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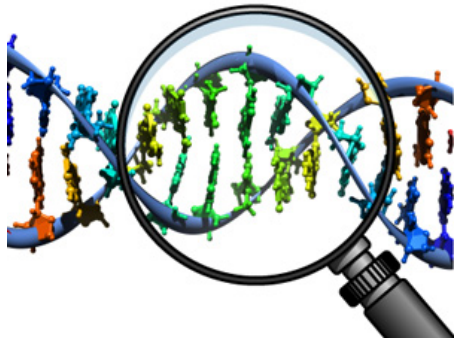
An Exploratory Ethnographic Study of Issues and Concerns with Whole Genome Sequencing

Emiliano De Cristofaro
University College London
http://emilianodc.com

Genomics 101



Genomes…

- Information to build/maintain an organism's living example
- At least one copy of the genome is in almost all cells
- Encoded in DNA (or RNA for viruses)
  - DNA: a double stranded polymer of nucleotides (A, C, G, T)
- In humans, 3.2B nucleotides (in 23 chromosome pairs)

Whole Genome Sequencing (WGS)…

- Determining the complete DNA sequence in a genome

WGS Progress

Some dates

1970s: DNA sequencing starts

1990: The “Human Genome Project” starts

2003: First human genome fully sequenced

2005: Personal Genome Project (PGP) starts

2012: UK announces sequencing of 100K genomes

Some numbers

$3B: Human Genome Project (2003)

$250K: Illumina (2008)

$5K: Complete Genomics (2009), Illumina (2011)

$1K: Illumina (2014)

The Good News

Affordable WGS facilitates the creation of large datasets for research purposes

- Crucial for hypothesis-driven research, e.g., GWAS

Low-cost WGS will bring genomics to the masses

- Large number of individuals will have the means to have their (fully) genome sequenced, and possibly store/retain it

Personalized medicine

- Diagnosis/treatment tailored to patient's genetic makeup

In general, genomic tests can be done "in silico", using specialized computation algorithms

The Bad News

The genome is a unique identifier

- Once leaked, you cannot “revoke” it
- Anonymization / de-identification useless
  - Gymrek et al., Identifying personal genomes by surname inference, Science, 2013

Genomic information is extremely sensitive

- Contains ethnic heritage, predisposition to diseases and conditions (even mental), many phenotypical traits
- Raises the risk of genetic discrimination – “genism”

It gets worse…

Leaking one's genome ≈ leaking relatives' genome

- ~99.9% of genomes of closely related humans identical
- Basis of Gymrek's attack
- The case of Henrietta Lacks
  - See Humbert et al. (ACM CCS, 2013)

Sensitivity of human genomes is (almost) perpetual

- Even if encrypted, can't guarantee security of the encryption algorithm past 30-50 years

More details:

- Ayday et al., Chills and Thrills of WGS, IEEE Computer

The Greater Good vs Privacy?

Advances in genomics often promoted as dependent on volunteers and data sharing

- Sharing is actually a requirement for most grants

Sharing is an important asset for research

- Chatterjee et al. (Nature, 2013) project that several million samples may be needed for robust GWAS

But privacy and discrimination fears may drive potential participants away?

- McGuire et al. (Genetics in Medicine, 2011) finds correlation between opting out and privacy fears

Open Questions

What do we understand about users' perceptions and attitudes with respect to Whole Genome Sequencing

Do privacy perceptions/concerns experienced by individuals correspond to what the scientific community would expect?

How to identify effective mechanisms to communicate risks and benefits? How to reconcile the greater good/ privacy tension?

(Little understanding from prior work in context of WGS)

Methodology 1/3

Recruited 16 study volunteers in SF Bay Area

- Sex: female (8), male (8)
- Age: 18-24 (2), 25-34 (7), 35-44 (3), 45-54 (1), 55-64 (1), 65-(2)
- Degree: College (4), Master (8), PhD (4)
- Income: <$50K (3), $50K-$75K (3), >$75K (10)
- Westin: Unconcerned (4), Pragmatist (7), Fundamentalist (5)

Participants skewed toward high-income/high-edu

Representative population for early WGS adopter, as per related work, e.g., Facio et al. (Nature, 2011), 2012 NPR study, …

Methodology 2/3

Participants guided through a set of slides depicting a few hypothetical scenarios

- Asked to comment on and rank these scenarios

Four experiments

- Exp A: Assessing perception of today's genetic tests
- Exp B: Comparing attitudes toward different WGS program
- Exp C: Assessing perception of privacy/ethical issues with WGS
- Exp D: Comparing the response to medical/genomic/personal information loss

Exp A – Trust

Genetic Tests: More to less inclined	Avg	Std
(A.6) Determine Cancer Treatment	5.81	0.39
(A.5) Determine Drug Dosage	4.63	0.70
(A.2) Genetic Compatibility	4.06	1.25
(A.1) Disease Predisp. (Doctor)	2.63	0.99
(A.4) Disease Predisp. (Company)	2.13	0.70
(A.3) Ancestry Testing	1.75	1.09

(A.6), (A.5), (A.2) statistically significantly higher than (A.1)

Mann-Whitney U Test ($U = 210{:}5$, $n1 = n2 = 16$, $P < 0.01$, two-tailed)

(A.1) and (A.4) close

(A.4) was ranked among the bottom because of mistrust in company

Exp B – Control

WGS Programs: More to less inclined	Avg	Std
(B.3) Data-only (DVD)	2.68	0.58
(B.1) Healthcare Provider	2.00	0.71
(B.2) Direct-to-Consumer (DTC) Company	1.31	0.46

(B.3) the “favorite” (12/16 ranking at the very top)

(B.2) the least “favorite” (11/16 ranking at the very bottom)

Diff b/w (B.1) and (B.2) stat. significant ($U = 194; P < 0.05$, two-tailed)

12/16 participants mention they wanted to “feel in control”

Mistrust against health provider: “use against me”, company “even worse”

When prospecting a $1,000 discount for (B.1), even more suspicious

Exp C – Discrimination

Incidents: More to less discomfort	Avg	Std
(C.1) Labor Discrimination	3.31	0.58
(C.2) Health Insurance Discrimination	3.00	0.94
(C.3) Sequenced Genome Leaked	2.56	0.93
(C.4) Sibling Donating Genome to Science	1.13	0.33

(C.4) least discomforting (14/16 at the very bottom), (C.1) most discomforting (15/16 participants ranking in top two)

- Some participants not surprised by (C.2)
- Some participants find (C.1) extremely unjust because of environmental factors

Exp D – Harm

Information loss: More to less frightened	Avg	Std
(D.1) Identity Theft	3.50	0.63
(D.3) Emails and Pictures Leaked	2.63	1.61
(D.4) Sequenced Genome Leaked	2.00	0.63
(D.2) Medical Records Leaked	1.88	0.48

(D.1) and (D.4) statistically significantly different

Correlation b/w lower income and (D.3), higher income and (D.1)

χ^2 (1;N = 32) = 8.60 $p < 0.01$ (both cases)

Correlation b/w fundamentalists and (D.1)

χ^2 (1;N = 32) = 4.36 $p < 0.05$

Summary of (Preliminary) Results

1) Users' perception depends on related perceived medical benefit

2) Participants prefer that doctors administer/explain genetic tests, mistrust of DTC companies

3) Raising issue of control – prefer to retain & own genomics data to minimize fear of potential discrimination

4) Labor/healthcare discrimination top concerns, also due to mistrust/unawareness of legislation

5) Genetic information disclosure not well understood

Related Work

Response to learning results of genetic tests

E.g. predisposition to cancer

Control

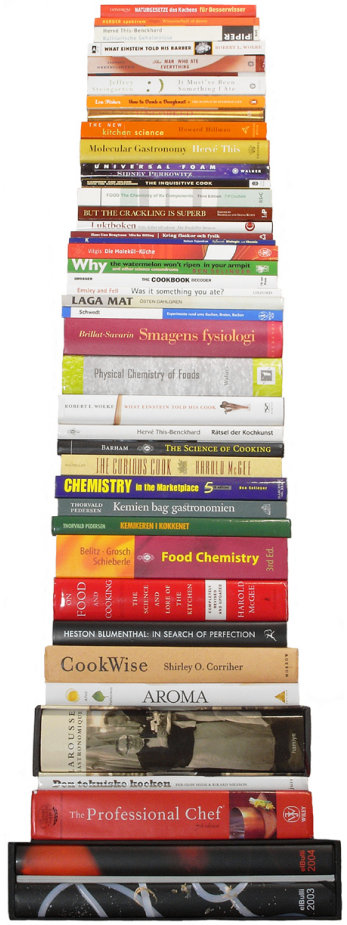
Participants want the opt-out option

Genetic discrimination

Well-known fear, not well contextualized

Informed consent issues

Very important area of research, very hard to get the consent right



Discussion

How to use preliminary study for a larger, more focused study?

- What to focus on?
- How to select/recruit participants?
- How to interview users on an emerging technology?

How to drive user-centered design of personal (computational) genomics?

How to raise awareness/communicate concerns and regulation from/to users?

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