



Global Alliance
for Genomics & Health
Collaborate. Innovate. Accelerate.

The DNA Dilemma

Providing individuals with their raw, uninterpreted genome

Neha Kumar, Paul Petit, Shweta Sen

Presenting on behalf of: The Personal Genome Project

Intended Audience: Global Alliance for Genomics & Health (aka GA4GH)

November 15, 2018



Genomes are now easier than ever to sequence



1970s

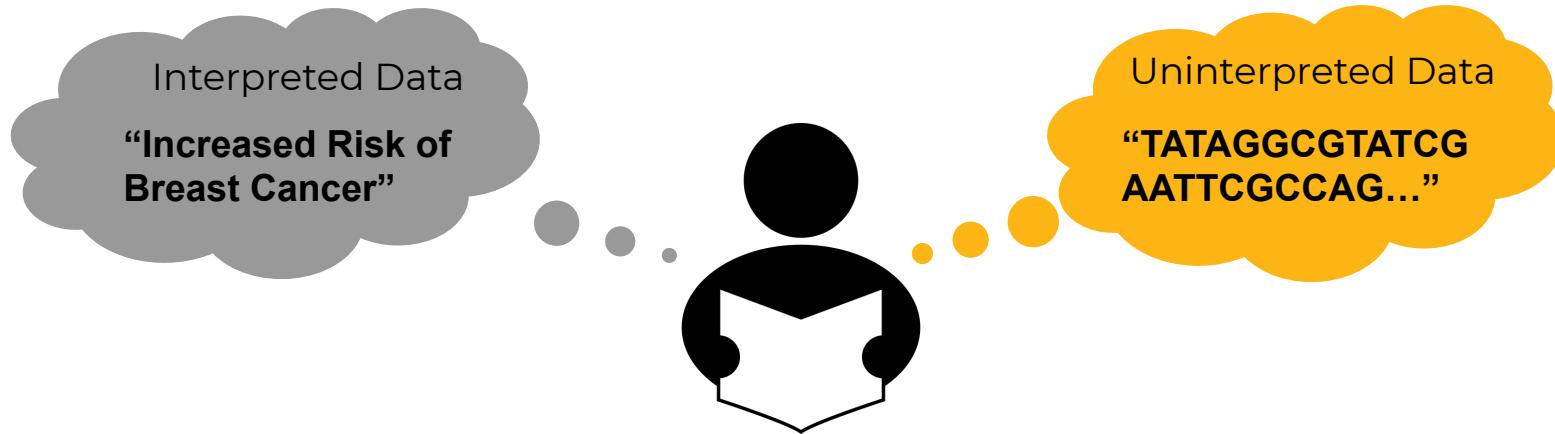
Sequencing
Technology
developed



Today

Cost to sequence
plummets from \$100M
(2000) to under \$1K

Should individuals have the right to access and distribute their full uninterpreted genomic data *in its entirety* as they see fit?



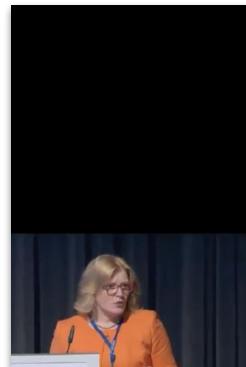
Actively Discussed in....



APPLaUD: Access for patients and participants to individual level uninterpreted genomic data (Human Genomics, 2018)



GA4GH 6th Plenary Meeting | GA4GH Connect -- Basel, Switzerland (Oct 2018)



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Regulatory and Ethics Work Stream

Lead: Professor Bartha Maria Knoppers

Lead: Professor Madeleine Murtagh

Manager: Adrian Thorogood



Centre of Genomics and Policy
Centre de génomique et politiques



McGill

Access to genomes enables research among a wider range of citizens and professional scientists

Primary reasons for requesting raw genome*

86% of patients



Learn about themselves / family history

80% of patients



Share data with other research groups

*Among patients who already have access to their genomes and opted to share their data [Haeusermann]

The debate has a cascading effect on multiple stakeholders

Primary Stakeholder | Policy-framing organization



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Drives Policy

Impacts

Data Collectors | Research Studies / DTC Services



Experiences the greatest fiscal impact

Impacts

Users | Research Participants / Patients



Drives demand

A series of policies contextualize the debate

"Everyone has the right.. to share in scientific advancements and its benefits"

Universal Declaration of Human Rights



HIPAA grants US patients right to genomic test results



A global standard to data access rights

1948

2014

2018

Looking ahead

Genomic sequencing becomes increasingly common



Personal Genome Project

GDPR sets EU standard to data access rights





Inconsistency among current studies begs the need for a uniform protocol

Increasing Restrictions



Personal
Genome
Project

Grants full access
to personal
genomic data



Grants access to DNA
snippets



Grants view only
access (no sharing)



Parties in favor of full access to uninterpreted genomic data



Pro Side

Access-Optimistic Individuals

“My DNA, my data! I have a right to it, and I’ll use it to make my life better.”

Scientists & Researchers

“Scientific possibilities are endless if this data is released for people to share with scientists.”

Corporations & Insurers

“The more data, the merrier!
Genome data can help drive customer value and reduce cost.”

Parties against full access to uninterpreted genomic data

Con Side

Access-Pessimistic Individuals

"This data is extremely sensitive!
I likely won't use it, and I don't want it in the wrong hands."

Data Collectors

"Releasing this data will cause a storm of misuse and drain precious resources as third parties misinterpret data."

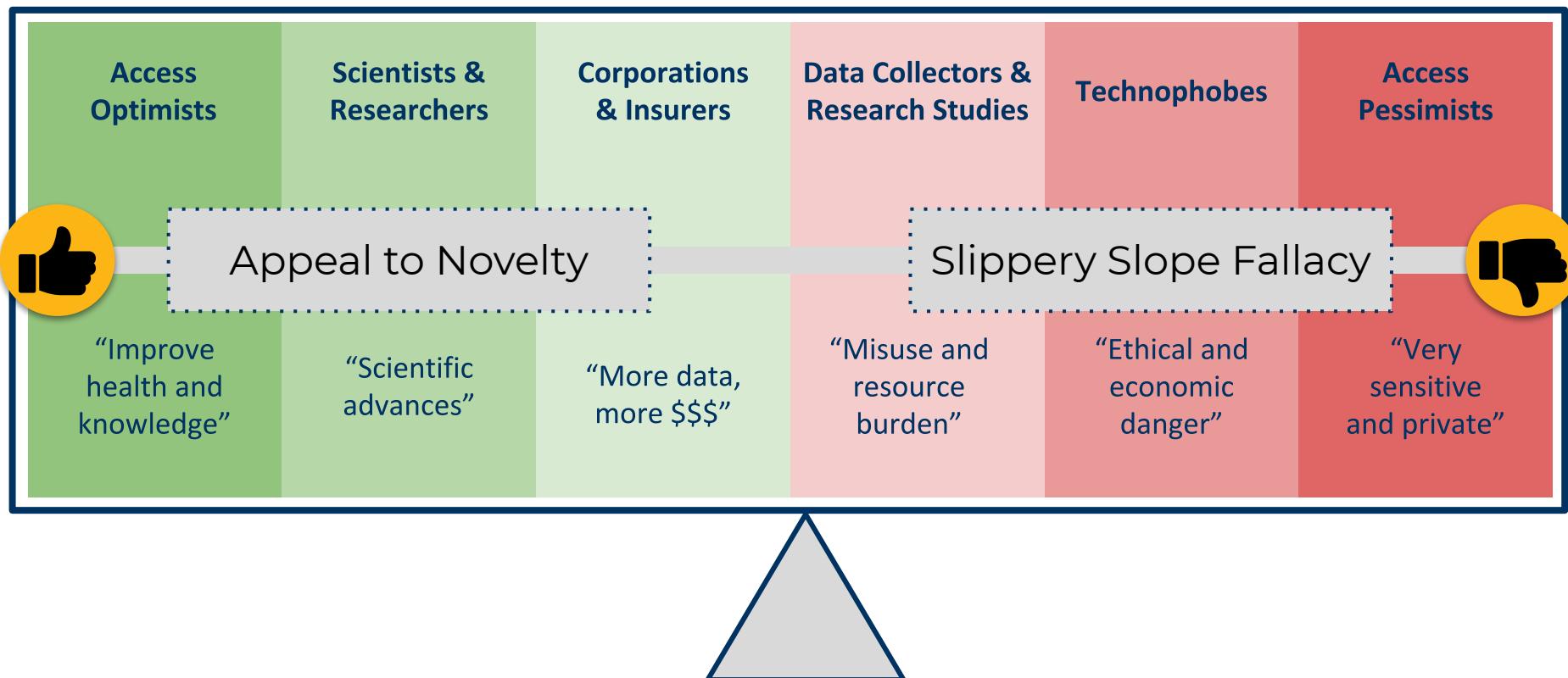
Technophobes

"Danger! Opening access to this data may have big bioethical and macroeconomic implications."





Pro-con Continuum and Fallacies of Argumentation





Looking Ahead

Projected Outcome: Pro-Side Wins

Individuals will have ownership of their entire genomic data and can distribute it in-full.

Key Reasons



Regulatory Shifts

HIPAA and GDPR aim to give control to individuals over their personal data.



Push towards Genomic Data Bank

Aggregation of genomic information can improve fidelity of interpretation services.



Potential for Medical Advancements

Expanding repository of genetic data can help pinpoint disease risk factors.

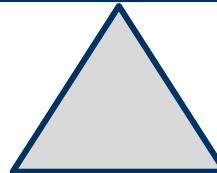


Calls for Data Transparency

Reducing corporate stronghold on data removes “behind the scenes” suspicions on data misuse.



Con-Side: Central Fears



How **secure** is my information?

How will I **control** my data and know where it's being used?

What if my information is **hacked** and gets used against me?

What if my data gets into the **wrong hands**?

Creating compromise: DS can give individuals security and control over their genomic data.

Privacy-Preserving Machine Learning



Healthcare informatics companies can anonymize data and use differential privacy techniques to maintain data sensitivity.

Used by: [Google](#), [Apple](#), [Microsoft](#), [Uber](#)

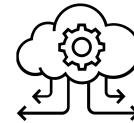
Blockchain Technology



Individuals can store genomic data on blockchain and release parts of it to receive gene interpretation services.

Used by: [Nebula](#), [GeneCoin](#), [LunaDNA](#)

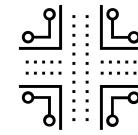
Cloud & Database Management



Data regarded as Public Health Information (PHI) can employ encryption, data access controls, and firewall implementation.

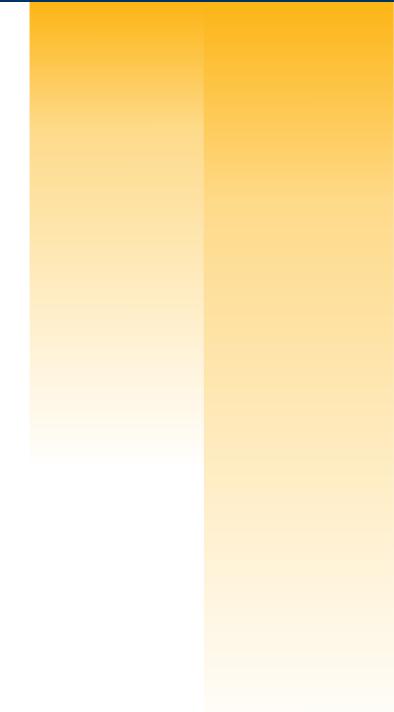
Used by: [Canadian Genomics](#), [Bionimbus](#)

Infrastructure Adaptations



Creation of DS regulatory body can help set standards for authentication, in-person account creation, data formats, and data transfer protocols.

Further reads: [NGS](#), [GCC](#), [NIH File Formats](#)



Questions?





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Appendix

Lower-cost sequencing has given rise to research studies and consumer services

Research Studies



Personal
Genome
Project

Genomics

england



Direct-To-Consumer Services



23andMe





How data science can enable progress

Arguments For & Against



Data Science Opportunities + Potential Use Cases

Data Mining & Machine Learning

Healthcare researchers can generate disease prediction models by identifying key risk factors and training inferences against a genomic data bank.

Database & Cloud Management

High-volume database provides hospitals secure and reliable access to patients' genomic history, paving the way for personalized medicine.

Business Intelligence

Using BI tools to detect data negligence and fraud. BI analysts can investigate corporate practices and consumer behavior to assess conformance.



How data science can create compromise

Arguments For & Against



Data Science Opportunities + Potential Use Cases

Companies Pay You for Your Data

Organizations profiting from your data could pay you a share of the earnings. The theory, posed by Eric Posner, states data creation should be a form of reciprocated labor costs.

Blockchain Technology

A user could store genomic information on blockchain and release parts of it to receive interpretation services, thus limiting impact of privacy erosion and enabling transparent data transfers.

Privacy-Preserving Machine Learning

Healthcare informatics companies can anonymize data and use differential privacy techniques to maintain data sensitivity.



Pro-con Continuum and Fallacies of Argumentation

Access Optimists	Corporations & Insurers	Scientists & Researchers	Data Collectors & Research Studies	Technophobes	Access Pessimists
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“Improve health and knowledge”

“More data, more \$\$\$”

“Scientific advances”

“Misuse and resource burden”

“Ethical and economic danger”

“Very sensitive and private”

Appeal to Novelty



Slippery Slope Fallacy