

Ethics, Genomics and Society

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Senior Social Scientist

SOCIETY+
ETHICS
RESEARCH

CONNECTING
SCIENCE

Genetics/genomics and ethics

Researching ethics

Genomics and data

Conclusions

Genetics and ethics

- Late 19th/early 20th century, concerns with genetic and racial health led to 'positive' and 'negative' eugenics
- Rewards for reproducing, forced sterilisation, involuntary research, genocide
- Members of population seen as of differing 'worth' to society



Genetics and ethics

- Reproductive decision making
- Genetic manipulation
- Privacy and confidentiality
- Risk of stigmatisation or unfair discrimination
- Incidental/additional findings
- Testing of those without capacity
- IP and gene patents



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President George W. Bush signs H.R. 493, the Genetic Information Nondiscrimination Act of 2008, Wednesday, May 21, 2008, in the Oval Office. White House photo by Eric Draper.



THE LANCET

COMMENT | VOLUME 394, ISSUE 10215, P2133-2135, DECEMBER 14, 2019

Should doctors have a legal duty to warn relatives of their genetic risks?

Anna Middleton • Richard Milne • Lauren Roberts • Jonathan Roberts • Christine Patch

Published: November 26, 2019 • DOI: [https://doi.org/10.1016/S0140-6736\(19\)32941-1](https://doi.org/10.1016/S0140-6736(19)32941-1) • Check for updates



United Kingdom
Genomics England 2012-
 100,000 Genomes: rare disease, cancer
 £350M (USD\$485M)
Scottish Genomes £6M (USD\$8M)
Welsh Genomics for Precision Medicine
 £6.8M (USD\$9M)
Northern Ireland Genomic Medicine Centre £3.3M (USD\$4.6M)

Switzerland
Swiss Personalized Health Network 2017-2020
 Infrastructure
 CHF68M (USD69M)

France
Genomic Medicine Plan 2016-2025
 Rare disease, cancer, diabetes
 €670M (USD\$799M)

Estonia
Estonian Genome Project 2000 –
 Infrastructure and population-based cohort
 2017: €5M for 100,000 individuals

Netherlands
RADICON-NL 2016-2025
 Rare disease
 Health Research Infrastructure

Finland
National Genome Strategy 2015-2020
 Infrastructure
 €50M (\$USD 59M)

Denmark
Genome Denmark 2012-
 DK 86M (USD\$13.5M)
FarGen 2011- 2017
 DK 10M (USD\$1.6M)
 Infrastructure, population-based cohort, pathogen project

United States of America
National Human Genome Research Institute 2007-
 Infrastructure and clinical cohorts
 USD\$427M
All of Us 2016-2025
 Population cohort
 USD\$500M (first two years)

Turkey
Turkish Genome Project 2017-2023
 Infrastructure, clinical and population-based cohorts

Brazil 2015-
Brazil Initiative on Precision Medicine
 Infrastructure, disease and population cohorts

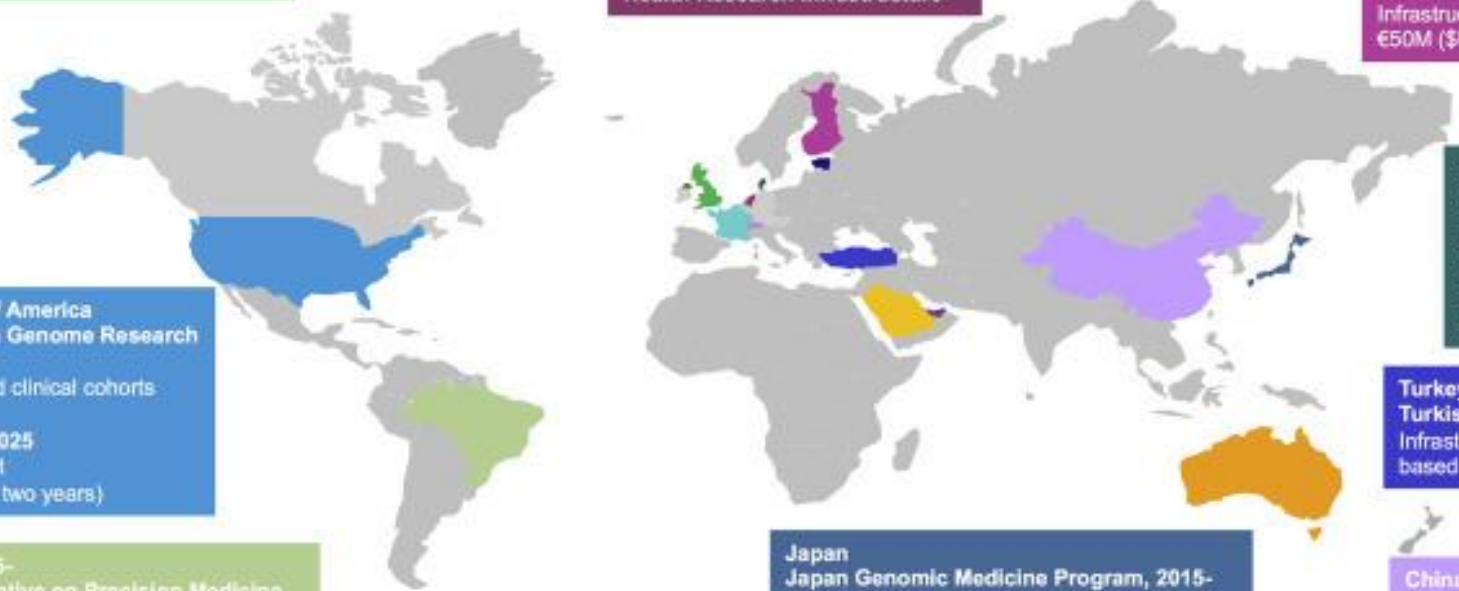
Japan
Japan Genomic Medicine Program, 2015-
 Infrastructure, clinical and population-based cohorts, drug discovery
 JPY10.2B (USD\$90.05M)

China Precision Medicine Initiative
 100,000,000 genomes
 CNY60 billion (USD\$9.2 billion)

Qatar
Qatar Genome 2015-
 Infrastructure, population cohort

Saudi Arabia
Saudi Human Genome Program, 2013-
 Infrastructure, clinical cohorts and population-based cohorts
 SAR300M (USD\$80M)

Australia
Australian Genomics 2016-2021
 Infrastructure, rare disease and cancer
 AUD\$125M (USD\$95M)
Genomics Health Futures Mission 2018-2028
 AUD\$500M (USD\$372M)



Genomics and ethics

- Reproductive decision making
- Genetic manipulation
- Privacy and confidentiality
- Risk of stigmatisation or unfair discrimination
- Incidental/additional findings
- Testing of those without capacity
- IP and gene patents
- Blurring of clinical care and research
- Obtaining valid consent
- Breadth and feedback of findings
- **Responsible data stewardship**
 - Future use of samples and data
- **Maintaining public trust and confidence**
- **Justice and equity of access**

Bioethics

- Moral reasoning applied to the life sciences
 - Consequentialism
 - Deontology
 - Virtue ethics
 - Principlism
 - Feminist bioethics

Social science

- Sociology, anthropology, geography applied to the sciences
- What are the potential social, political, economic impacts of genomics?
- How do and should societal concerns and priorities shape science?
- Which topics come to be seen as ethical concerns and why?
- What do 'the public' think about ethical questions and what are the implications of this for science?

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Genomics and data

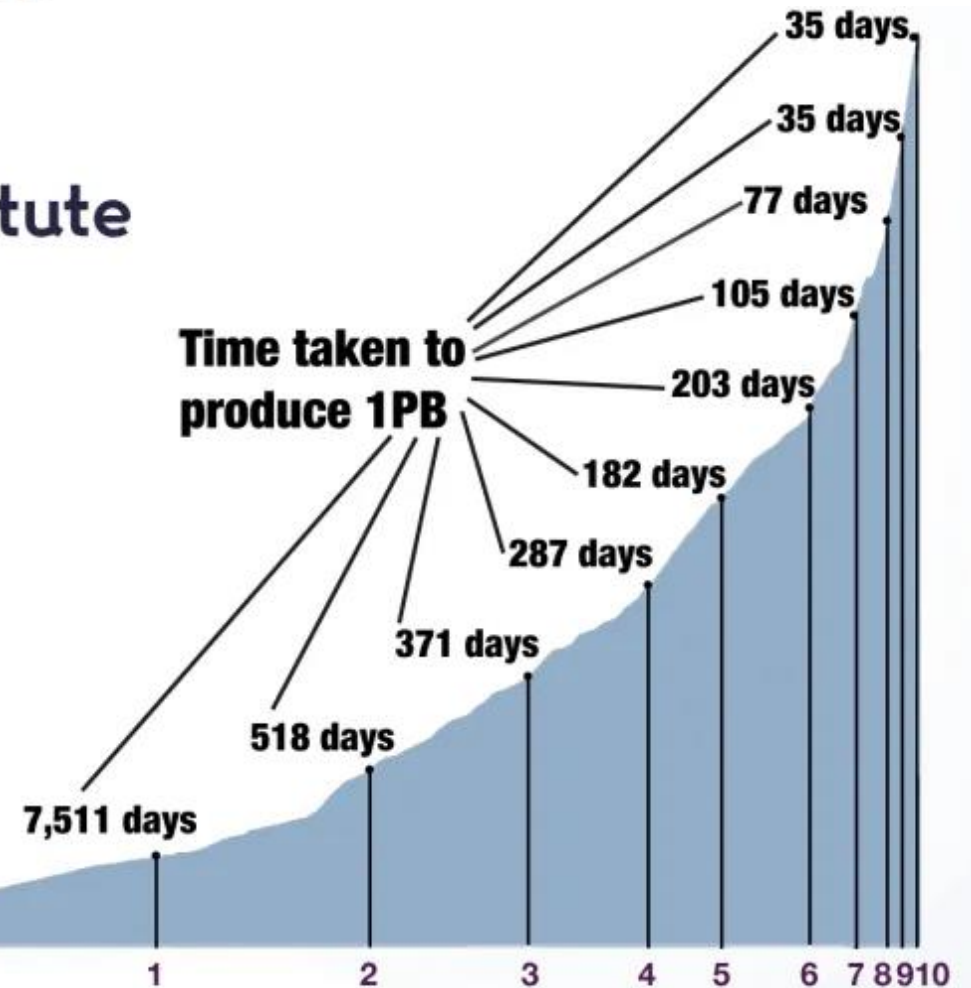
Sanger's super-sized sequencing scales new heights

May 1, 2019 6 min read

We're celebrating: we've just read the same amount of DNA in one year as we achieved in the previous 25 years combined. This dizzying speed offers unprecedented possibilities.

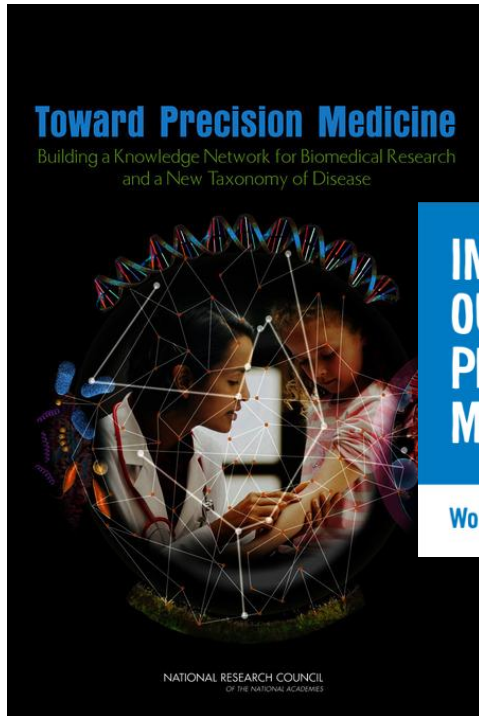
By: Ali Cranage, science writer

Total amount of DNA read by the Wellcome Sanger Institute



Cumulative total of Petabases of DNA sequenced by the Wellcome Sanger Institute

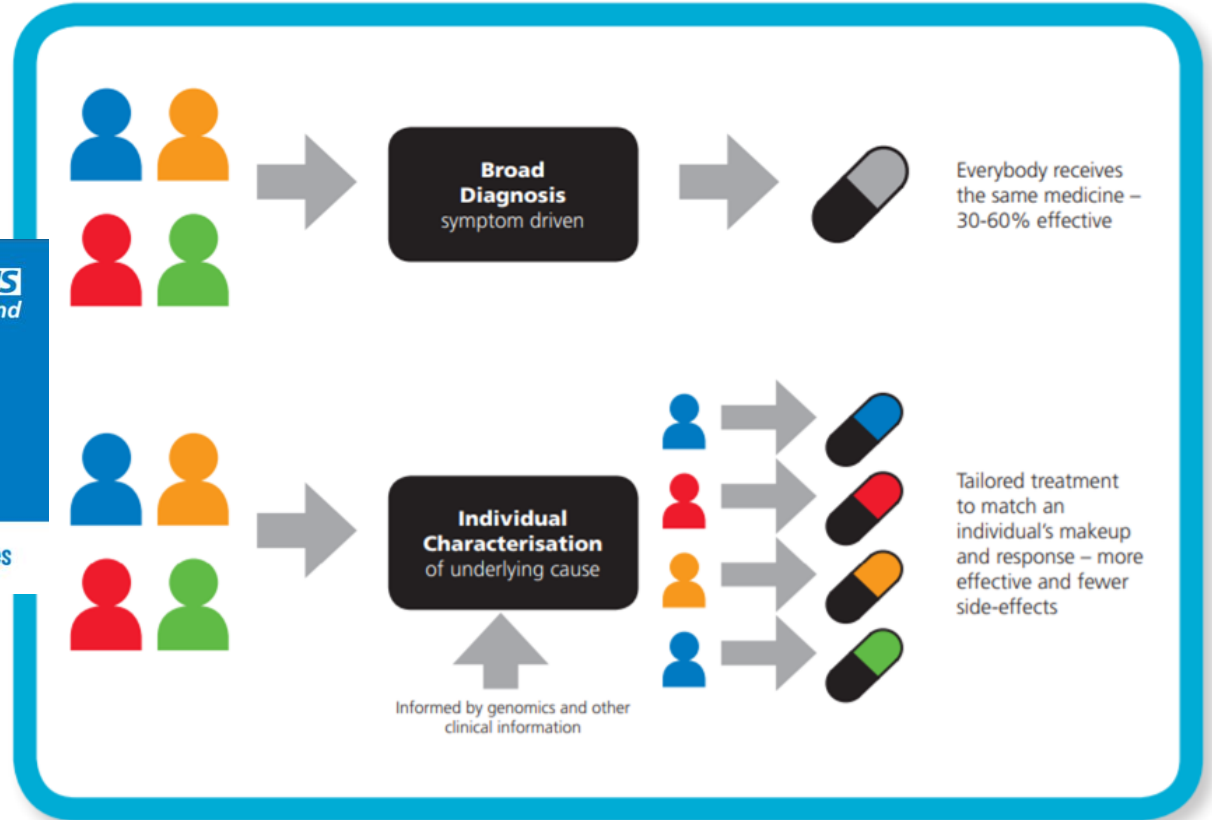
Precision and data-driven medicine



IMPROVING OUTCOMES THROUGH PERSONALISED MEDICINE

NHS England

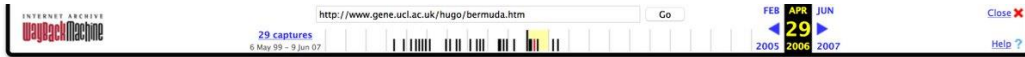
Working at the cutting edge of science to improve patients' lives



Sharing data



NATIONAL CANCER INSTITUTE GENOMIC DATA COMMONS



THE WELLCOME TRUST

Summary of principles agreed at the International Strategy Meeting on Human Genome Sequencing

Bermuda, 25th-28th February 1996
Sponsored by the Wellcome Trust

The following principles were endorsed by all participants. These included officers from, and scientists supported by, the Wellcome Trust, the UK Medical Research Council, the NIH NCHGR (National Center for Human Genome Research), the DOE (U.S. Department of Energy), the German Human Genome Programme, the European Commission, HUGO (Human Genome Organisation) and the Human Genome Project of Japan. It was noted that some centres may find it difficult to implement these principles because of legal constraints and it was, therefore, important that funding agencies were urged to foster these policies.

Primary Genomic Sequence Should be

It was agreed that all human genomic seq... encourage research and development and



Challenges of collecting, sharing and linking genomic data

- Consent
- Privacy and harms
- Justice and fairness
- Trust
- The problems of non-use

Consent

- Limits of consent in face of open-ended data and sample storage and use
- Limits on autonomy in terms of controlling use of samples
- Unclear risks to privacy due to data sharing
- Acceptability of 'broad consent'
- Risks to individuals and to research

Privacy and harms

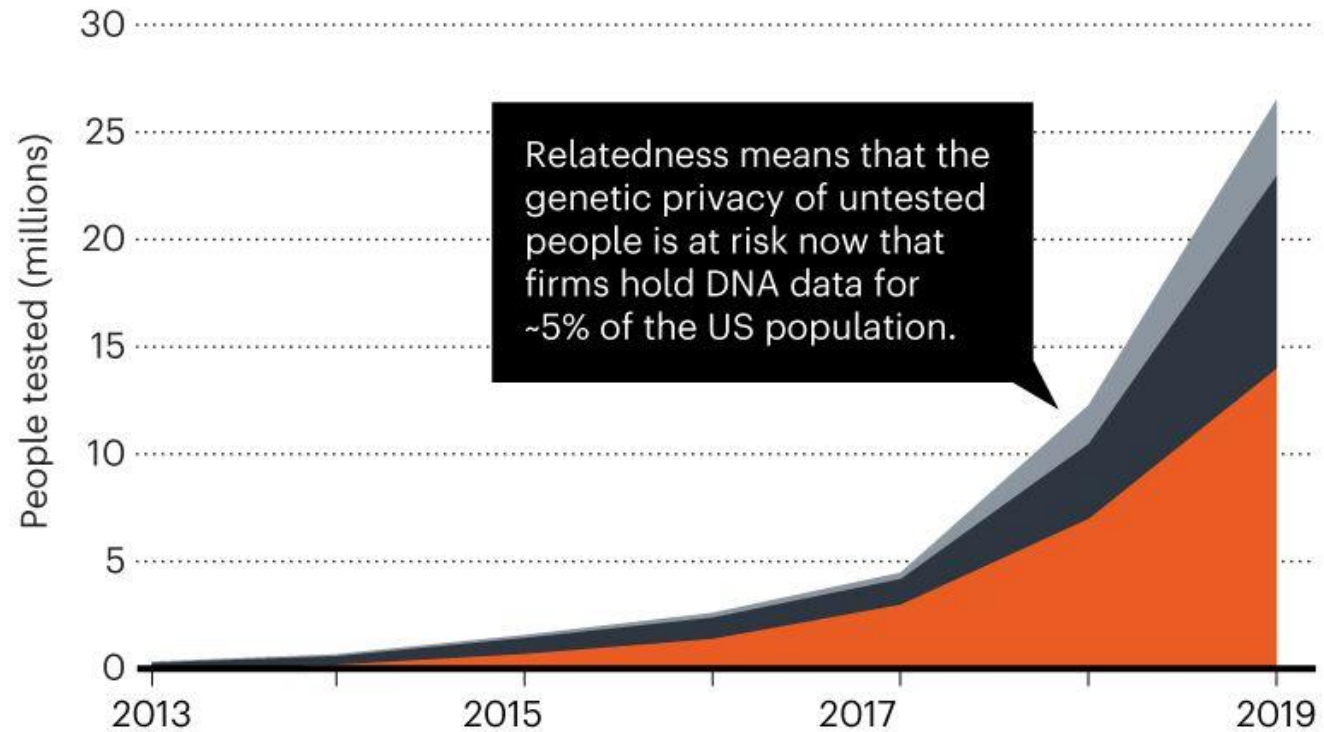
Unintended and misuse of data leading to:

Breaches of privacy
Stigma
Discrimination

DNA TESTING FOR ALL

An increasing number of people are having their DNA analysed by consumer-genomics companies.

■ Ancestry ■ 23andMe ■ Others

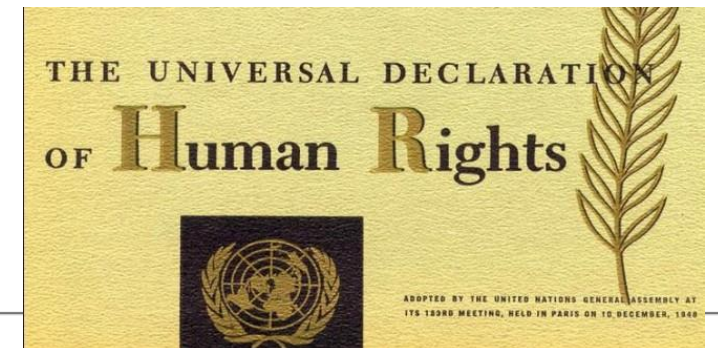
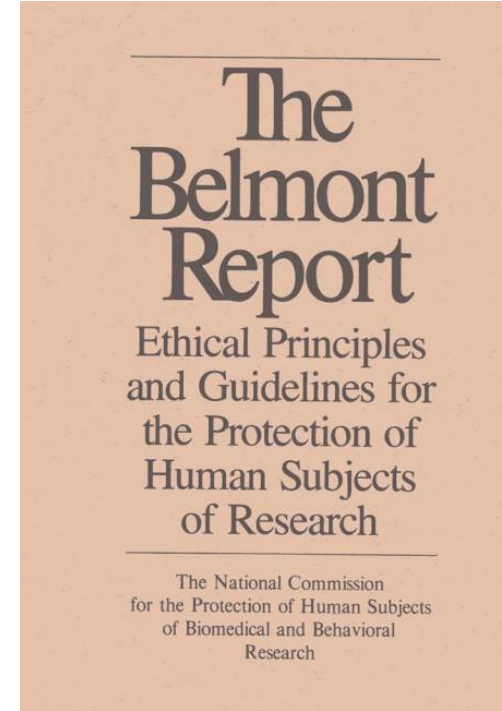


Relatedness means that the genetic privacy of untested people is at risk now that firms hold DNA data for ~5% of the US population.

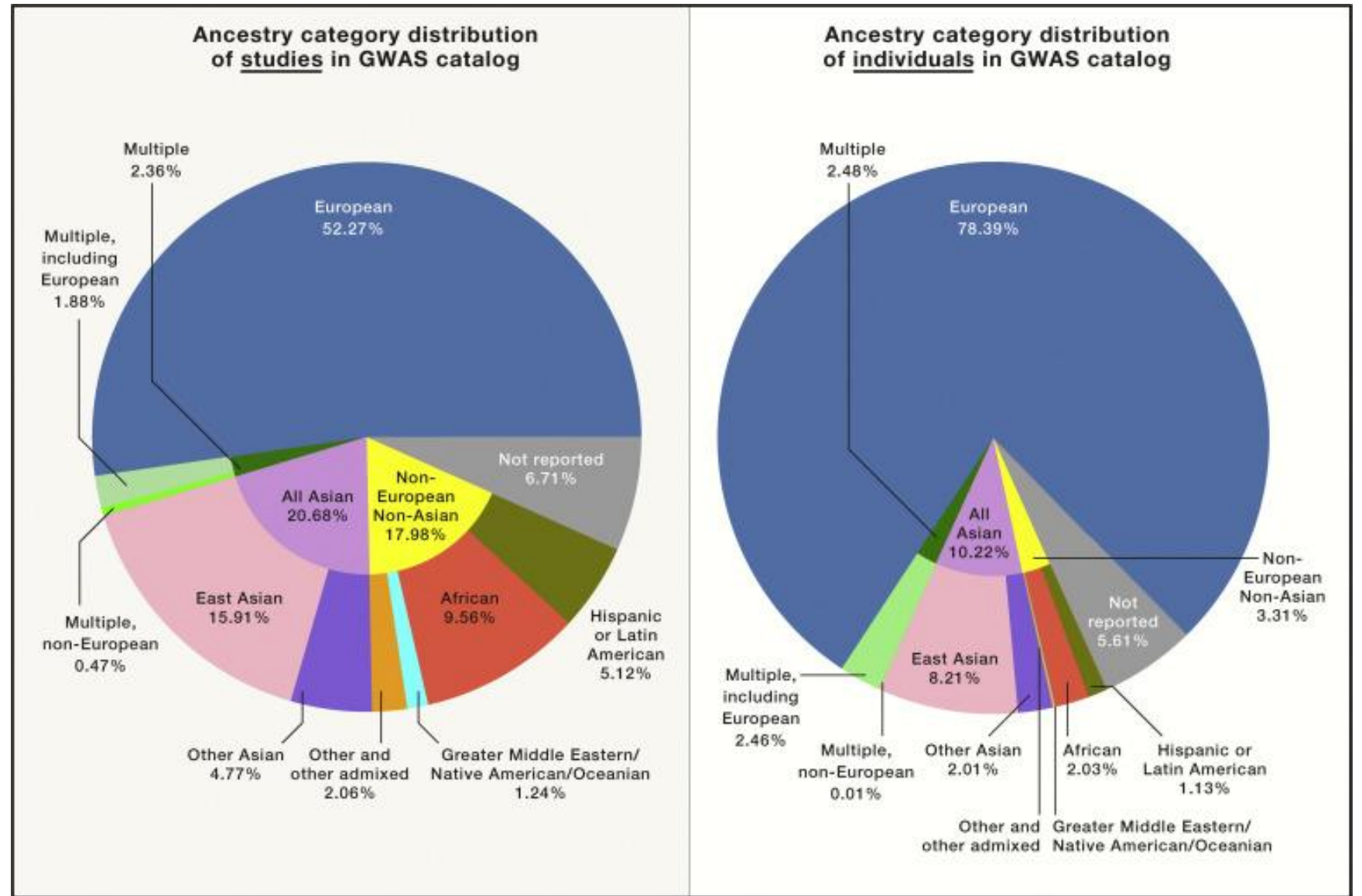
Justice

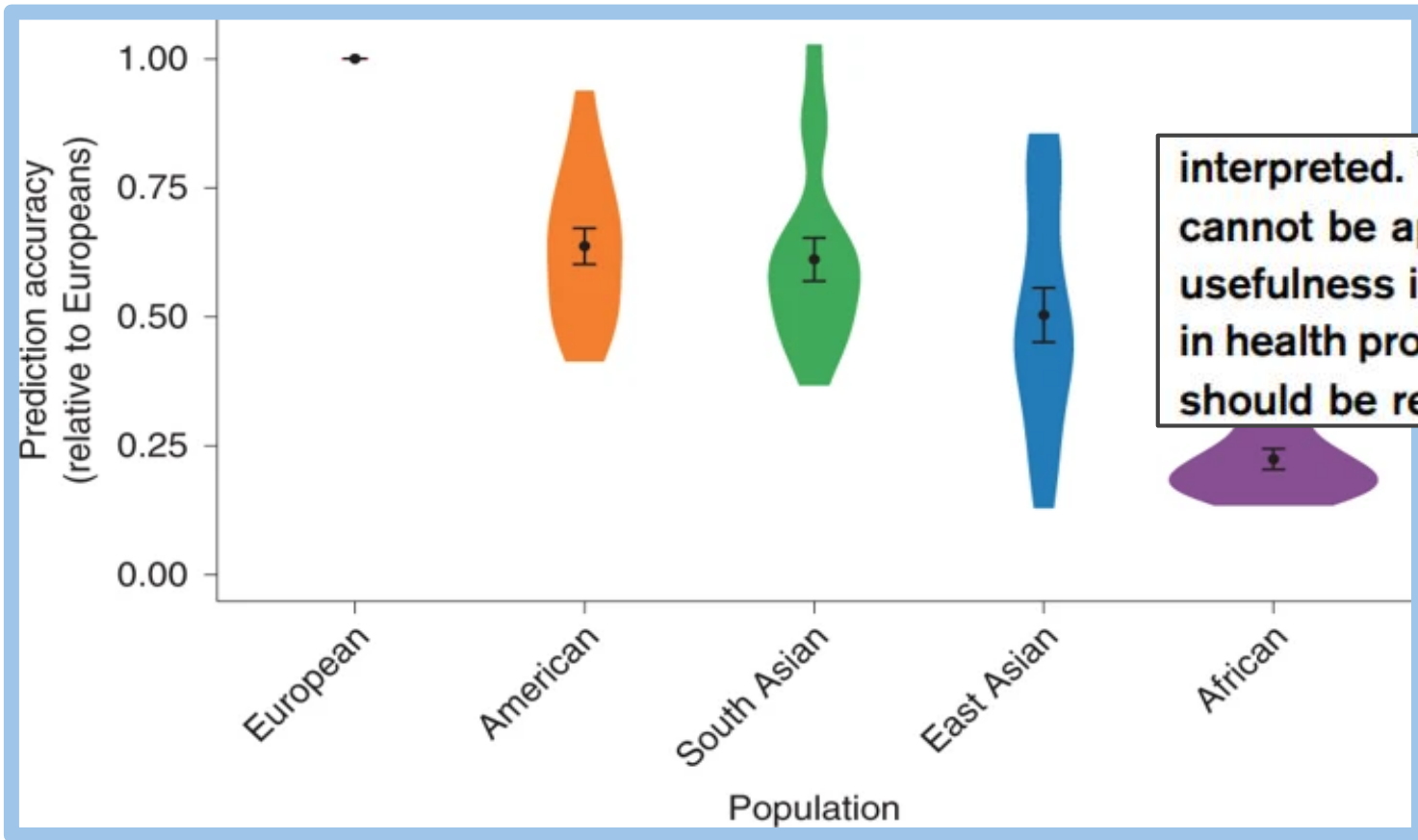
“Who ought to receive the benefits of research and bear its burdens?” (Belmont Report)

“Everyone has the right freely to participate in the cultural life of the community, to enjoy the arts **and to share in scientific advancement and its benefits.**” (Article 27 UNDHR)



Sirugo *et al.* (2019)
<https://doi.org/10.1016/j.cell.2019.02.048>





Martin, A. et al., *Nature Genetics* (2019);
<https://doi.org/10.1038/s41588-019-0379-x>.

interpreted. The PRS derived from European subjects cannot be applied to non-Europeans, limiting its potential usefulness in clinical settings and raising issues of inequity in health provision. Previous studies that have used the PRS should be re-examined in the light of these findings.

Curtis, D. *Psychiatric Genetics* 28, no. 5 (2018)
<https://doi.org/10.1097/YPG.0000000000000206>.

RESEARCH ARTICLE

ECONOMICS

Dissecting racial bias in an algorithm used to manage the health of populations

Ziad Obermeyer^{1,2*}, Brian Powers³, Christine Vogeli⁴, Sendhil Mullainathan^{5*†}

Obermeyer et al., *Science* (October 25, 2019)
<https://doi.org/10.1126/science.aax2342>.

Prediction accuracy relative to European-ancestry individuals across 17 quantitative traits and 5 continental populations in the UKBB.



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THE FUTURE OF HEALTH BEGINS WITH YOU

Trust and trustworthiness

Use of data relies on all stakeholders trusting in the organisations responsible for decision making

Where trust is absent, the social license for data use may be lost

Potentially problematic in contexts of private sector involvement



◆ WSJ NEWS EXCLUSIVE | TECH

Google's 'Project Nightingale' Gathers Personal Health Data on Millions of Americans

Search giant is amassing health records from Ascension facilities in 21 states; patients not yet informed



Patient data from GP surgeries sold to US companies

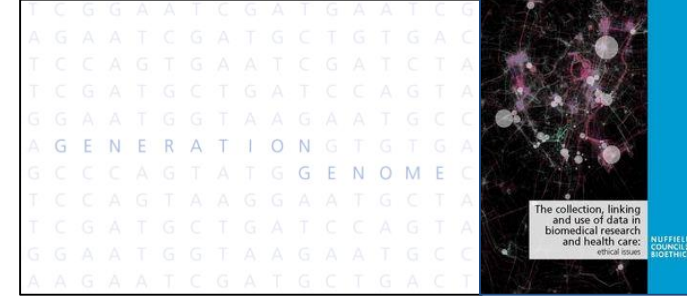
Dealings with international pharma raise new fears about American ambitions to access NHS



THE SUNDAY TIMES

Amazon ready to cash in on free access to NHS data

Achieving a balance



Pursuit of public interest

Protection of private interests

How to maximise societal benefit from scientific research

How to protect individuals from misuse of data about them

How to realise collective benefits by protecting individuals



How to help individuals benefit from collective action

“what is the set of morally reasonable expectations about the use of data and what conditions are required to give sufficient confidence that those expectations will be satisfied?”

- respect for persons
- respect for human rights
- participation of those with morally relevant interests
- accounting for decisions



Multiple stakeholders

“decision makers should not merely imagine how people ought to expect their data to be used, but **should take steps to discover how people do, in fact, expect their data to be used, and engage with those expectations.**”

(Nuffield Council on Bioethics, 2015)



“Facilitate deliberation about the wider societal implications of genomic and health-related data sharing among all stakeholders, especially citizens.” (Knoppers 2016)

Your DNA, Your Say

Global public views on sharing genomic data



Global public survey

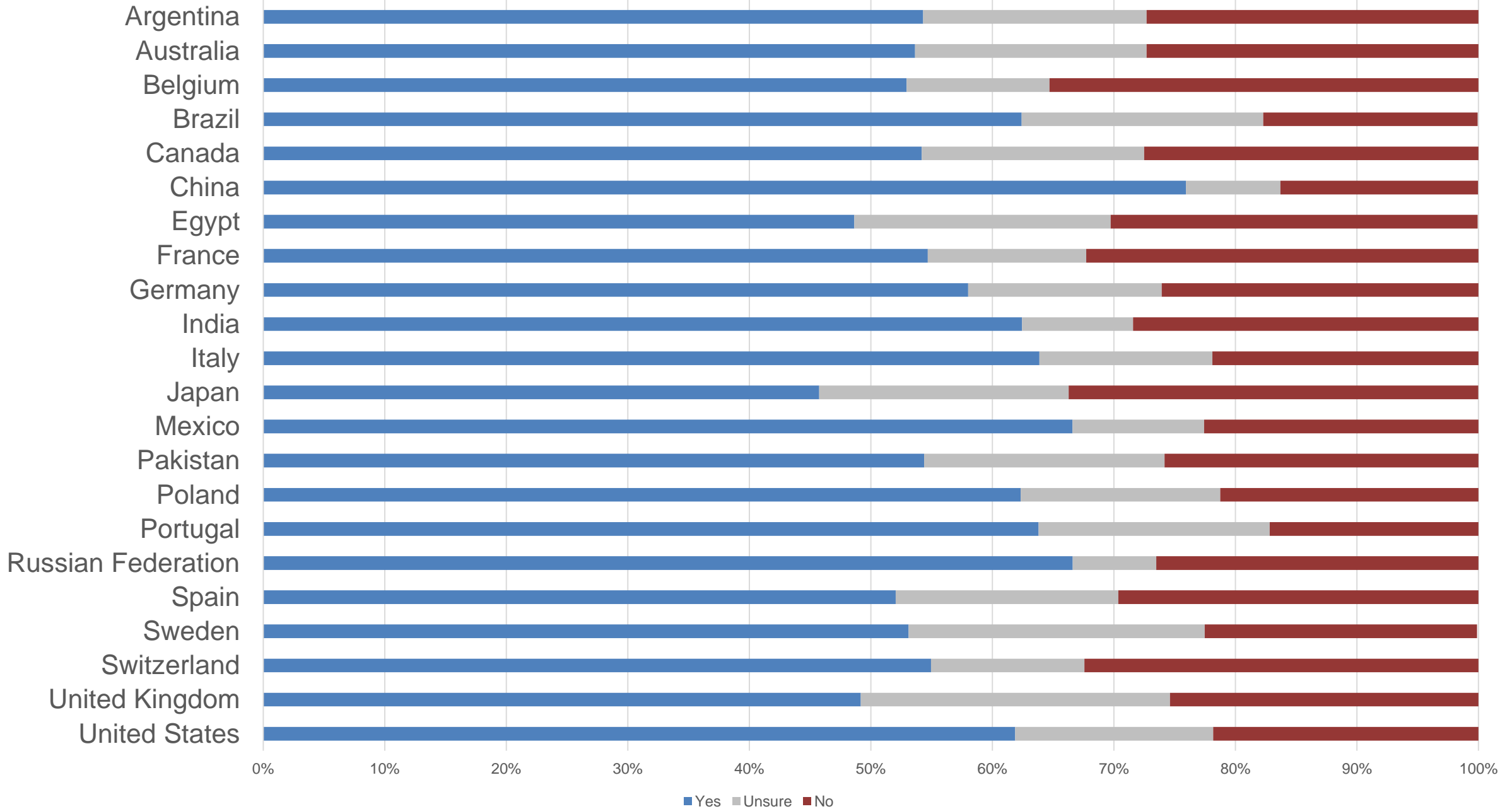
Sample size: 37,000 completed samples, 'representative' public recruited via Dynata (global market research company)

22 countries, 15 languages

Data collected 2017-2019



Willingness to donate DNA and health information



The importance of familiarity

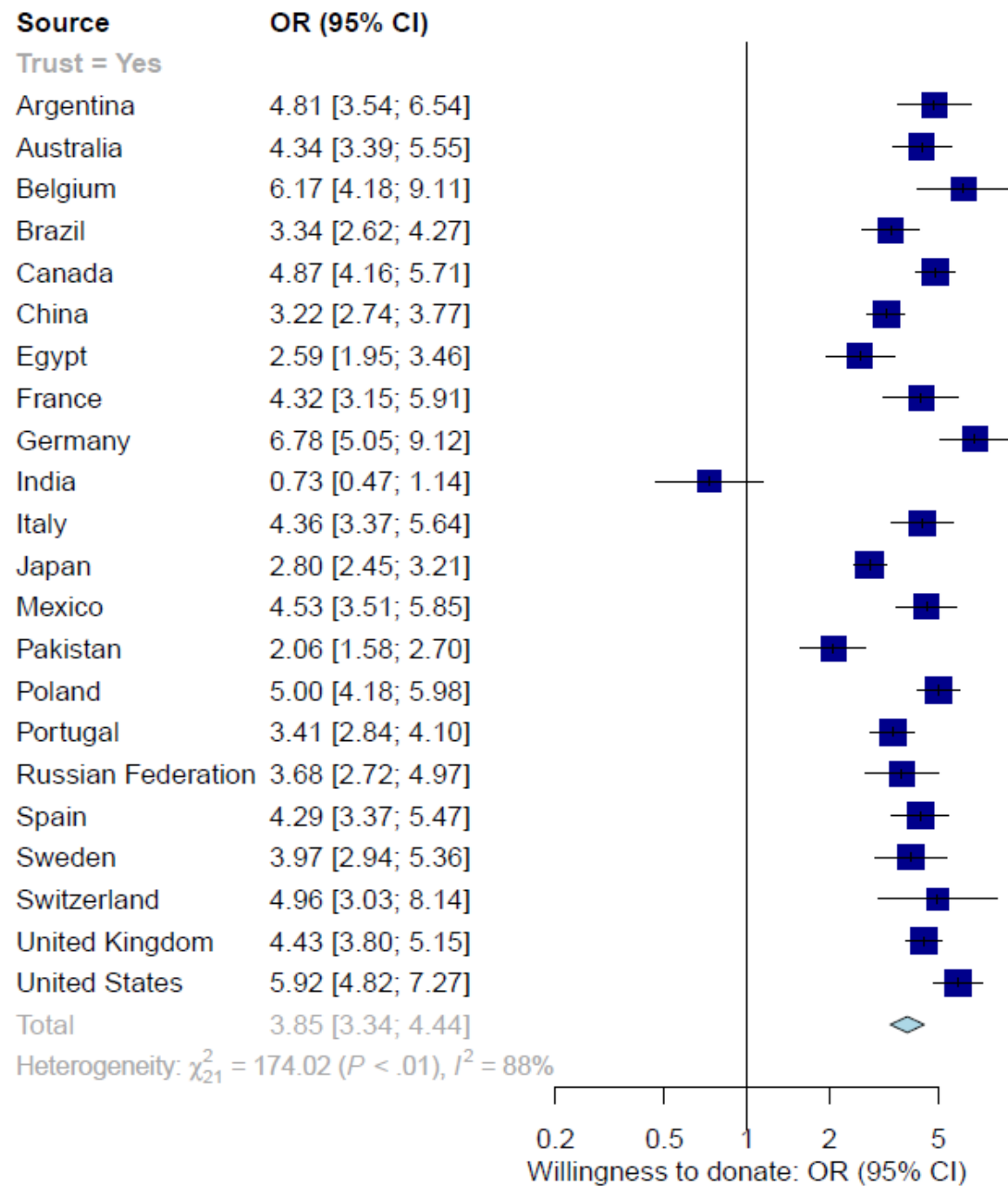
Familiarity with genomics is low (<50% in 20/22 countries)

Those people who are most familiar with DNA are more willing to donate (Overall OR 1.85, 95% CI 1.71-2.00)

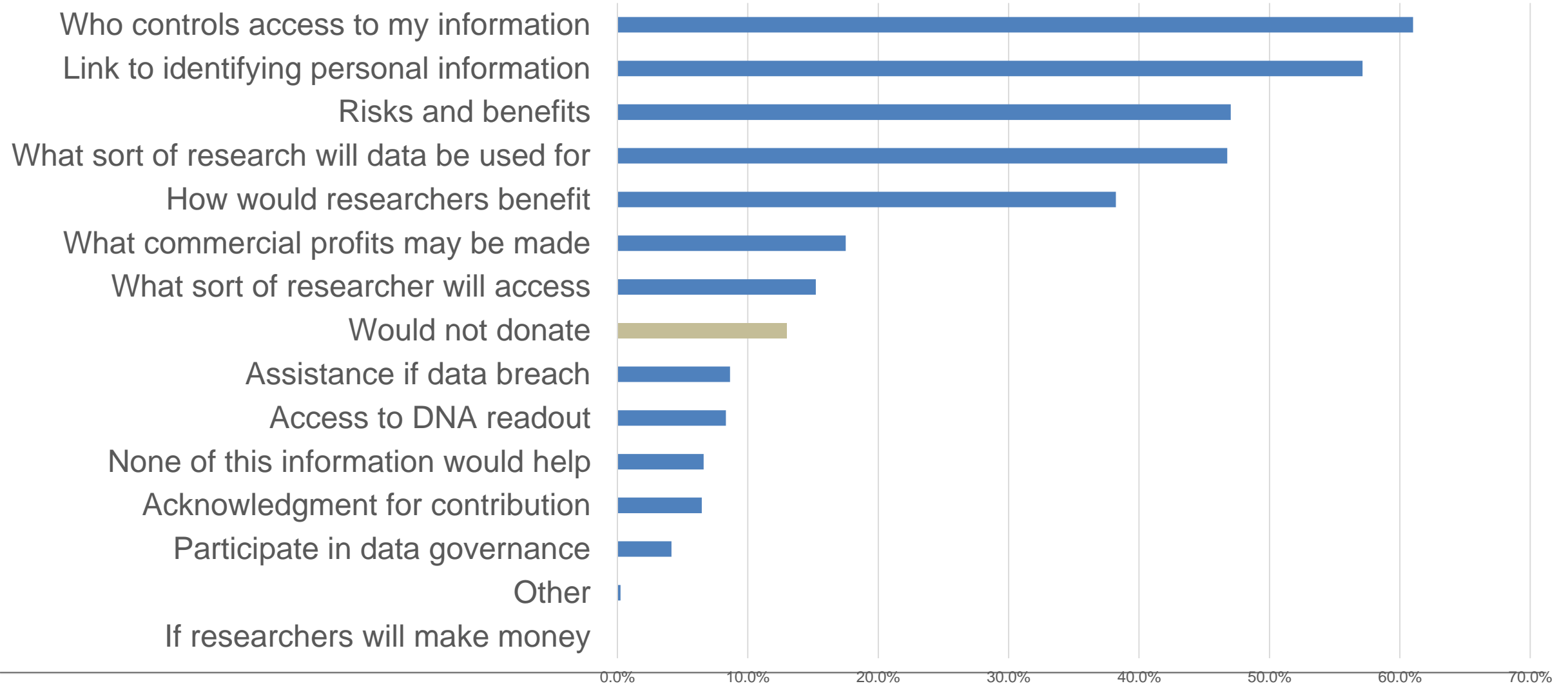
Those with personal familiarity are most willing to donate (Overall OR 2.7, 95% CI 2.37-3.09)

Total percentage trusting each organisation or individual with DNA and health information

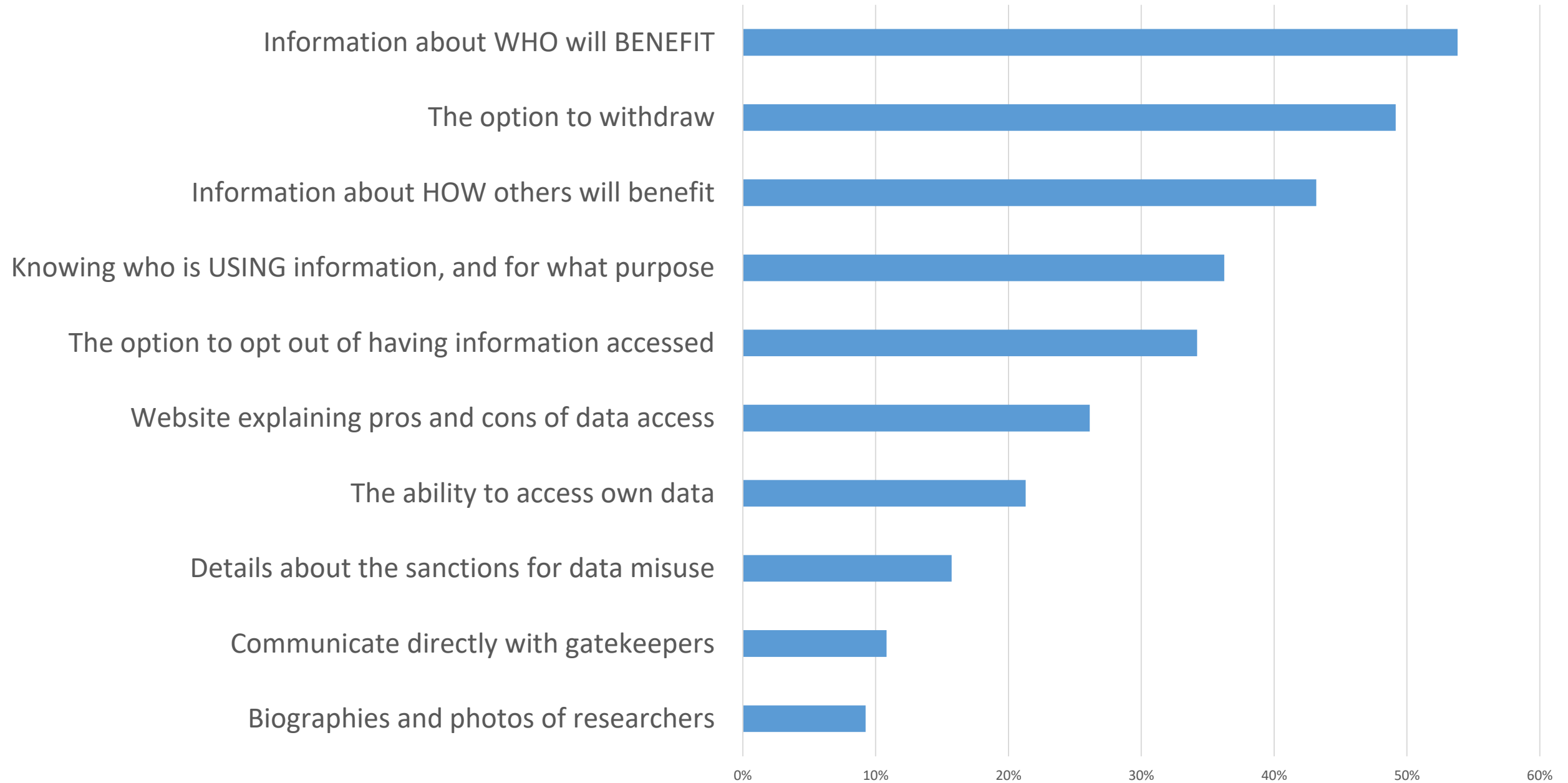




What information would help you decide whether or not to donate? (UK only)



What would help trust (UK only)





Data — from objects to assets

How did data get so big? Through political, social and economic interests, shows **Sabina Leonelli**, in the fourth essay of a series on how the past 150 years have shaped science.

“Building robust records of the judgements baked into data systems, supplemented by explicit reflections on whom they represent, include or exclude will enhance the accountability of future uses of data. It also helps to bring questions of value to the heart of research, rather than pretending that they are external to the scientific process” (Leonelli 2019)

Conclusions

- Genomic research raises distinctive socio-ethical questions
- These cover research findings, research practice and the products of genomic research
- Researchers should consider what makes their research 'ethical' and anticipate consequences of their research – both positive and negative, drawing on other expertise where necessary
- For data, questions of who controls and who benefits are important

Middleton et al (2019) Members of the public in the USA, UK, Canada and Australia expressing genetic exceptionalism say they are more willing to donate genomic data. *European Journal Human Genetics*

Milne et al (2019) Trust in genomic data sharing among members of the general public in the UK, USA, Canada and Australia. *Human Genetics*

Middleton A, et al (2019). Attitudes of publics who are unwilling to donate DNA data for research. *European Journal of Medical Genetics*. Volume 62, Issue 5, Pages 316–323.

Middleton A et al (2018) ‘Your DNA, Your Say’: global survey gathering attitudes toward genomics: design, delivery and methods. *Personalized Medicine*, Vol. 15, issue 4, pp.311-318.

Middleton, A (2018) Society and personal genome data. *Human Molecular Genetics*. Vol. 27, No. R1

Middleton A(2017) Your DNA, Your Say, *The New Bioethics*, 23:1, 74-80,