetic testing companies address protection of personal information and biological samples. Of the 32 company websites we studied, 20 provide a privacy policy on their websites. Some focus on internet use (e.g. how the website tracks visitors) and some briefly address handling of samples and/or results. Seven of the 20 companies have more comprehensive policies that explain how the company collects and uses information via its website and how samples and results are handled.”

3. Human Evolution and Trait Variation

- Svante Pääbo
  - One of the founders of paleogenetics (evolutionary genetics)
  - In May 2010 published the draft Neanderthal genome in *Science*
  - enabled researchers to isolate differences between the genome of *Homo Sapiens* and Neanderthals

3. Human Evolution and Trait Variation

- A comparison of the genomes of people alive today provides new insights into the migration of *homo sapiens* throughout the world.

3. Human Evolution and Trait Variation

- But what if geographic differences or variations emerge in areas such as intelligence and behavior? Already, genetic studies are examining variations within populations on intelligence, personality type, religiosity, and even the ability to make money.

3. Human Evolution and Trait Variation

- Dr. Spencer Wells, head of the Genographic Project run by the National Geographic Society and IBM, would say probably no.

3. Human Evolution and Trait Variation

- Dr. Yang Huanming, head of the BGI (formerly the Beijing Genomics Institute), would say probably maybe.

3. Human Evolution and Trait Variation

- But…remember the principle of controlling for variables. Intelligence has a hereditary aspect. Parenting and education and life experience matter. But it is also possible that each human brain develops differently.

Trait Variation and Responsibility

"…there can be no doubt that genes do make some contribution to behavioural traits, including fundamental aspects of human character. Since people do not choose their genes, and are therefore not responsible for them, it seems to follow that they are not responsible for these aspects of their character. But if this is so, then how far are they responsible for themselves at all? Does research in behavioural genetics undermine the normal sense of responsibility?"

Nuffield Council on Bioethics

4. Genetic Testing and Medicine

Meet Claudine Wrighter

- 34 years old
- Family history led to doctor recommendation to undergo genetic testing at Dana Farber Institute
- Inherited BRCA1 gene mutation from her mother
- Recommendation of oncologist and genetic counselor: surgical removal of ovaries and fallopian tubes

Claudine Wrighter’s Case

- While her decision was based on carefully considered personal choices in careful consultation with her doctors, cases such as this raise a number of questions:
  - about the advisability of the treatment;
  - about the decision-making process used to make a treatment decision;
  - the nature of the patient-doctor relationship;
  - and affordability and social equity.

5. Ethical Implications

- Genetic research in general, and human genetic research in particular, has been a contentious subject for both religious and secular ethical systems.

5. Ethical Implications

Playing God?

The Tale of The Prince and The Biologist.

Charles Philip Arthur George
(Prince)

- “Mixing genetic material from species that cannot breed naturally, takes us into areas that should be left to God. We should not be meddling with the building blocks of life in this way.”

Richard Dawkins
(Biologist)

- “Almost every morsel of our food is genetically modified – admittedly by artificial selection not artificial mutation, but the end result is the same. A wheat grain is a modified grass seed, just as a Pekinese is a modified wolf. Playing God? We have been playing God for centuries!”

5. Ethical Implications

- Religious perspectives

The Seven Deadly Sins

- Lust
- Gluttony
- Greed
- Sloth
- Wrath
- Envy
- Pride

The Seven Deadly Sins 2.0

- Lust
- Gluttony
- Greed
- Sloth
- Wrath
- Envy
- Pride

- Genetic modification
- Human experimentation
- Polluting the environment
- Social injustice
- Causing poverty
- Financial gluttony
- Taking drugs

5. Ethical Implications

Christian ethicists believe that using genome-sequencing technology to determine behavioral choices should be the lowest priority in personal genetic research.

Instead, genetic research should emphasize the care and management of patients and in developing new treatments. Behavioral genetics, it states, does not do these things.

5. Ethical Implications

In the Islamic world, the approach taken toward genetics is grounded in the decisions of The Islamic Jurisprudence Council of the Islamic World League (Organization of Islamic Cooperation).

5. Ethical Implications

In its 15th session in October 1998, the IJC decided:

1) to permit use of genetic engineering for disease prevention, treatment, or amelioration on the condition that such uses do not cause further damage;
2) to forbid the use of genetic engineering for evil and criminal uses or what is forbidden religiously;
3) to forbid using genetic engineering and its tools to change human personality and responsibility, or interfering with genes to improve the human race;

5. Ethical Implications

4) to forbid doing any research or therapy of human genes except in extreme need, after critical evaluation of its benefits and dangers and after an official consent of the concerned, respecting the extreme confidentiality of the information and human rights and dignity as dictated by Islamic Sharia'ha;
5) to allow the use of bio-engineering in the field of agriculture and animals, on the condition that precautions are taken not to inflict harm (even in the long term) on humans, animals or vegetation.

6. Group Identity

- Human and personal genomics can have a profound impact on cultural, ethnic, linguistic, and indigenous groups.

Group Identity

- Indigenous groups around the world have taken a defensive stance concerning the genetic testing of their populations.

Group Identity

- Indigenous groups worry about how the testing will be used and how it could hurt their cultural identity.

Arizona State University and the Havasupai Indians

"They used our blood
for all these studies,
people got degrees and
grants, and they never
asked our permission."

- Carletta Tilousi (Lead Plaintiff)

7. Consent and Privacy

- Laws and regulations surrounding consent and privacy are still unclear and evolving.

The Genetic Information Nondiscrimination Act (GINA)

- Federal US law (21 May 2008)
- Prohibits discrimination in health coverage and employment on the basis of genetic information.

But…

- GINA’s non-discrimination protections do not extend to life, disability, and long term care insurance.
- GINA’s employment provisions do not apply to employers with fewer than 15 employees
- GINA does not prohibit health insurers or administrators from obtaining and using genetic test results in making health insurance payment determinations.

Around the world…

- UK moratorium on use of genetic information by insurance industry until 2014.
- France and Spain ban all use of genetic test results by all insurers.
- The Council of Europe’s Convention on Human Rights and Biomedicine prohibits genetic discrimination.

And in Canada…

Genetic Discrimination in Canada

- 27-year-old Katie Lingard was recently told by a major insurance provider that she would have to prove that she didn't carry the gene for Huntington disease, a disease that runs in her family, in order to qualify for the life and long-term disability insurance she needed to set up a chiropractic practice.

Genetic Discrimination in Canada

- A 2006 survey of Canadians at risk for Huntington disease found that 39.9% had experienced discrimination.

Genetic Discrimination in Canada

- Life and disability insurance companies were the main source of discrimination, with 29.2% of respondents reporting their applications for coverage were rejected, their premiums were increased, or they were forced to take a predictive test before they could obtain coverage.

Canadian Law

- No specific laws on genetic discrimination exist in Canada.

Canadian Law

- Article 15 of the **Canadian Charter of Rights and Freedoms** guarantees equality and grants each person the right to not be subjected to discrimination. However, the Charter applies only to disputes between people and state institutions.
- The **Canadian Human Rights Act** protects individuals against discrimination based on disability (Art. 3) and could therefore protect individuals whose genetic information reveals the risk of disease and therefore perceived or imputed disability.
- The **Tri-Council Policy Statement** recognizes that discrimination is a risk in genetic research. Researchers who are carrying out genetic research should reveal possible risks to research ethics boards and participants.
- Canada’s **Personal Information Protection and Electronic Documents Act** protects the personal information of individuals.

Canadian Law

- Ten Principles in the Model Code for Protection of Personal Information (incorporated into the **Personal Information Protection and Electronic Documents Act**):

Accountability
Consent
Limiting Use
Accuracy
Openness
Identify Purposes
Limiting Collection
Disclosure and Retention
Safeguards
Individual Access

Canadian Law

- None of these instruments address future disability, perceived disability, or imputed disability.
- None of these instruments are aimed at the prevention of discrimination, instead offering remedies or recourse after discrimination has occurred.

Canadian Law

- A Private Members Bill (C536), titled “An Act to Amend the Canadian Human Rights Act (genetic characteristics)” was introduced in parliament in April 2010.

- It did not become law.

8. Patent Law

- The debate

Patent Law

- The debate
  - Discovering a new function of a known DNA sequence (for example, its association with a particular disease) requires a gene to be isolated and thus could be considered ‘new’, because these copies of genes are ‘made’ (as they do not exist in nature prior to synthesis in a test tube).

Patent Law

- The debate
  - An opposing view argues that genes are discoveries, not inventions and that it is unethical to attempt to privately own a human gene.

Patent Law

- Genomics and patent law: a moving target
- *Association for Molecular Pathology vs. Myriad Genetics and the University of Utah Research Foundation*

Patent Law

- Genomics and patent law: a moving target
- the creation of the world’s first living organism with a completely synthetic genome: JCVI-syn1.0.

The Glofish Debate

- "Because tropical aquarium fish are not used for food purposes, they pose no threat to the food supply. There is no evidence that these genetically engineered zebra danio fish pose any more threat to the environment than their unmodified counterparts which have long been widely sold in the United States. In the absence of a clear risk to the public health, the FDA finds no reason to regulate these particular fish." (US FDA)

- "It's clear this sets a precedent for genetically engineered animals. It opens the dams to a whole host of nonfood genetically engineered organisms. That's unacceptable to us and runs counter to things the National Academy of Sciences and other scientific review boards have said, particularly when it comes to mobile GM organisms like fish and insects." (Center for Food Safety)

9. Global Governance

Guidelines, but no rules

- There are currently no institutions or international treaties solely responsible for the global governance of personal genomics.

Declaration of Bilbao (1993)

- The first international document to address the human genome. The declaration denounces all uses of genetic information causing or leading to discrimination in work relations, in the insurance domain or in any other sector.

UNESCO’s Universal Declaration on the Human Genome and Human Rights (1997)

- proclaims that “[n]o one shall be subjected to discrimination based on genetic characteristics that is intended to infringe or has the effect of infringing human rights, fundamental freedoms and human dignity.”

UNESCO’s International Declaration on Human Genetic Data (2003)

- specifies that “[e]very effort should be made to ensure that human genetic data … are not used for purposes that discriminate in a way that [infringes on] human rights, fundamental freedoms or human dignity of an individual … a family, a group or communities.”

ECOSOC Resolution 2004/09 on Genetic Privacy and Non-Discrimination (2004)

- The United Nations Economic and Social Council’s Resolution 2004/09 on Genetic Privacy and Non-Discrimination (2004) “[u]rges States to ensure that no one shall be subjected to discrimination based on genetic information” and to take the appropriate measures to attain this goal.

The Developing World

- 90% of health research dollars are currently being spent on health problems that affect only 10% of the world’s population.

The Developing World

- Genomic sovereignty
  - a state should exercise control over the genetic material of their populations.
  - desire to develop national expertise and infrastructure in this area, to avoid dependence and domination by foreign science and expertise.

The Developing World

- But…
  - Governments may be thinking of the genetic heritage of their populations as a resource, to be “mined” like any other.
  - Many groups in developing countries worry that governments will use genetic testing against them.