# From Genomes to GPS:
## Socially responsible research and the future of data-driven medicine

Richard Milne
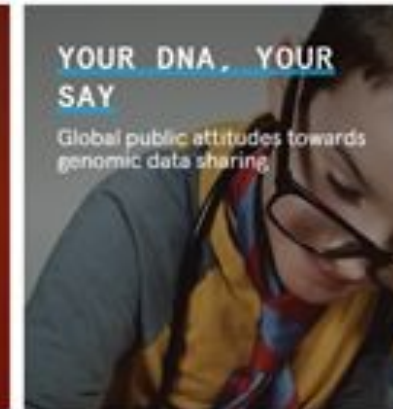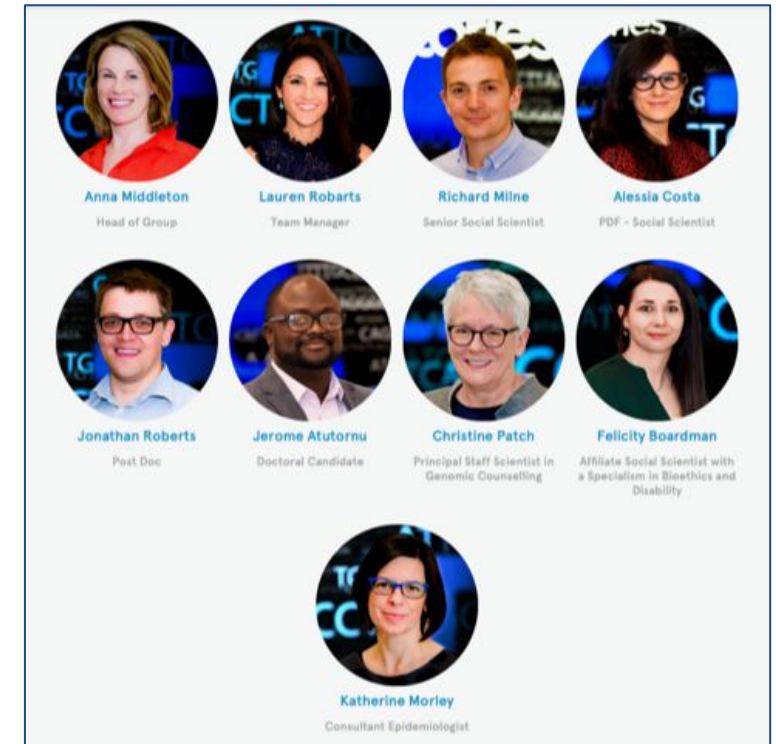Anna Middleton
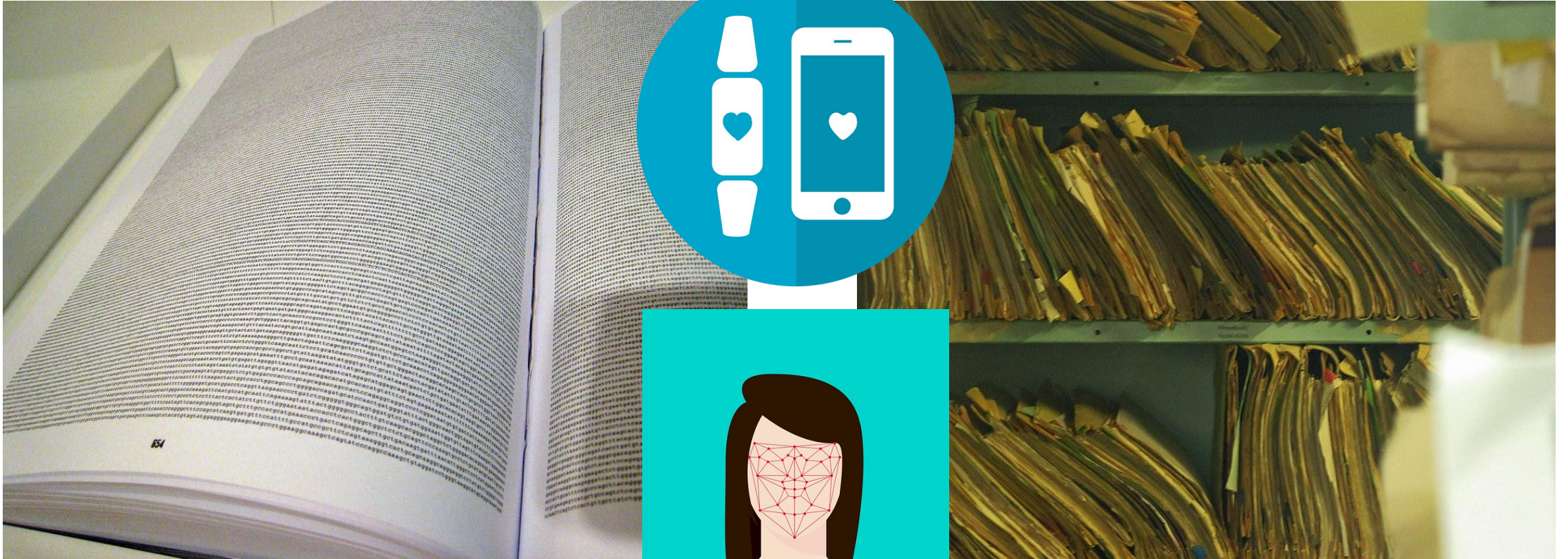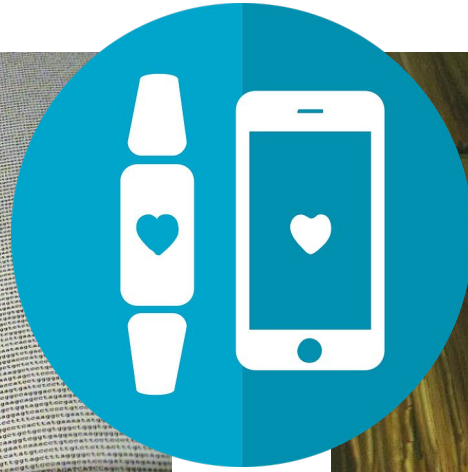
SOCIETY+
ETHICS
RESEARCH

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# Society and Ethics Research

Empirical social science research exploring the translation of genomics from bench to bedside and beyond
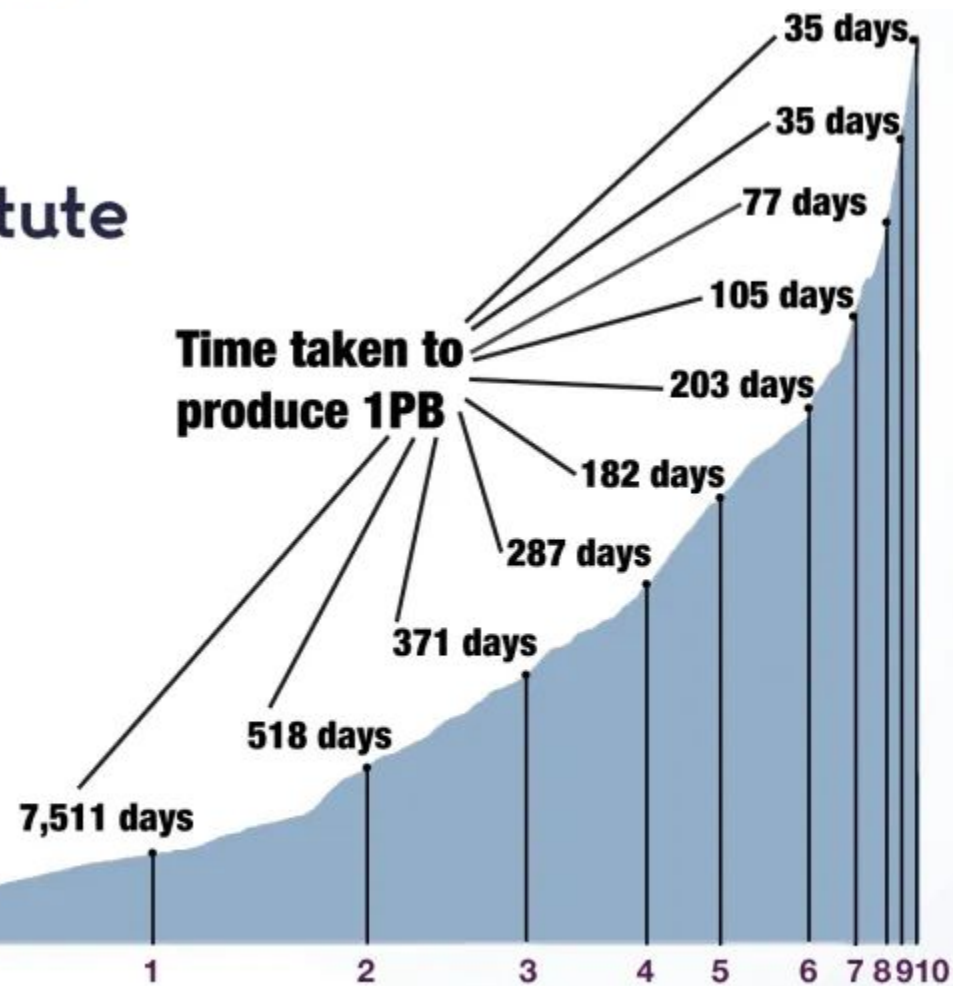
# Biomedical and health data

# 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 possib

By: Ali Cranage, science write



**Total amount of DNA read by the Welcome Sanger Institute**

Time taken to produce 1PB

- 35 days
- 35 days
- 77 days
- 105 days
- 203 days
- 182 days
- 287 days
- 371 days
- 518 days
- 7,511 days

wellcome **sanger** institute

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

# A DAY IN DATA

The exponential growth of data is undisputed, but the numbers behind this explosion – fuelled by internet of things and the use of connected devcies – are hard to comprehend, particularly when looked at in the context of one day

## 463EB
of data will be created every day by 2025

## 500m
tweets are sent every day

## 4PB
of data created by Facebook, including

**350m** photos

**100m** hours of video watch time

### DEMYSTIFIYING DATA UNITS

From the more familiar 'bit' or 'megabyte', larger units of measurement are more frequently being used to explain the masses of data

| Unit | | Value | Size |
|------|------|------|------|
| b | bit | 0 or 1 | 1/8 of a byte |
| B | byte | 8 bits | 1 byte |
| KB | kilobyte | 1,000 bytes | 1,000 bytes |
| MB | megabyte | $1,000^2$ bytes | 1,000,000 bytes |
| GB | gigabyte | $1,000^3$ bytes | 1,000,000,000 bytes |
| TB | terabyte | $1,000^4$ bytes | 1,000,000,000,000 bytes |
| PB | petabyte | $1,000^5$ bytes | 1,000,000,000,000,000 bytes |
| EB | exabyte | $1,000^6$ bytes | 1,000,000,000,000,000,000 bytes |
| ZB | zettabyte | $1,000^7$ bytes | 1,000,000,000,000,000,000,000 bytes |
| YB | yottabyte | $1,000^8$ bytes | 1,000,000,000,000,000,000,000,000 bytes |

*A lowercase "b" is used as an abbreviation for bits, while an uppercase "B" represents bytes.

## 95m
photos and videos are shared on Instagram

## 294bn
billion emails are sent

## 320bn
emails to be sent each day by 2021

## 306bn
emails to be sent each day by 2020

## 65bn
messages sent over WhatsApp and two billion minutes of voice and video calls made

## 3.9bn
people use emails
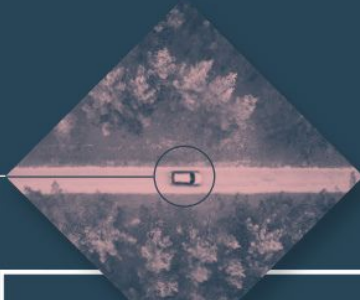
## 4TB
of data produced by a connected car

## 28PB
to be generated from wearable devices by 2020

Searches made a day — **5bn**

Searches made a day from Google — **3.5bn**

### ACCUMULATED DIGITAL UNIVERSE OF DATA

**4.4ZB** 2013

**44ZB** 2020

# The promise of data-driven medicine
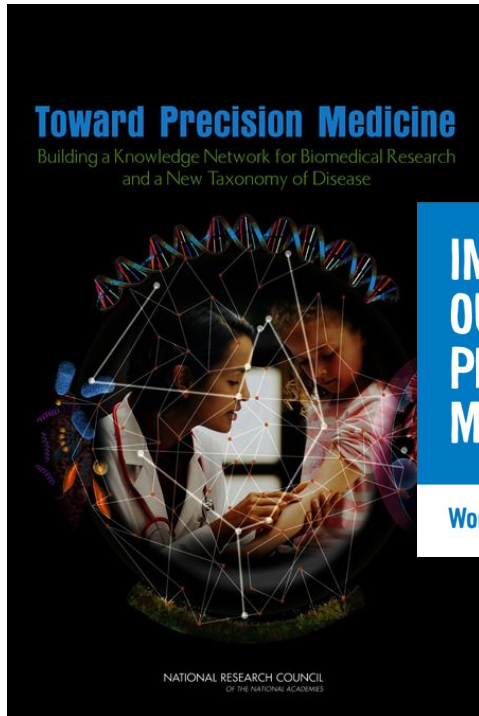
# Convergence

## Prediction of cardiovascular risk factors from retinal fundus photographs via deep learning
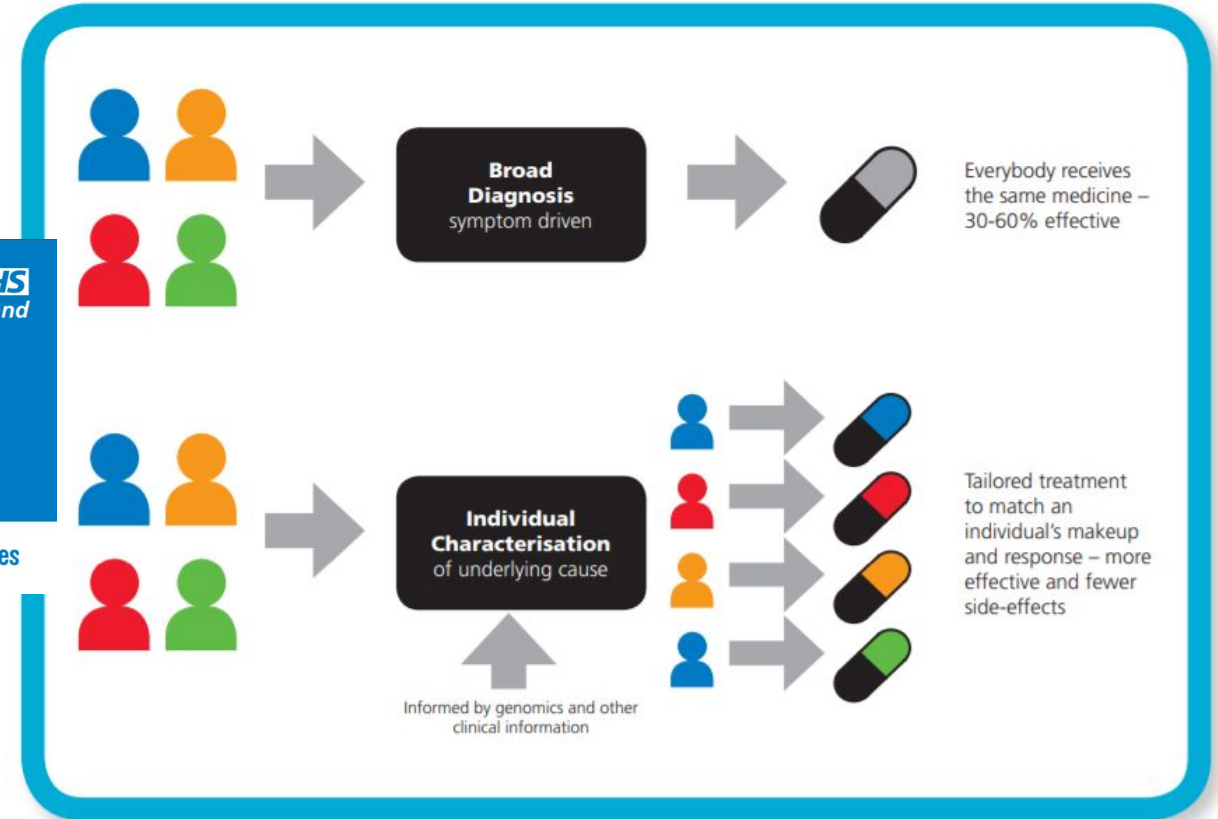
## Individualized Medicine from Prewomb to Tomb

Eric J. Topol[1],*

In 2010, Eric Schmidt of Google said "The power of individual targeting—the technology will be so good it will be very hard for people to watch or consume something that has not in some sense been tailored for them" (Jenkins, 2010). Although referring to the capability of digital technology, we have now reached a time of convergence of the digital and biologic domains. It has been well established that 0 and 1 are interchange-

# Sharing data



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

NATIONAL CANCER INSTITUTE
GENOMIC DATA COMMONS

Access the Data

#NCIGDC

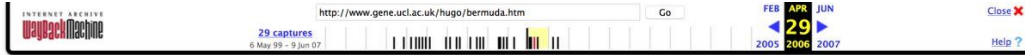## Summary of principles agreed at the International Strategy Meeting on Human Genome Sequencing

**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 in the Publ**

It was agreed that all human genomic sequence infor
encourage research and development and to maximise

National Institutes of Health
Office of Science Policy

DRAFT NIH Policy for Data Management and Sharing
November 2019

**DRAFT NIH Policy for Data Management and Sharing**

**I. Purpose**

The NIH Policy for Data Management and Sharing (herein referred to as the Policy) reinforces NIH's longstanding commitment to making the results and outputs of the research that it funds and conducts available to the public. Data sharing enables researchers to rigorously test the validity of research findings, strengthen analyses through combined datasets, reuse hard-to-generate data, and explore new frontiers of discovery. In addition, NIH emphasizes the

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# **The challenges**

Data misuse and the threat
to privacy

- Who is using data
- Trust and trustworthiness

Justice and equity

# Data misuse

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.

©nature

Moreau, Y. (2019) *Nature*

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1 square = 1 article

Legend:
- Gender
- Age
- Politics
- Location
- Occupation
- Ethnicity and Race
- Family and Relationships
- Education
- Income
- Language
- Health
- Religion
- Sexual Orientation
- Social Class
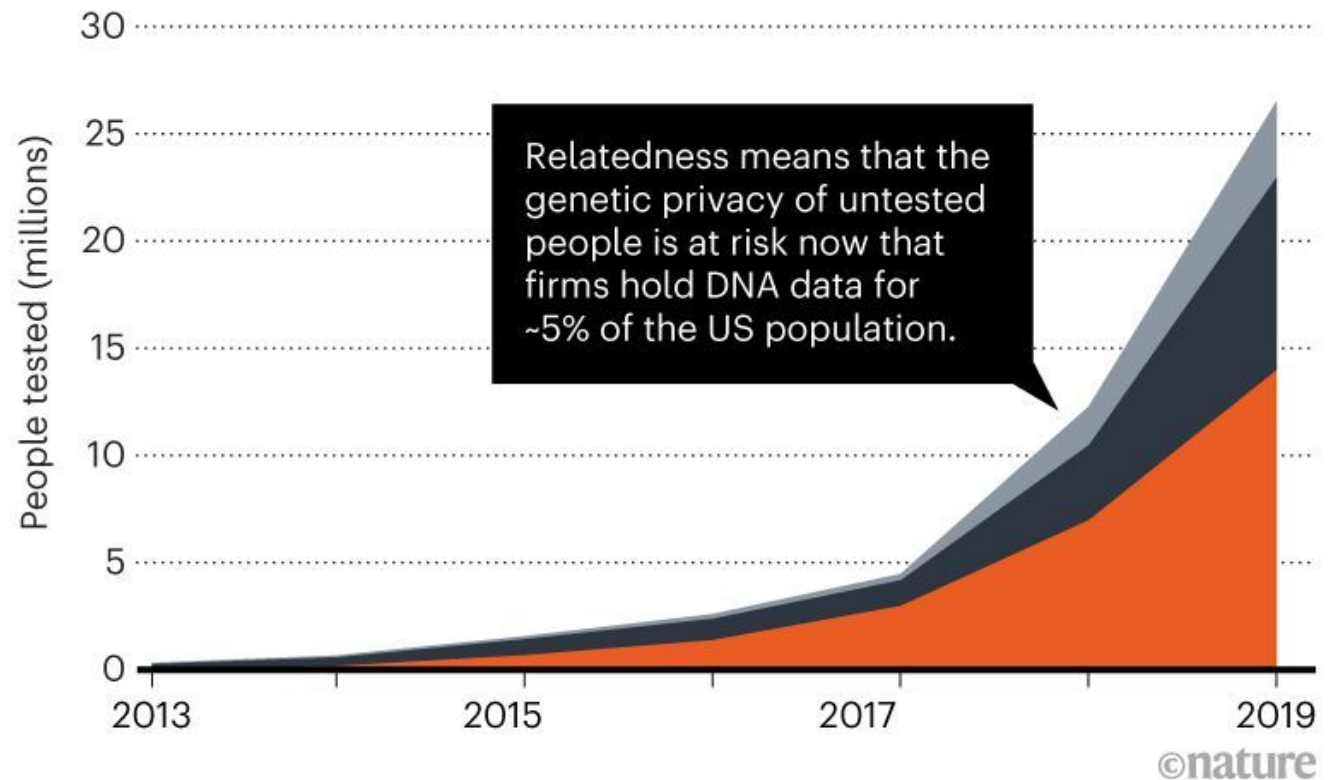
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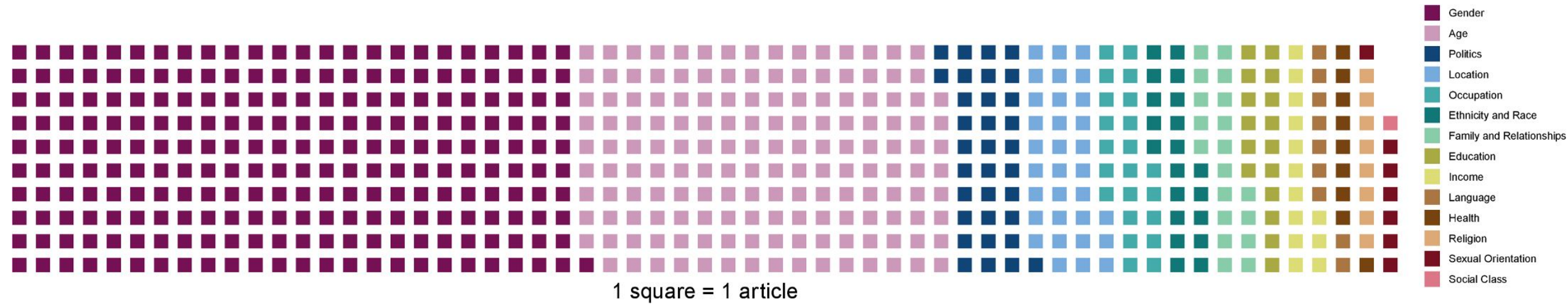
- Data from WebMD's symptom checker, and diagnoses received, including "drug overdose" shared with Facebook.
- "Heart disease" and "considering abortion" were shared from sites like the British Heart Foundation, Bupa and Healthline



User data is sent to dozens of third parties from health websites

Flow of user data from BabyCenter.com, accessed from UK, Nov 7 2019

Source: Crownpeak
Graphic by Alan Smith, Max Harlow
© FT
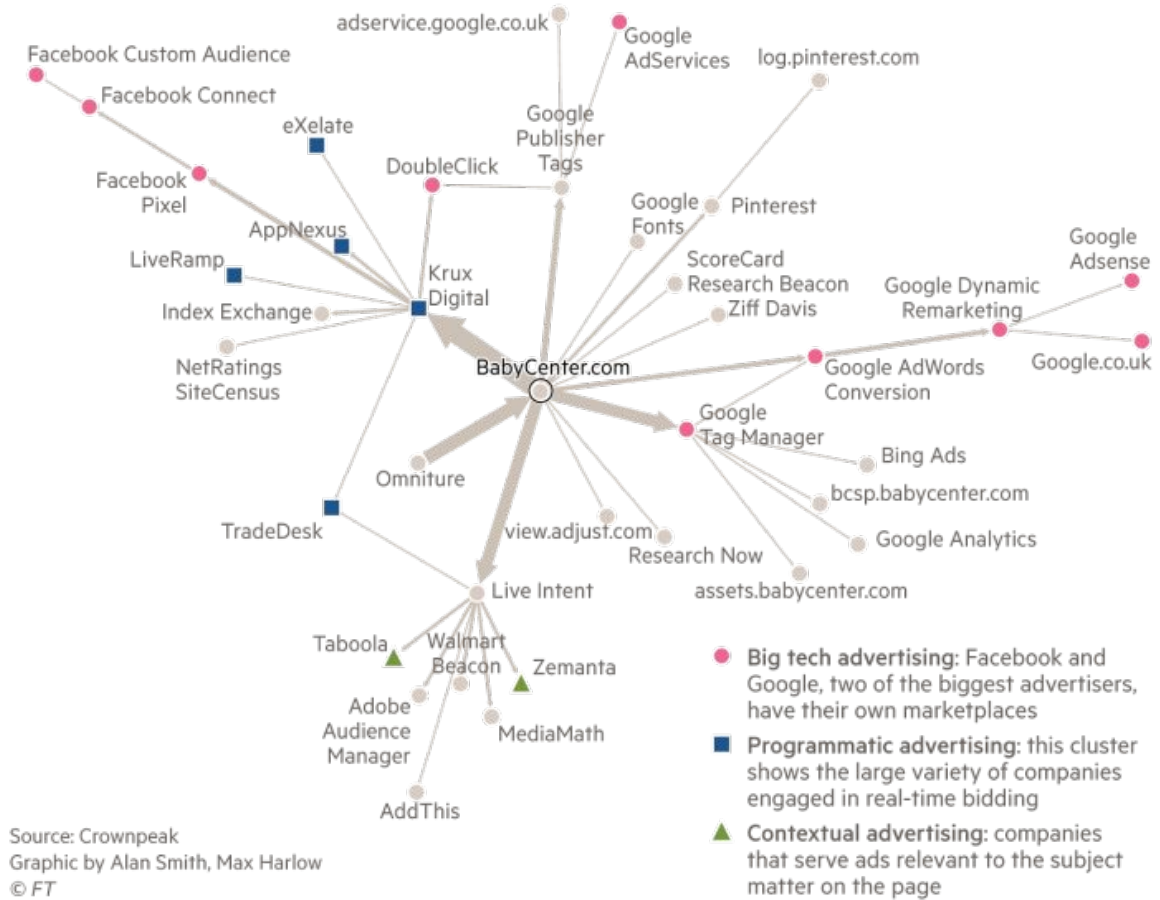
● **Big tech advertising:** Facebook and Google, two of the biggest advertisers, have their own marketplaces

■ **Programmatic advertising:** this cluster shows the large variety of companies engaged in real-time bidding

▲ **Contextual advertising:** companies that serve ads relevant to the subject matter on the page

# Sharing with whom?

THE COMMON THREAD
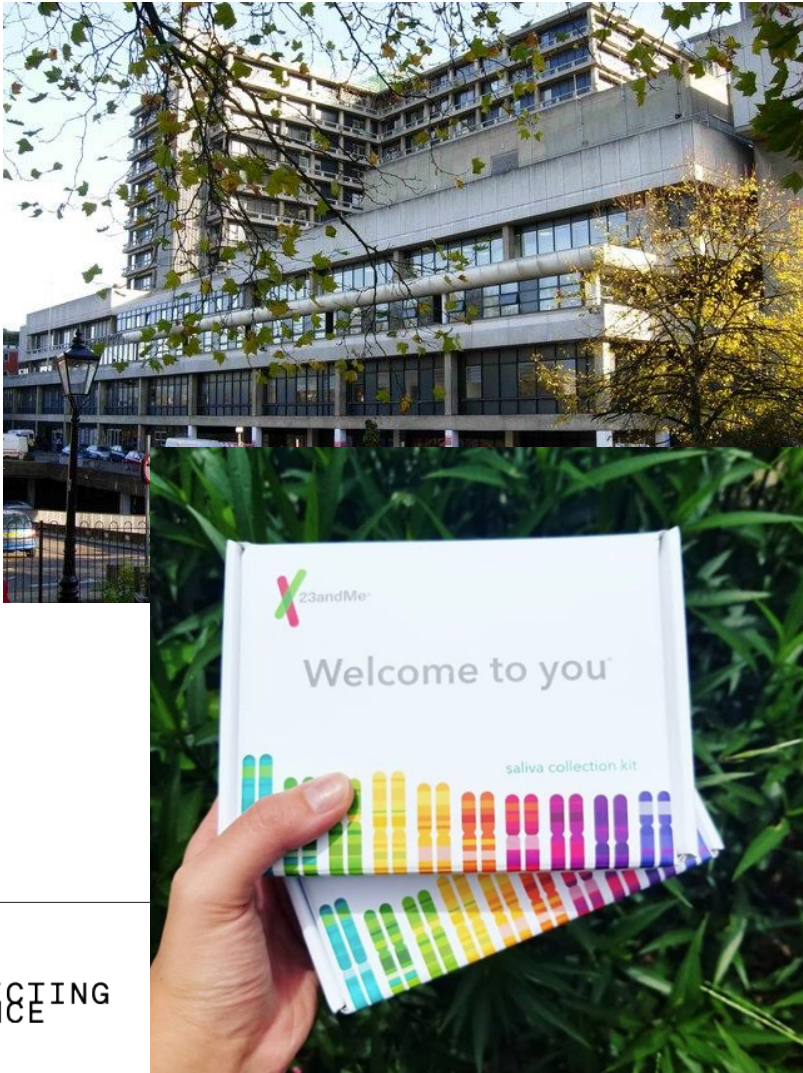
THE SUNDAY TIMES

## Amazon ready to cash in on free access to NHS data

MYR

The Observer
General election 2019

## Patient data from GP surgeries sold to US companies

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

23andMe

Welcome to you®

saliva collection kit

◆ 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

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# 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



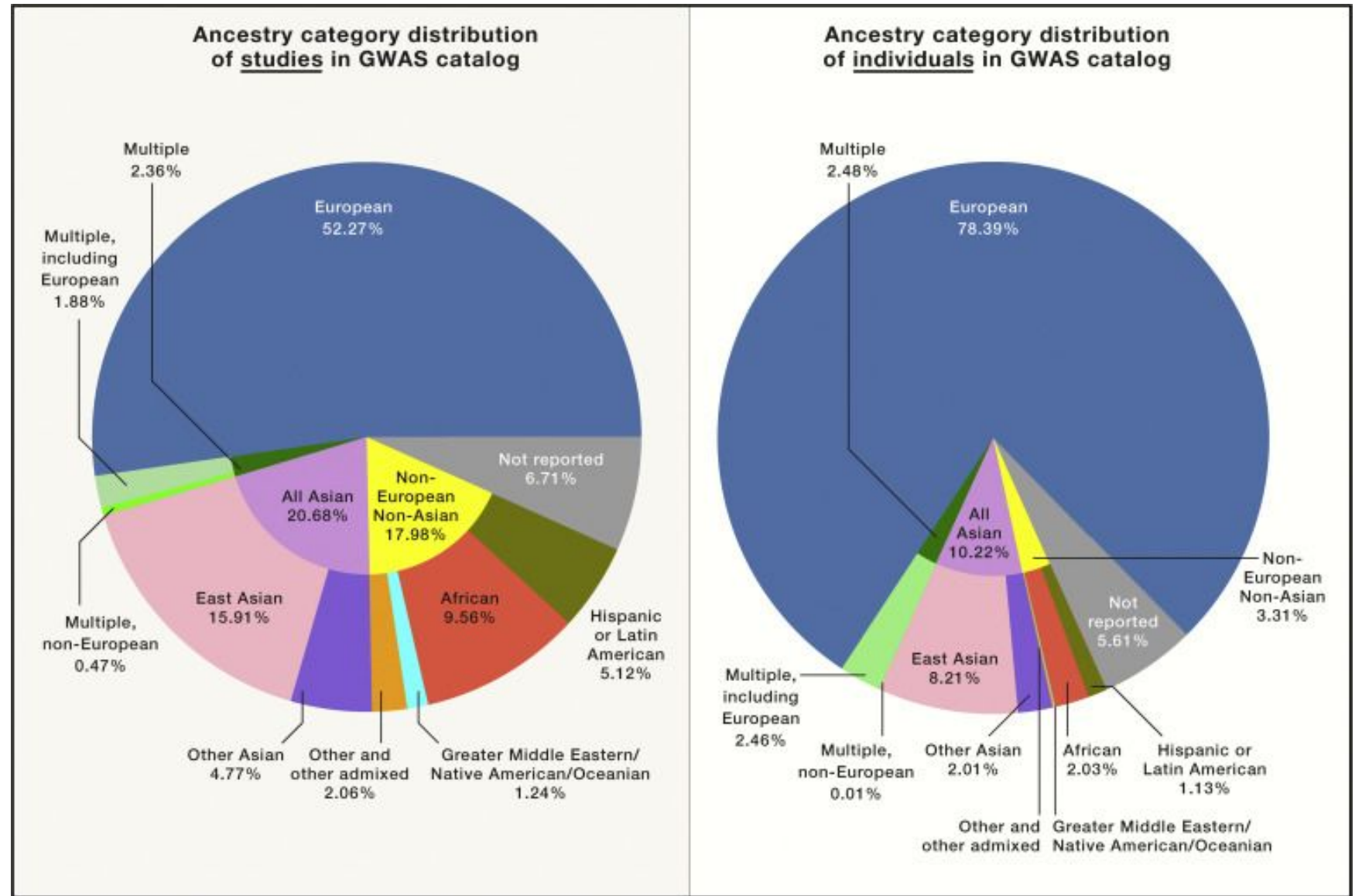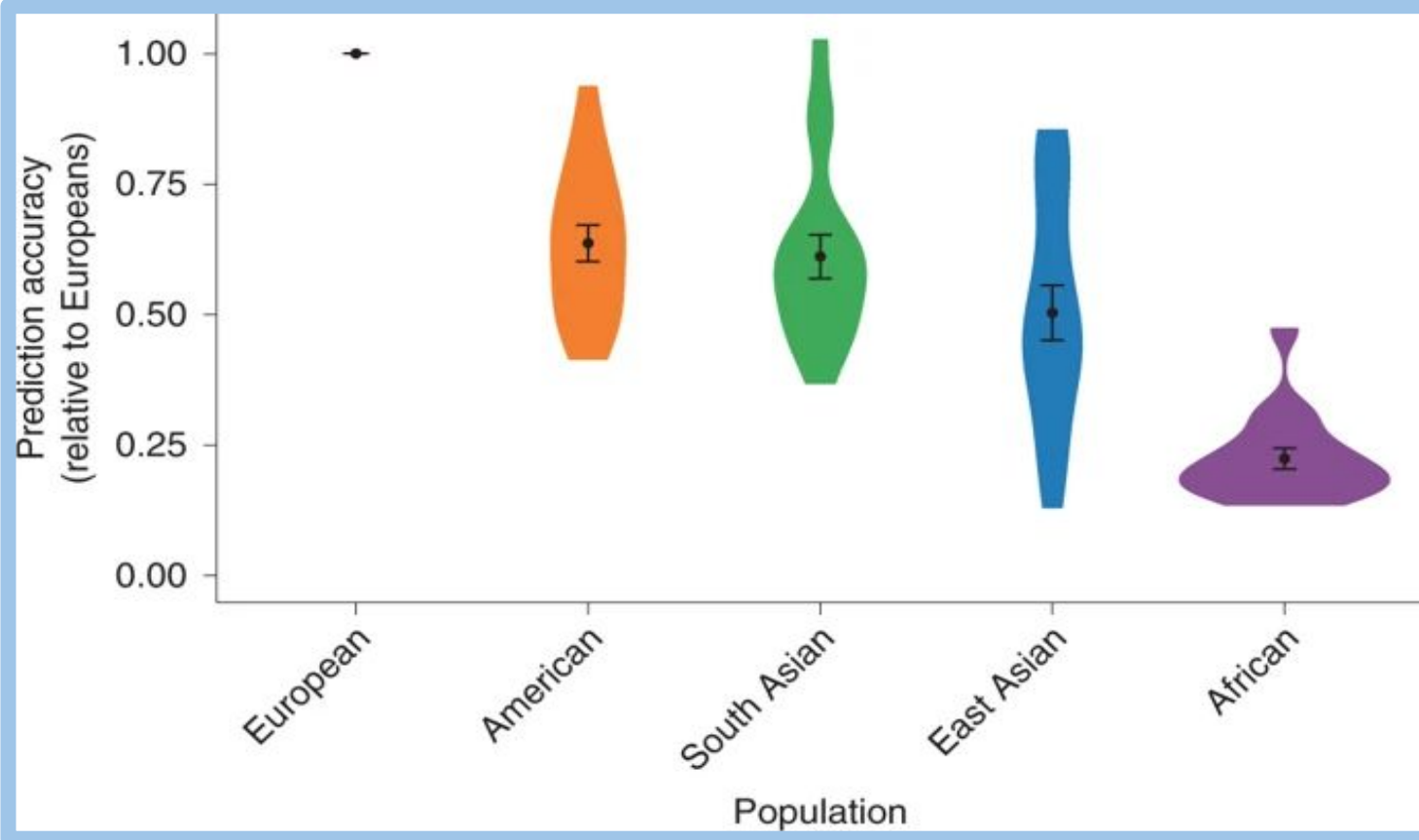Better information means better care

# 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)





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**Ancestry category distribution of studies in GWAS catalog**

- Multiple 2.36%
- Multiple, including European 1.88%
- Multiple, non-European 0.47%
- European 52.27%
- All Asian 20.68%
- Non-European Non-Asian 17.98%
- Not reported 6.71%
- East Asian 15.91%
- Other Asian 4.77%
- Other and other admixed 2.06%
- Greater Middle Eastern/ Native American/Oceanian 1.24%
- African 9.56%
- Hispanic or Latin American 5.12%

**Ancestry category distribution of individuals in GWAS catalog**

- Multiple 2.48%
- Multiple, including European 2.46%
- Multiple, non-European 0.01%
- European 78.39%
- All Asian 10.22%
- Non-European Non-Asian 3.31%
- Not reported 5.61%
- East Asian 8.21%
- Other Asian 2.01%
- Other and other admixed
- Greater Middle Eastern/ Native American/Oceanian
- African 2.03%
- Hispanic or Latin American 1.13%

Sirugo *et al.* (2019)
https://doi.org/10.1016/j.cell.2019.02.048
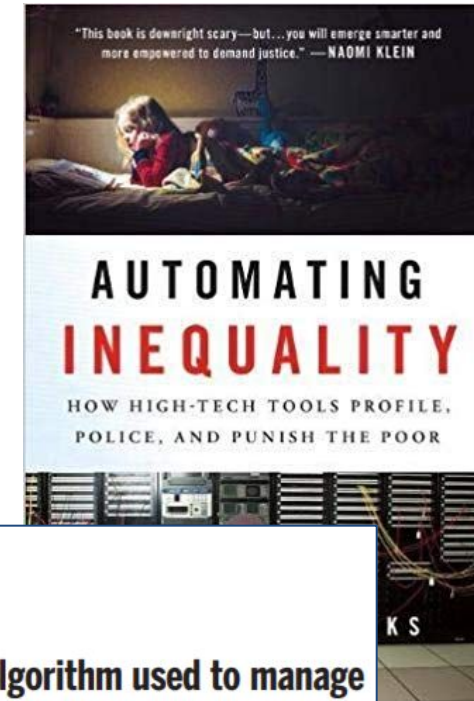
*Prediction accuracy relative to European-ancestry individuals across 17 quantitative traits and 5 continental populations in the UKBB.*

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

**RESEARCH ARTICLE**
ECONOMICS

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

Ziad Obermeyer[1,2]*, Brian Powers[3], Christine Vogeli[4], Sendhil Mullainathan[5]*†

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

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.

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# Achieving a balance

Recognition of common interest

How to maximise societal benefit from scientific research

Protection of private interests

How to protect individuals from misuse of data about them (privacy, discrimination, stigma)

*How to realise collective benefits by protecting individuals*

←——→

*How to help individuals benefit from collective action*

# **Socially Responsible Research**

Realising collective benefits from data involves ensuring that data collection sharing and use are done in a manner that ensures

- respect for persons
- respect for human rights
- participation
- accounting for decisions

The collection, linking and use of data in biomedical research and health care: ethical issues

NUFFIELD COUNCIL ON BIOETHICS

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# 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)



The collection, linking and use of data in biomedical research and health care: ethical issues

NUFFIELD COUNCIL ON BIOETHICS



Knoppers *The HUGO Journal* 2014, **8**:3
http://www.thehugojournal.com/content/8/1/3

The HUGO Journal
a SpringerOpen Journal

**REPORT**                                    **Open Access**

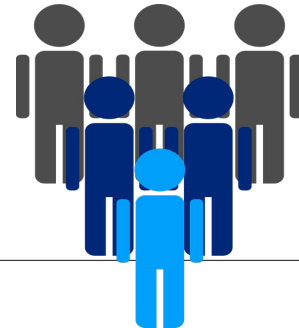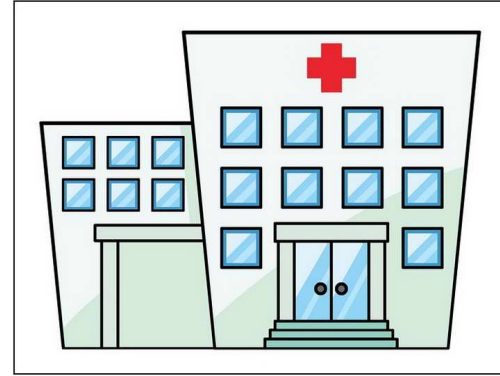Framework for responsible sharing of genomic and health-related data

Bartha Maria Knoppers

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

CONNECTING SCIENCE

# Society and Ethics Research

Embedded in clinic

… And with science

… Conducting research on stakeholder perspectives on genomics, data and society

# Global public survey

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

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جوابات دوبارہ ترتیب دیں اور شروع کریں

خوش آمدید

یہ ایک سروے ہے جو آپ کے آن لائن صحت کے ڈیٹا کے بارے میں ہے اور اسے دوسرے کیسے استعمال کر سکتے ہیں۔

یہ بہت آسان ہے - فلمیں کو دیکھیے اور چند سوالات کا جواب دیجے۔ فلمیں آپ کو سب جان کاری دینگی - اپنے ماوس سے ان الفاظ کو دیکھیے جن کے نیچے لائن ہے اور آپ کو ایک فہرست نظر آئے گی۔

سروے کوئی بھی اور سبھی بھر سکتے ہیں، لہذا آگے بڑھیں۔ آپ کی شرکت مکمل طور پر گمنام ہے اور ہم ویلکم جینوم کیمپس، کیمبرج، برطانیہ میں تمام جوابات محفوظ رکھیں گے۔

اگر آپ سوالات کے جواب شروع کرتے ہیں تو یہ آپ کی رضامندی کا اظہار ہے؛ اس کا گمنام ڈیٹا مستقبل میں سماجی سائنس دانوں کے ساتھ تحقیق کے لئے کام آسکتا ہے۔

کسی بھی وقت، آپ اس صفحہ کو بک مارک کر سکتے ہیں اور بعد میں واپس آسکتے ہیں۔

یہ تحقیق منافع کے لئے نہیں بلکہ انسانوں کی خدمت کے لیے ہے۔

films can be skipped, paused or watched again

**اگلا »**

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reset answers and start again

**الحمض النووي الخاص بك**

✓ أهلاً وسهلاً
✓ البصمة الإلكترونية
✓ إمكانية الوصول للبيانات من قبل آخرين
✓ إمكانية الوصول من قبل الأطباء المختصين
✓ إمكانية الوصول من قبل الباحثين غير هادفي الربح
✓ إمكانية الوصول من قبل الباحثين الهادفين للربح
• الأضرار المتوقعة
  ○ المتوقع من المعلومات
  ○ الثقة
  ○ آخر الأسئلة المتعلقة بك
  ○ أرسل إجاباتك

"شارلوك هولمز"

films can be skipped, paused or watched again

**الأضرار المتوقعة**

لنفترض أنه قد أعطيت الخيار للتبرع بالحمض النووي الخاص بك و بمعلوماتك الطبية بحيث تكون مجهول الهوية وذلك من أجل أن تستخدم من قبل أيا مما يلي:

• الأطباء المختصين (مثال : من أجل مساعدتك في تشخيص الحالات المرضية والقيام بنشر الأبحاث العلمية)
• الباحثين الغير هادفي الربح (مثال: من أجل القيام بأبحاث طبية و أيضا للحصول على تمويل جديد)
• الشركات الهادفة للربح (مثال : من أجل تطوير الأدوية والحصول على مال من وراء ذلك)

ضمن السيناريوهات الثلاثة المذكورة أعلاه، لنفترض أيضا أن هناك فرصة أن يتم التعرف عليك شخصيا من قبل أحد الباحثين القادرين على الوصول الى قاعدة بياناتك. نريد معرفة ماذا تعتقد ...

○ لن أقوم بالتبرع بمعلومات عن الحمض النووي الخاص بي و معلوماتي الطبية.
● سوف أقوم بالتبرع بمعلومات عن الحمض النووي الخاص بي و معلوماتي الطبية ليتم استخدامها من قبل ... ( اختر من الاجابات التالية بالضغط على المربع )

**الأطباء المختصين**

| لا أعلم | سأقبل بنسبة 90% أن يتم التعرف علي | سأقبل بنسبة 50% أن يتم التعرف علي | سأقبل بنسبة 10% أن يتم التعرف علي | سأقبل بنسبة 1% أن يتم التعرف علي | لن أقوم بالتبرع |

**الباحثون الغير هادفي الربح**

| لا أعلم | سأقبل بنسبة 90% أن يتم التعرف علي | سأقبل بنسبة 50% أن يتم التعرف علي | سأقبل بنسبة 10% أن يتم التعرف علي | سأقبل بنسبة 1% أن يتم التعرف علي | لن أقوم بالتبرع |

**الباحثون الهادفون للربح**

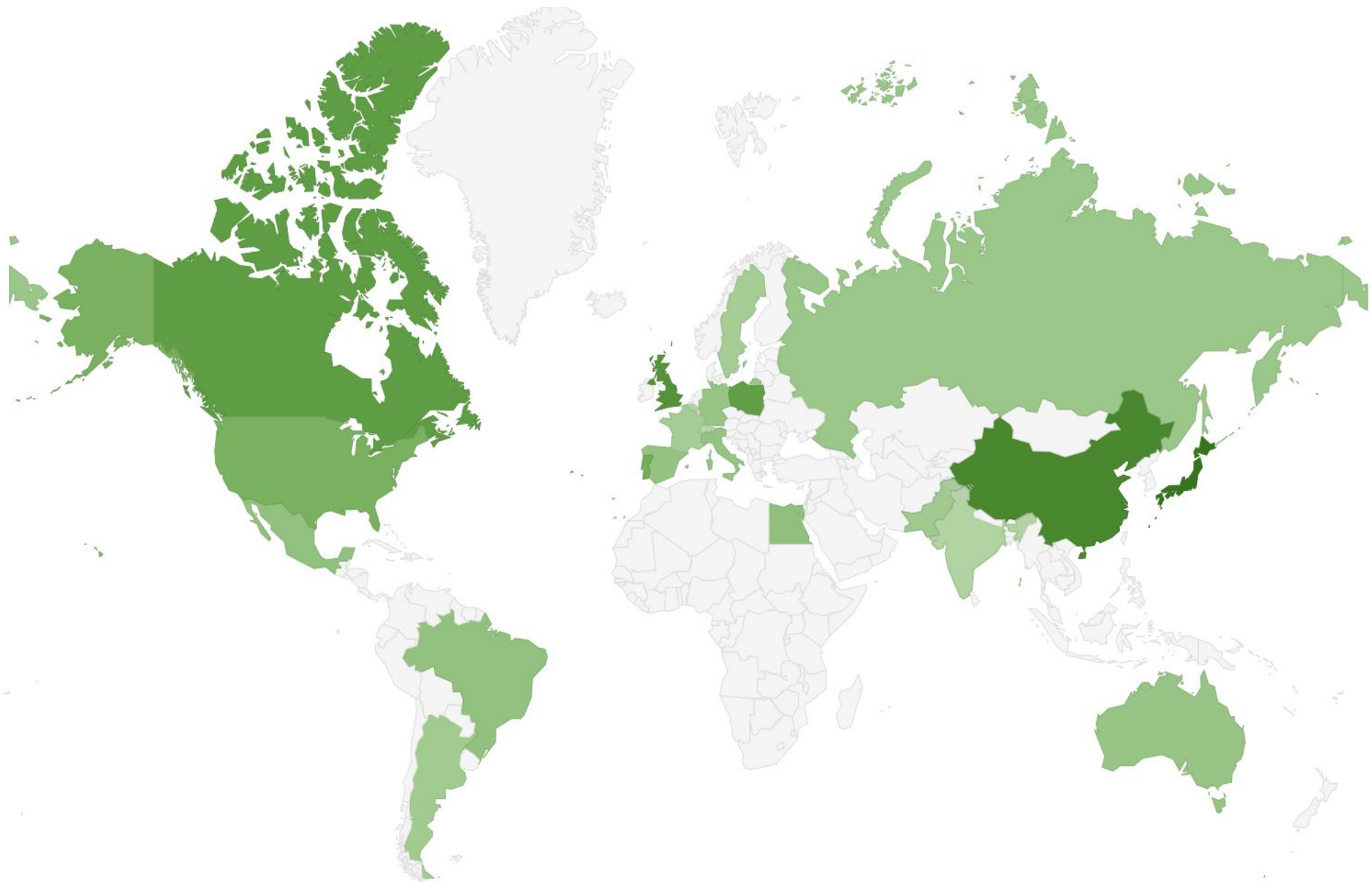| لا أعلم | سأقبل بنسبة 90% أن يتم التعرف علي | سأقبل بنسبة 50% أن يتم التعرف علي | سأقبل بنسبة 10% أن يتم التعرف علي | سأقبل بنسبة 1% أن يتم التعرف علي | لن أقوم بالتبرع |

تعليقات

**»التالي** — **»السابق**

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回答をリセットし、やり直す

**あなたのDNA、あなたの意見**

الحمض النووي الخاص بك

**العربية**

Ihre DNA, Ihre Entscheidung
**Deutsch**

Your DNA, Your Say
**English**

Tu DNA, Tu Decisión
**Español**

Votre ADN, Votre AVIS
**Français**

Þitt erfðaefni, þín ákvörðun
**Íslenska**

É il tuo DNA, Decidi Tu
**Italiano**

Twoje DNA, Twoje zdanie
**Polski**

O seu ADN, a sua voz
**Português**

Ваши гены - Вам решать
**Русский**

films can be skipped, paused or watched again

**ようこそ**

このアンケートでは、オンライン上にあるみなさんの健康情報や、他の人によるその情報の利用について質問します。

動画を見ていくつかの質問に答えるという簡単な形式です。回答に必要な情報は動画に含まれています。下線が引かれた単語の上にマウスを置くと、その用語の解説をみることができます。

このアンケートは誰でも入力できますので気軽にご参加ください。みなさんの回答は、英国ケンブリッジにあるWellcome Genome Campusにおいて匿名化された状態で安全に保管されます。

質問への回答開始をもって、アンケートへの参加に同意したものとみなされます。このアンケートで得られる匿名化されたデータは、将来の研究のために他の社会科学者と共有する可能性があります。

どの時点でも、このページをブックマークすれば後で戻ってくることができます。

この研究は、利益のためではなく、ひとびとのために行います。

**次 »**

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CONNECTING SCIENCE

Canada

*Iceland*

Sweden

Russia

Belgium

Poland

UK

Germany

China

France

USA

Portugal

Italy

Spain

Japan

Switzerland

Egypt

Pakistan

Mexico

*Qatar*

India

*Ghana*

Brazil

Argentina

Australia

Currently available

*Coming soon*

# Willingness to donate DNA and health data

# Willingness to donate (overall)



Legend: Yes (dark blue) | Unsure (light blue) | No (grey)

Countries (top to bottom):
Mexico, Russian Federation, Italy, Portugal, India, Brazil, Poland, United States, Germany, Switzerland, France, Pakistan, Argentina, Canada, Australia, Sweden, Belgium, Spain, United Kingdom, Egypt, Japan

X-axis: 0%, 25%, 50%, 75%, 100%
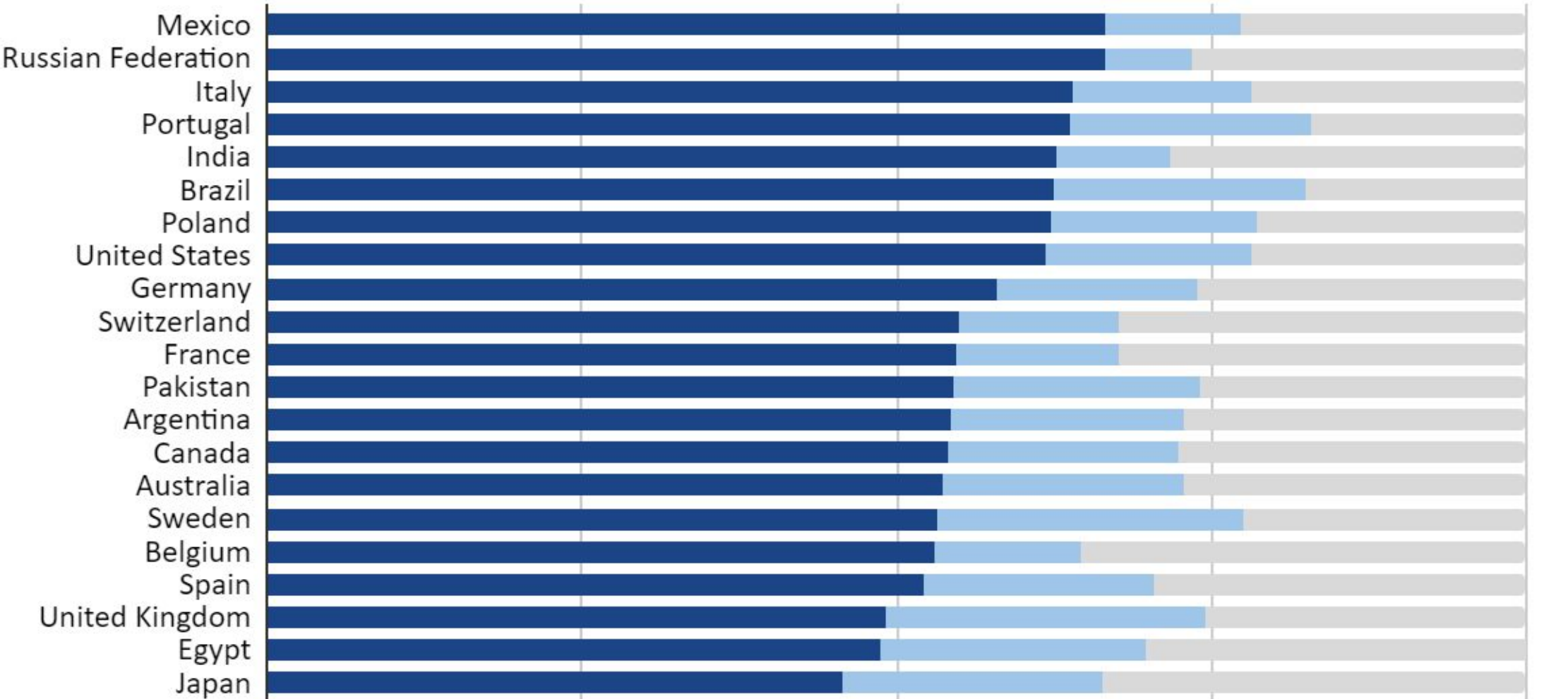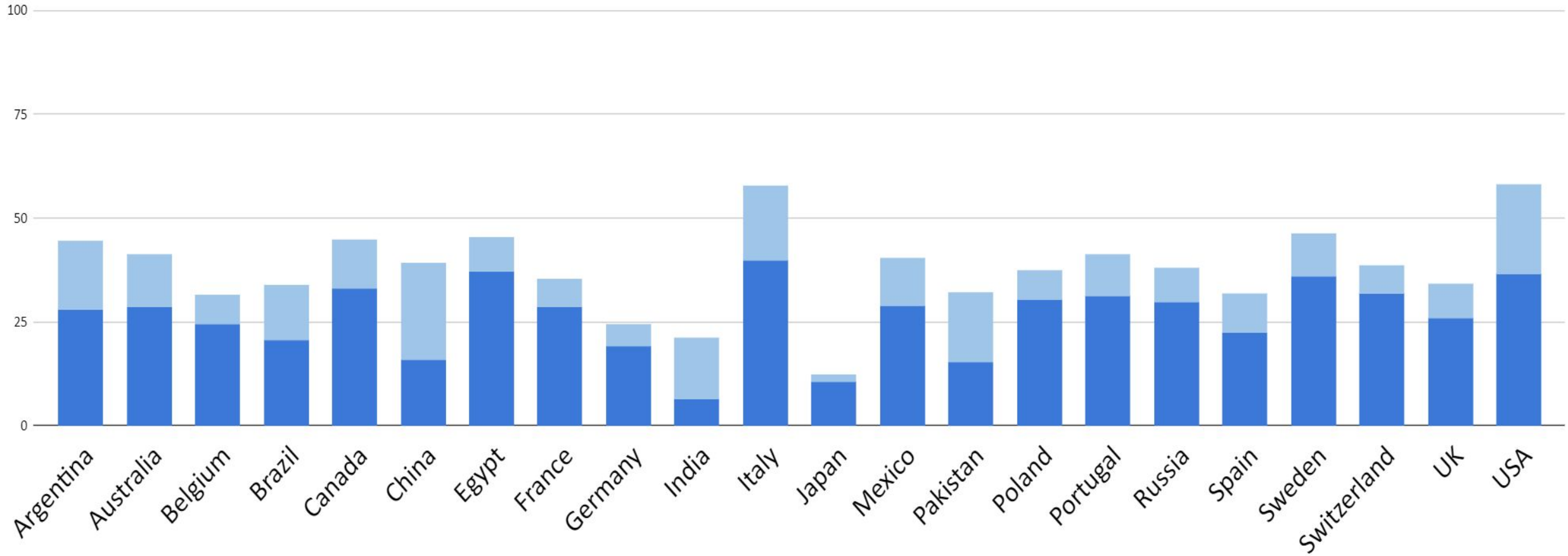
**What is being donated?**

**With whom?**

**Why?**

- Familiarity with genomics
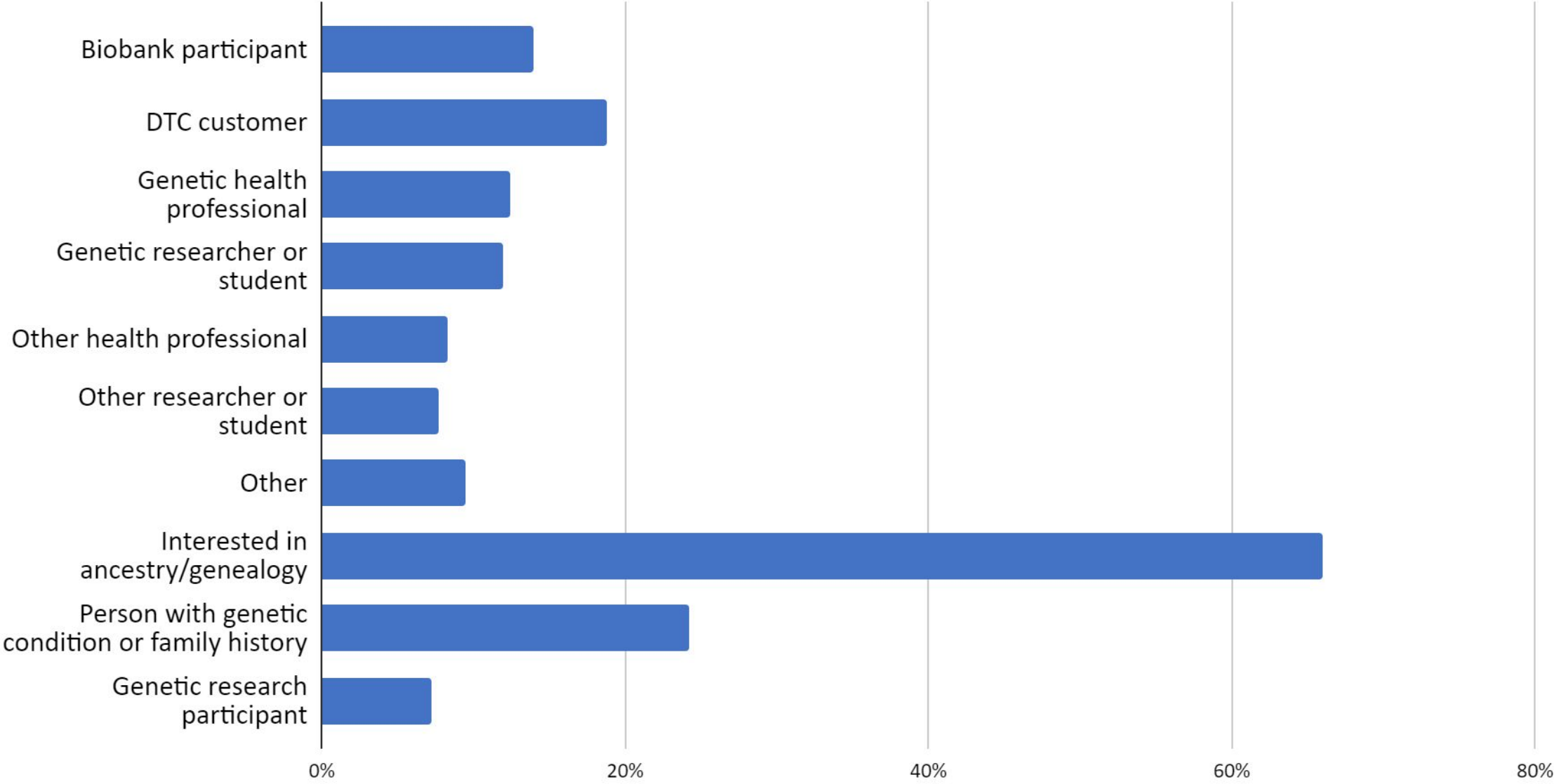- Is there is something special about genomic data?

# Familiarity

"Are you familiar with DNA, genetics or genomics?"

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Familiarity with genomics

# Source of familiarity with DNA/genetics/genomics - USA only



United States

| Country | OR [95% CI] |
|---|---|
| Argentina | 1.47 [1.07; 2.02] |
| Australia | 1.60 [1.22; 2.08] |
| Belgium | 1.69 [1.11; 2.57] |
| Brazil | 2.16 [1.61; 2.90] |
| Canada | 1.77 [1.50; 2.09] |
| China | 2.42 [1.89; 3.11] |
| Egypt | 1.96 [1.53; 2.51] |
| France | 1.56 [1.12; 2.17] |
| Germany | 1.70 [1.26; 2.30] |
| India | 2.23 [1.00; 4.97] |
| Italy | 1.45 [1.12; 1.87] |
| Japan | 2.20 [1.81; 2.68] |
| Mexico | 1.81 [1.39; 2.37] |
| Pakistan | 1.56 [1.06; 2.28] |
| Poland | 1.80 [1.52; 2.12] |
| Portugal | 1.50 [1.23; 1.82] |
| Russian Federation | 2.35 [1.78; 3.10] |
| Spain | 1.62 [1.22; 2.16] |
| Sweden | 1.75 [1.28; 2.38] |
| Switzerland | 1.47 [0.89; 2.41] |
| United Kingdom | 2.13 [1.81; 2.50] |
| United States | 2.61 [2.10; 3.25] |
| Total | 1.85 [1.71; 2.00] |

Heterogeneity: $\chi^2_{21}$ = 40.05 ($P$ < .01), $I^2$ = 48%
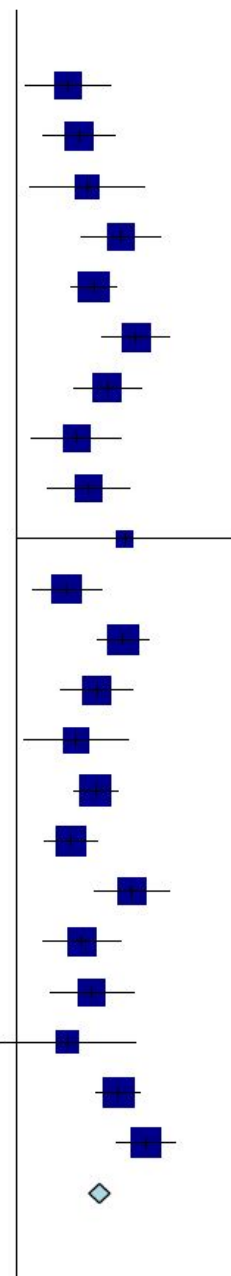
Genetics experience = Personal

| Country | OR [95% CI] |
|---|---|
| Argentina | 2.95 [1.91; 4.56] |
| Australia | 2.50 [1.73; 3.61] |
| Belgium | 2.54 [1.23; 5.26] |
| Brazil | 2.47 [1.72; 3.55] |
| Canada | 2.74 [2.15; 3.51] |
| China | 3.65 [2.87; 4.64] |
| Egypt | 3.29 [2.19; 4.96] |
| France | 3.08 [1.61; 5.91] |
| Germany | 4.60 [2.56; 8.24] |
| India | 3.88 [2.13; 7.06] |
| Italy | 1.68 [1.21; 2.32] |
| Japan | 2.97 [1.90; 4.64] |
| Mexico | 2.28 [1.53; 3.39] |
| Pakistan | 2.12 [1.46; 3.10] |
| Poland | 3.13 [2.28; 4.31] |
| Portugal | 1.75 [1.29; 2.38] |
| Russian Federation | 3.47 [2.14; 5.64] |
| Spain | 1.51 [1.00; 2.27] |
| Sweden | 1.79 [1.10; 2.90] |
| Switzerland | 3.60 [1.25; 10.39] |
| United Kingdom | 3.05 [2.36; 3.95] |
| United States | 4.16 [3.17; 5.47] |
| Total | 2.70 [2.37; 3.09] |

Heterogeneity: $\chi^2_{21}$ = 53.81 ($P$ < .01), $I^2$ = 61%
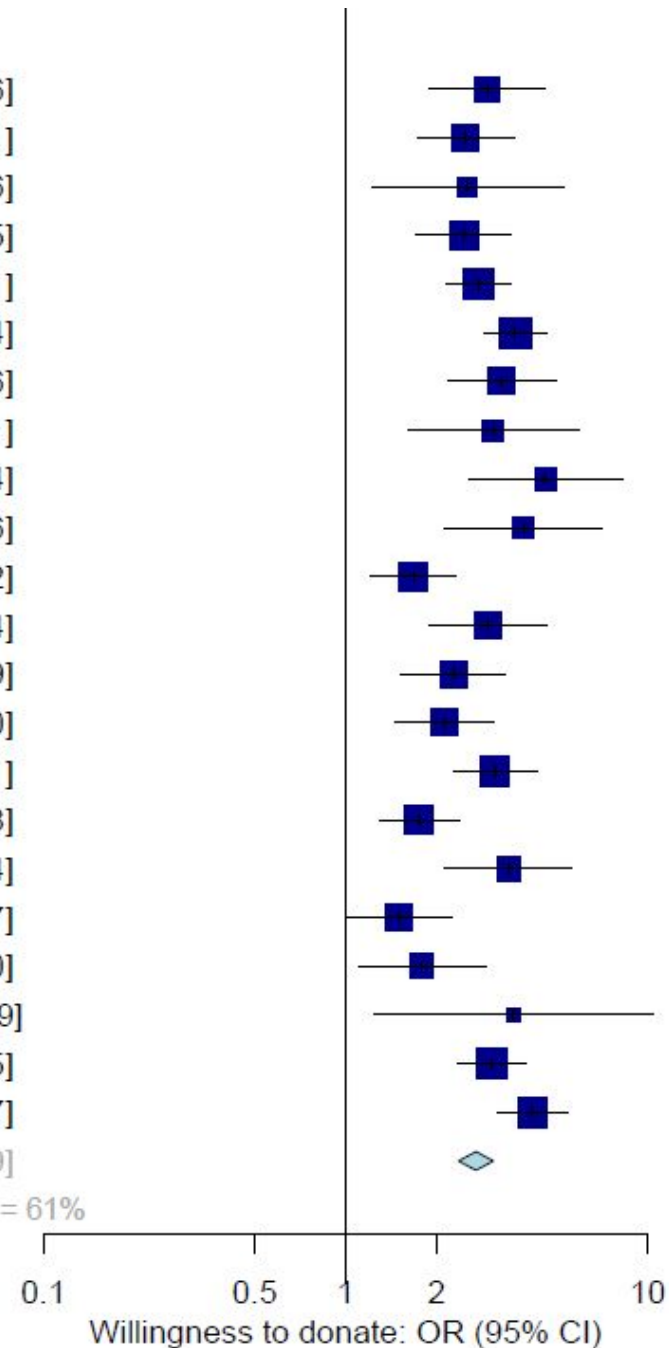
0.1   0.5   1   2   10

Willingness to donate: OR (95% CI)

# The importance of familiarity

Overall familiarity with genomics is low

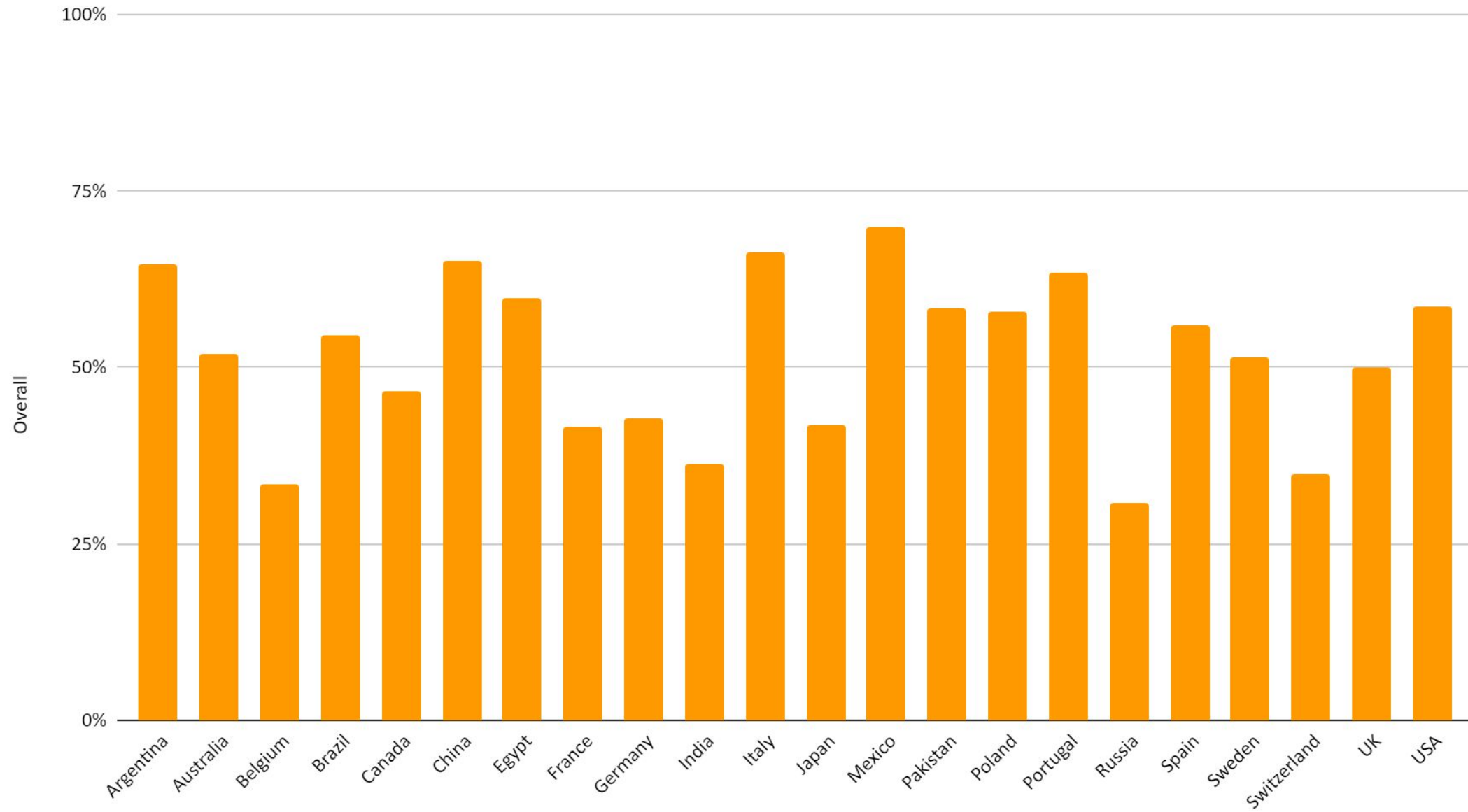Those people who are most familiar with DNA are more willing to donate

Those with personal familiarity are most willing to donate

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# Are genomic data different?

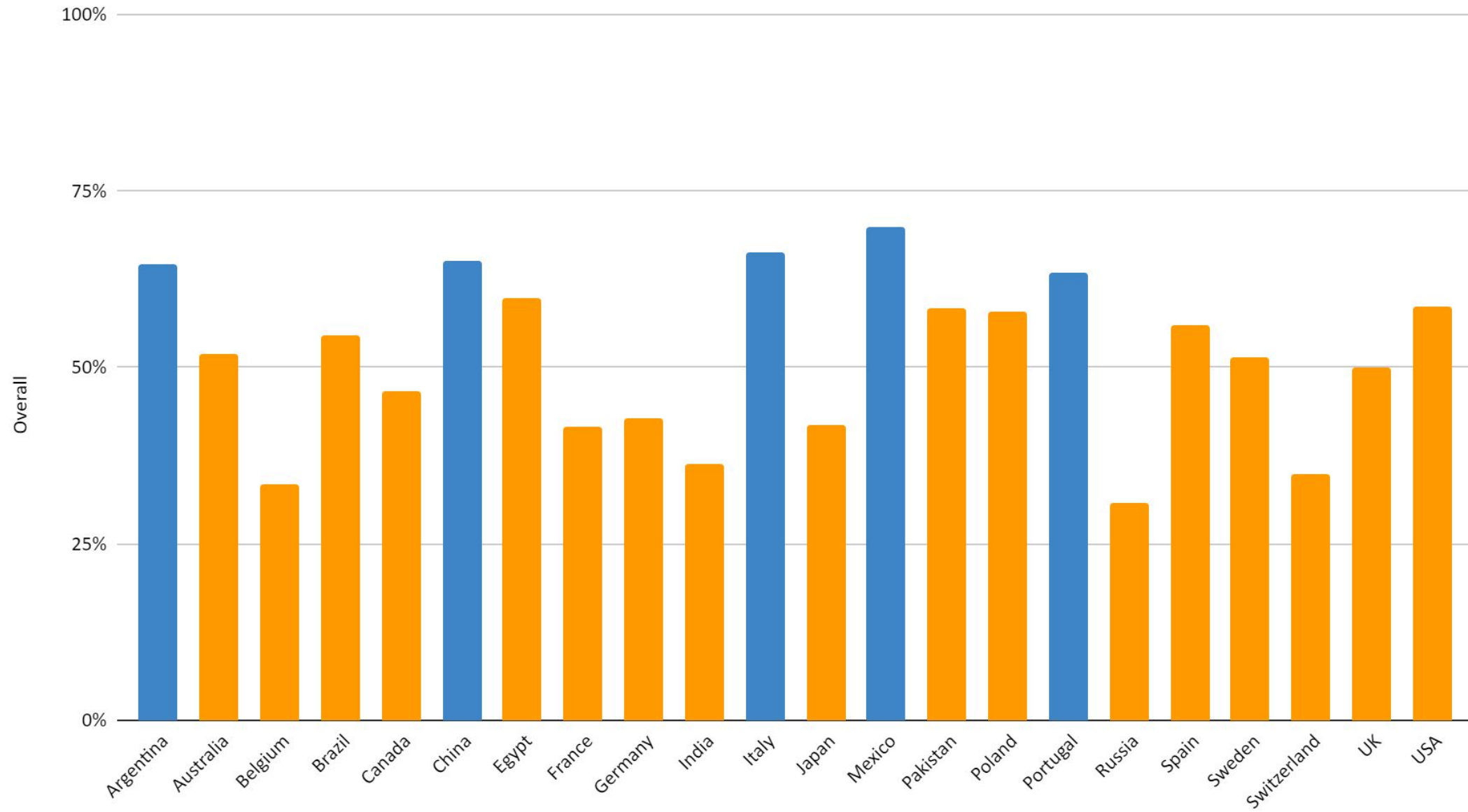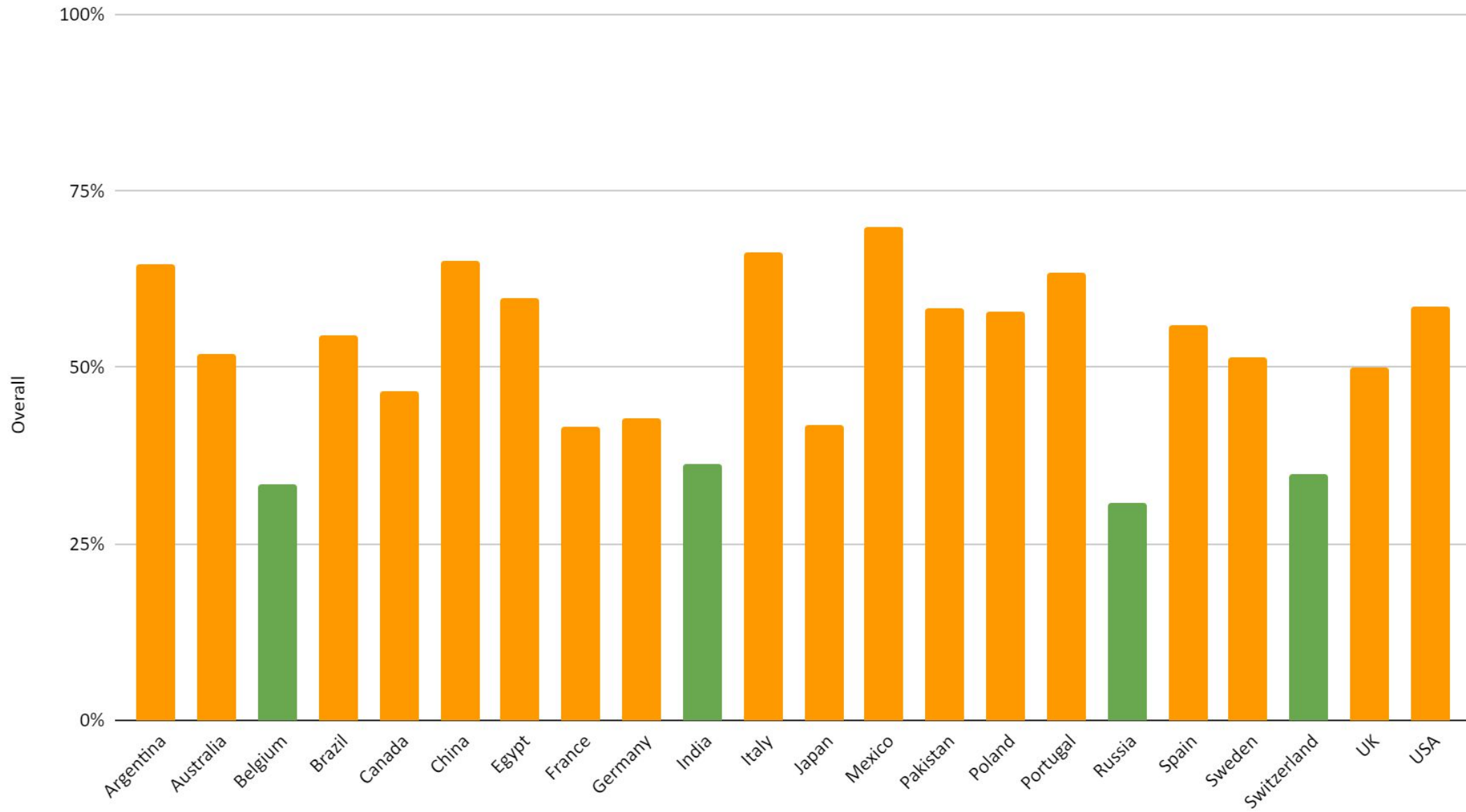# Exceptionalism

"For me DNA information is different to other medical information"

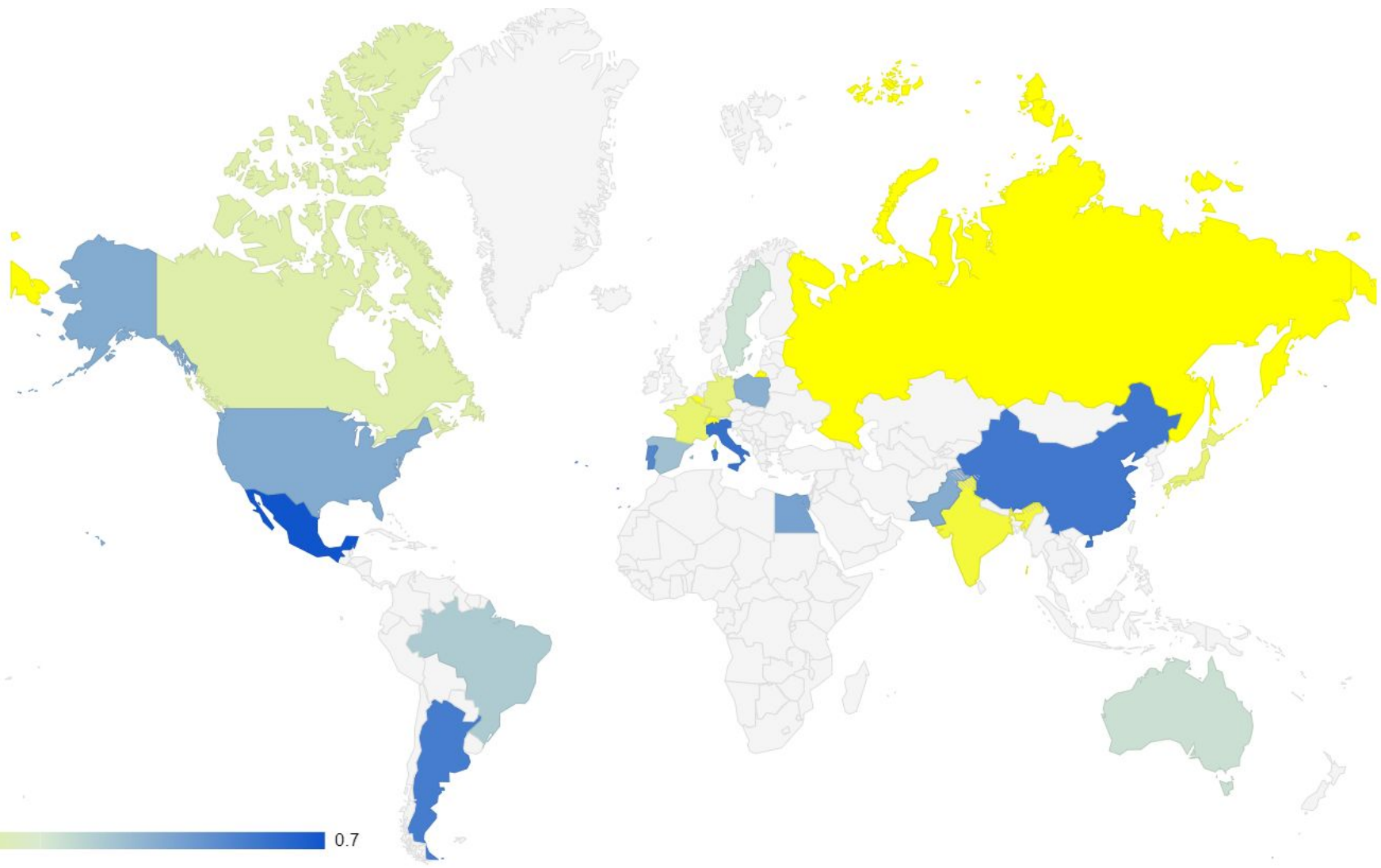"For Me DNA Information is Different to Other Medical Information"

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"For Me DNA Information is Different to Other Medical Information"

39

"For Me DNA Information is Different to Other Medical Information"
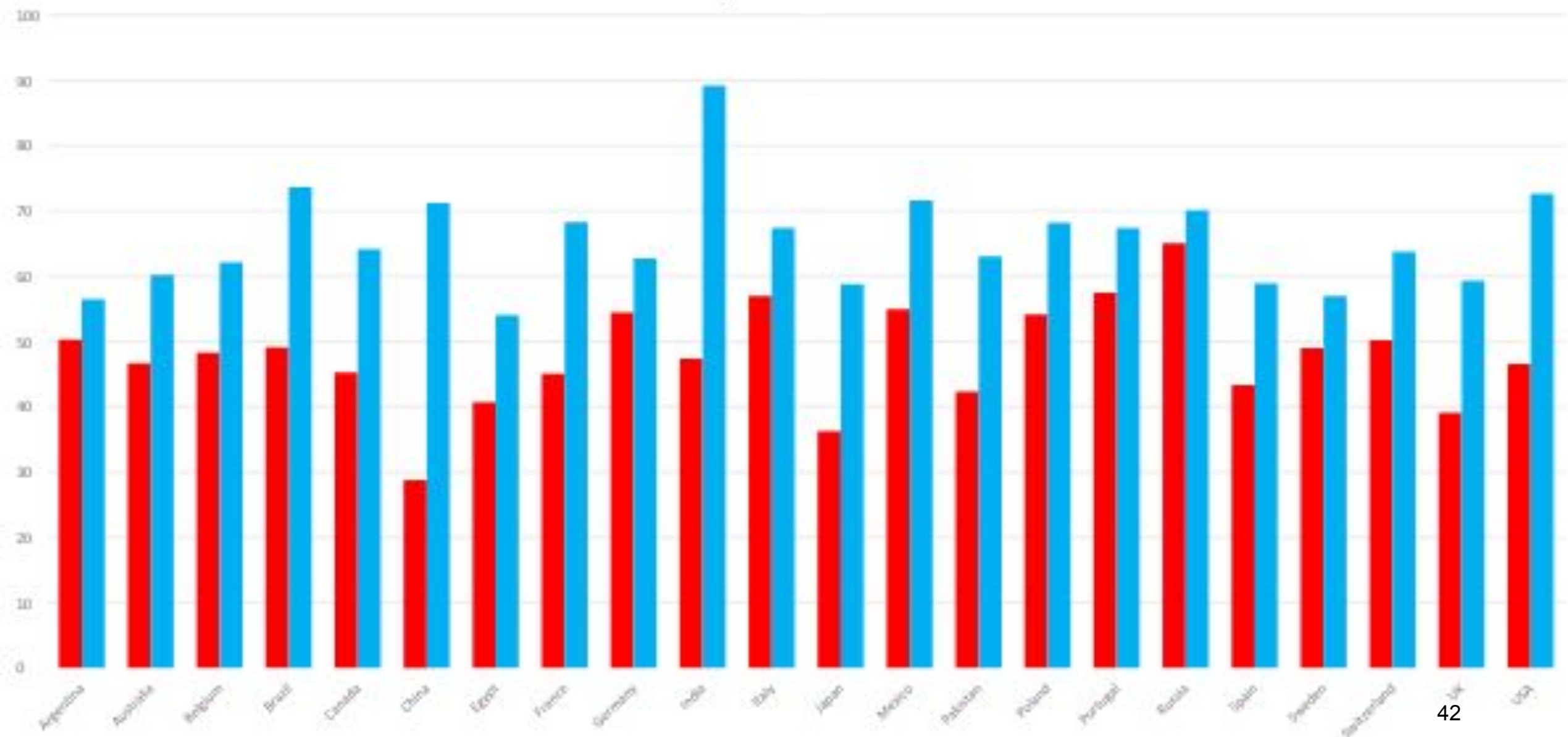
40

0.308 ▮ 0.7

% of those who see DNA information as same/different to other medical information who are willing to donate DNA data

■Same/unsure ■Different

| Source | OR (95% CI) |
|---|---|
| **DNA Status = Different** | |
| Argentina | 1.51 [1.13; 2.01] |
| Australia | 1.32 [1.05; 1.67] |
| Belgium | 1.15 [0.79; 1.66] |
| Brazil | 2.15 [1.72; 2.69] |
| Canada | 1.57 [1.36; 1.83] |
| China | 2.01 [1.72; 2.35] |
| Egypt | 1.35 [1.07; 1.71] |
| France | 1.62 [1.20; 2.17] |
| Germany | 1.36 [1.07; 1.72] |
| India | 3.19 [2.10; 4.85] |
| Italy | 1.20 [0.94; 1.53] |
| Japan | 1.78 [1.56; 2.02] |
| Mexico | 1.87 [1.46; 2.38] |
| Pakistan | 2.30 [1.75; 3.03] |
| Poland | 1.40 [1.21; 1.63] |
| Portugal | 1.41 [1.19; 1.69] |
| Russian Federation | 1.19 [0.91; 1.55] |
| Spain | 1.40 [1.12; 1.76] |
| Sweden | 1.39 [1.05; 1.85] |
| Switzerland | 1.64 [1.02; 2.63] |
| United Kingdom | 1.77 [1.54; 2.04] |
| United States | 1.99 [1.64; 2.42] |
| Total | 1.60 [1.47; 1.75] |

Heterogeneity: $\chi^2_{21} = 69.22$ ($P < .01$), $I^2 = 70\%$
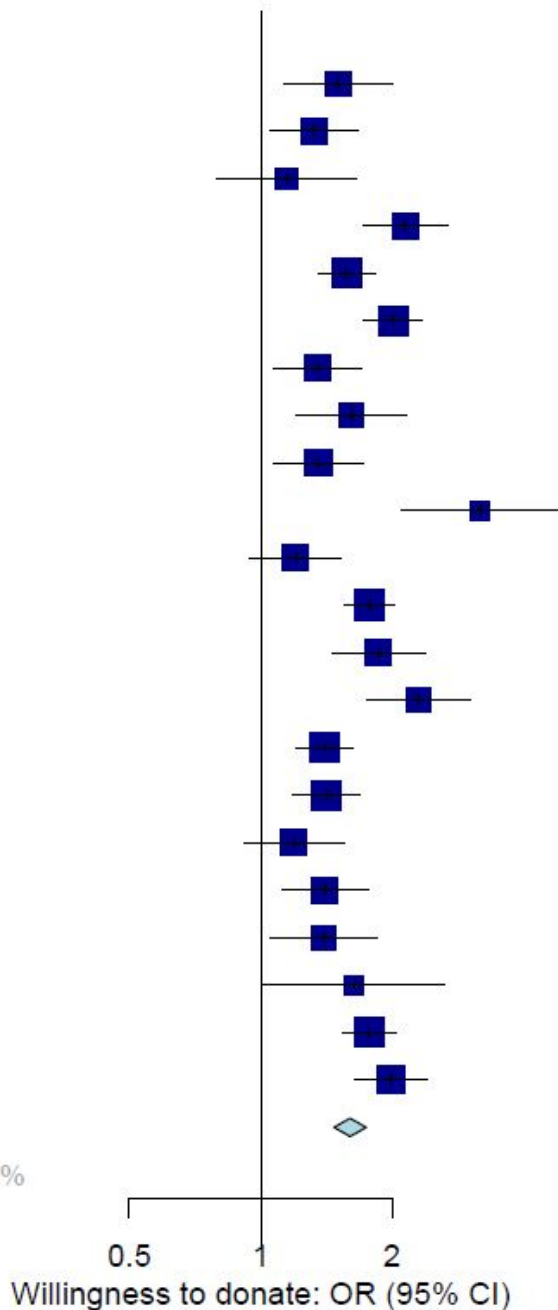
Willingness to donate: OR (95% CI)

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## Members of the public in the USA, UK, Canada and Australia expressing genetic exceptionalism say they are more willing to donate genomic data

Anna Middleton ✉, Richard Milne, Heidi Howard, Emilia Niemiec, Lauren Robarts,
Christine Critchley, Dianne Nicol, Barbara Prainsack, Jerome Atutornu, Danya F. Vears,
James Smith, Claire Steed, Paul Bevan, Erick R. Scott, Jason Bobe, Peter Goodhand,
Erika Kleiderman, Adrian Thorogood, Katherine I. Morley & on behalf of the
Participant Values Work Stream of the Global Alliance for Genomics and Health

Those who saw genomic data as different were:

- more likely to be familiar with or have personal experience with genomics
- more likely to see genomic information as having personal and economic value.
- more likely to make decisions about donation based on whether they could obtain a copy of their own raw data (cf Ruiz-Canela et al, 2011)
- Those with personal experience + genetic exceptionalist views were far more likely to be willing to donate their anonymous DNA and medical information for research than those without

**What is being shared?**

**With whom?**

**Why?**

- Familiarity with genomics
- Is there is something special about genomic data?

- Who the recipient is
- Trust in the recipient

# Who are data donated to?

# Would you donate your anonymous DNA information and medical information for use by:

- Medical doctors

- Non-profit researchers

- For-profit researchers

Willingness to donate DNA and medical information

Legend: Donate for use by Dr · Donate for use by non-profit researcher · Donate for use by for-profit researcher

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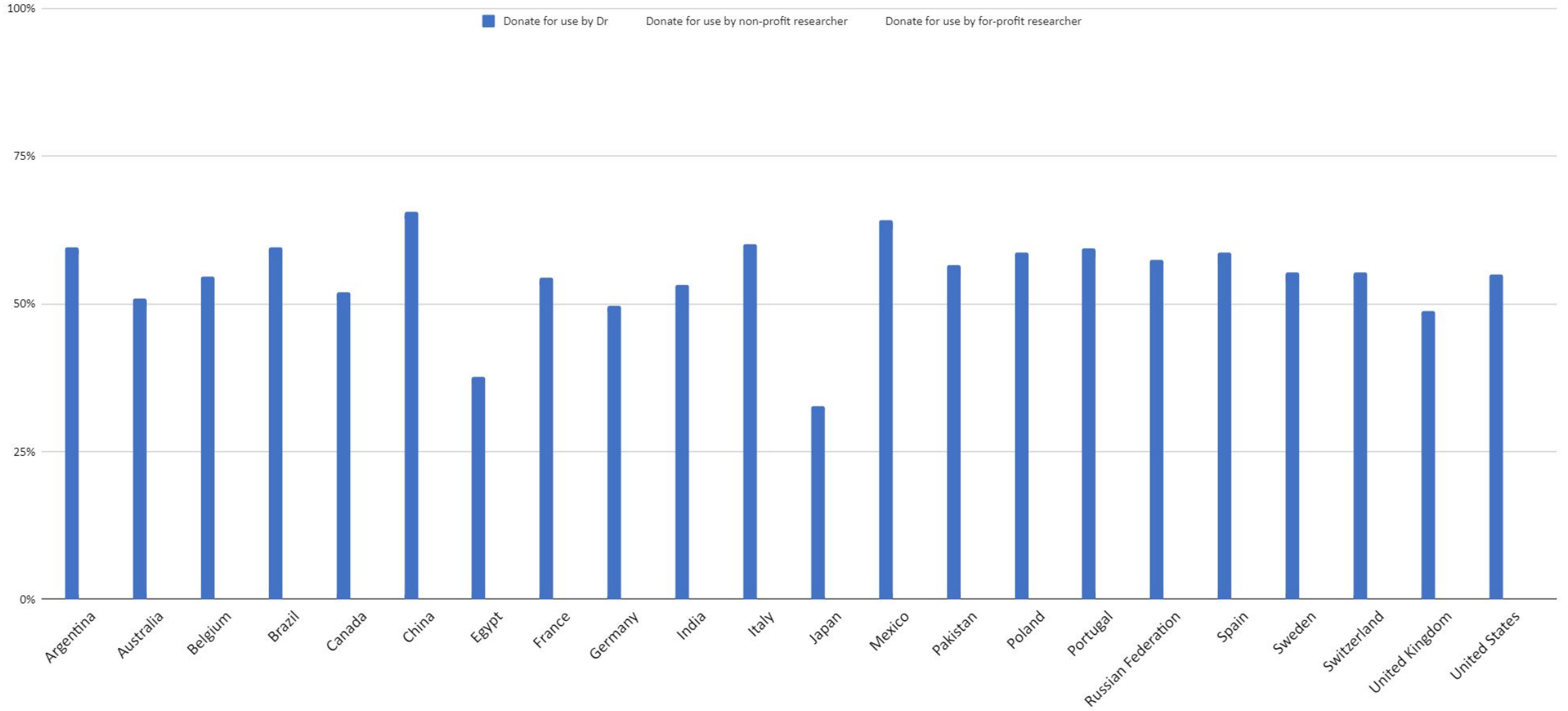
Willingness to donate DNA and medical information

Willingness to donate DNA and medical information

Donate for use by Dr | Donate for use by non-profit researcher | Donate for use by for-profit researcher
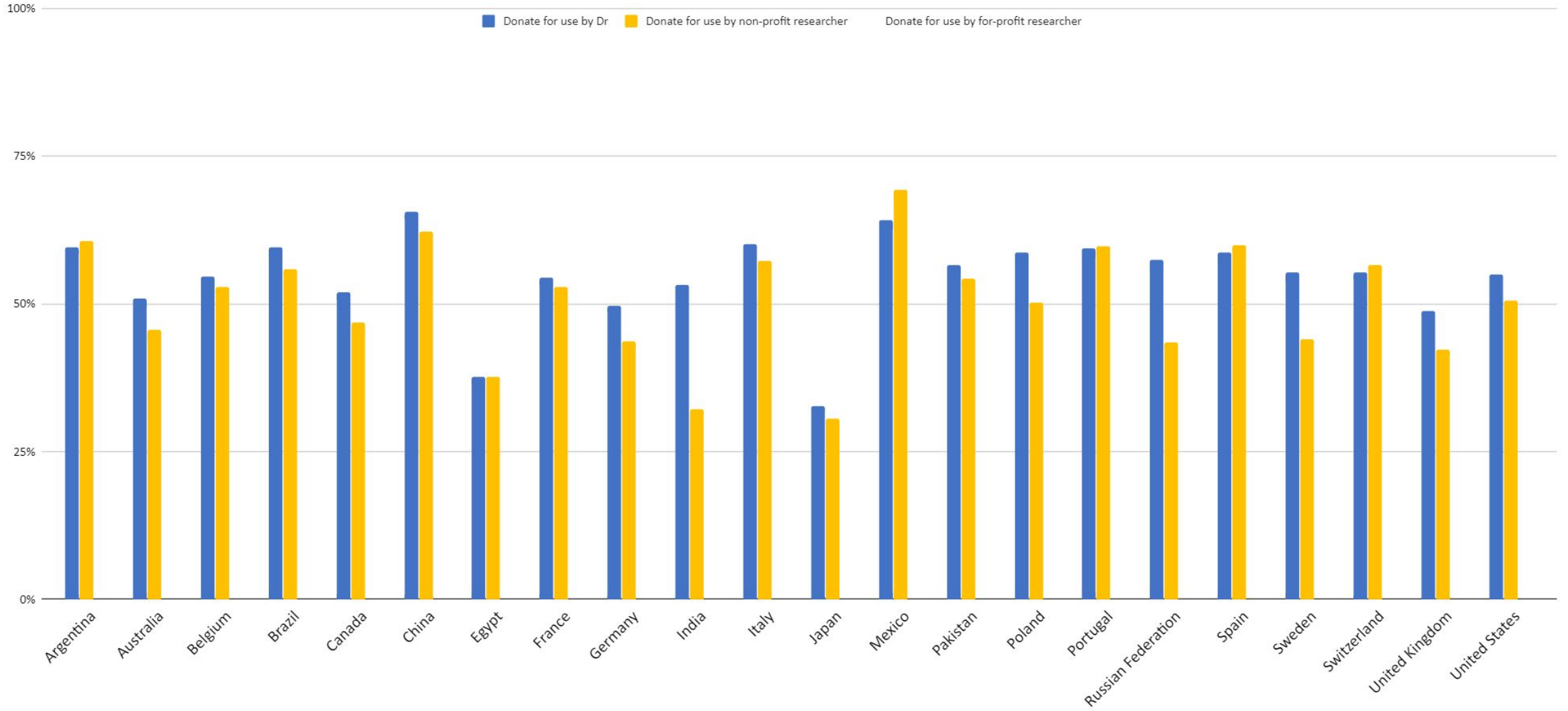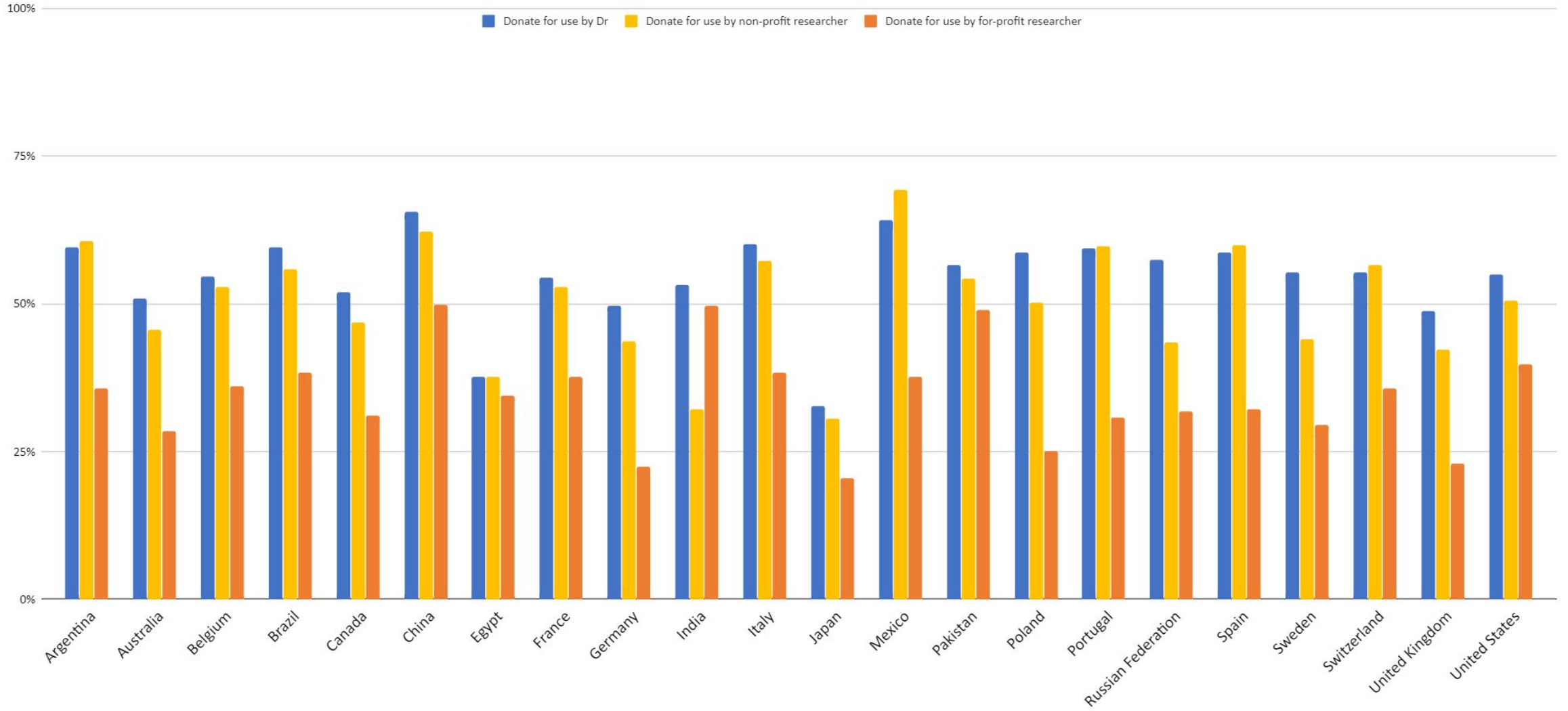
Across 22 countries, **global publics** are **more comfortable** with their data being used by **doctors** than **for-profit companies**

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# Trust

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

■ Own doctor  ■ Any doctor in country  ■ Non-profit researcher in my country  ■ Company researcher in my country  ■ My government
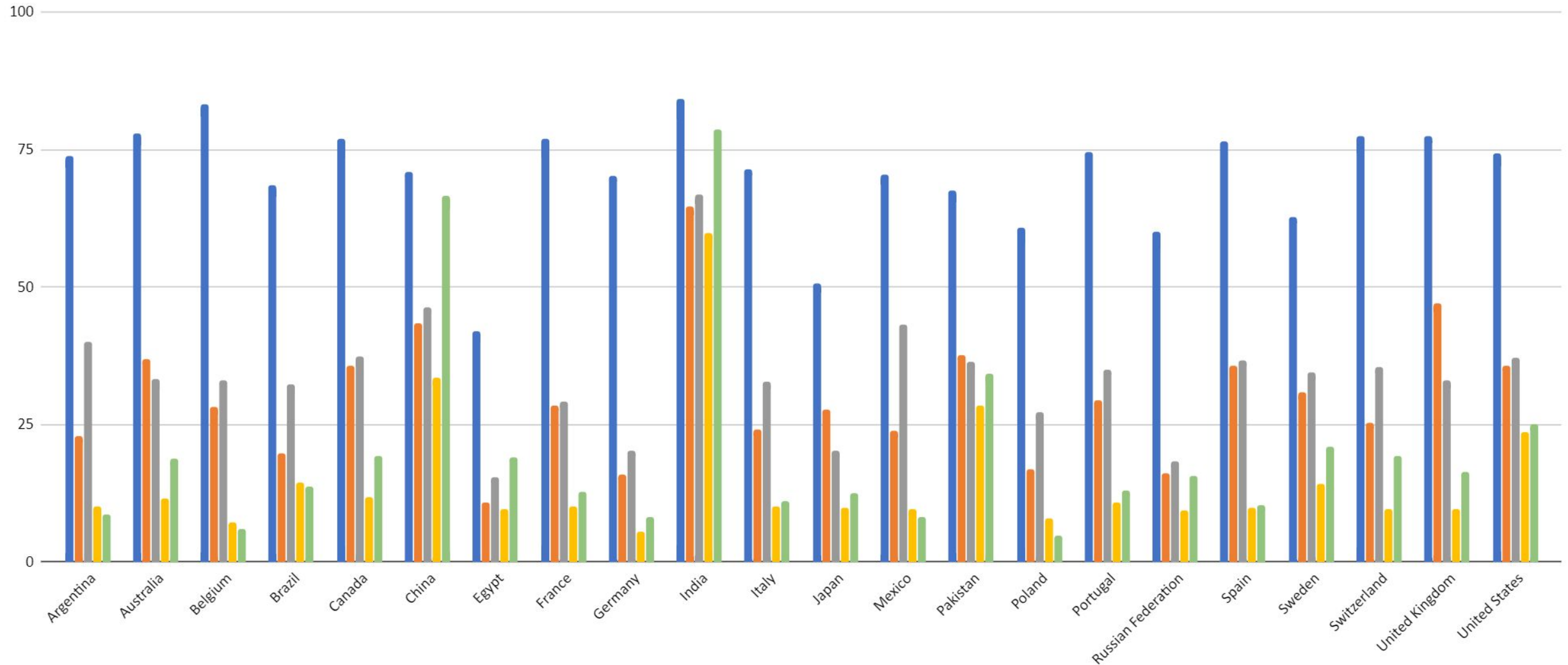
| Source | OR (95% CI) |
|---|---|
| Trust = Yes | |
| Argentina | 4.81 [3.54; 6.54] |
| Australia | 4.34 [3.39; 5.55] |
| Belgium | 6.17 [4.18; 9.11] |
| Brazil | 3.34 [2.62; 4.27] |
| Canada | 4.87 [4.16; 5.71] |
| China | 3.22 [2.74; 3.77] |
| Egypt | 2.59 [1.95; 3.46] |
| France | 4.32 [3.15; 5.91] |
| Germany | 6.78 [5.05; 9.12] |
| India | 0.73 [0.47; 1.14] |
| Italy | 4.36 [3.37; 5.64] |
| Japan | 2.80 [2.45; 3.21] |
| Mexico | 4.53 [3.51; 5.85] |
| Pakistan | 2.06 [1.58; 2.70] |
| Poland | 5.00 [4.18; 5.98] |
| Portugal | 3.41 [2.84; 4.10] |
| Russian Federation | 3.68 [2.72; 4.97] |
| Spain | 4.29 [3.37; 5.47] |
| Sweden | 3.97 [2.94; 5.36] |
| Switzerland | 4.96 [3.03; 8.14] |
| United Kingdom | 4.43 [3.80; 5.15] |
| United States | 5.92 [4.82; 7.27] |
| Total | 3.85 [3.34; 4.44] |

Heterogeneity: $\chi^2_{21} = 174.02$ ($P < .01$), $I^2 = 88\%$

Willingness to donate: OR (95% CI)

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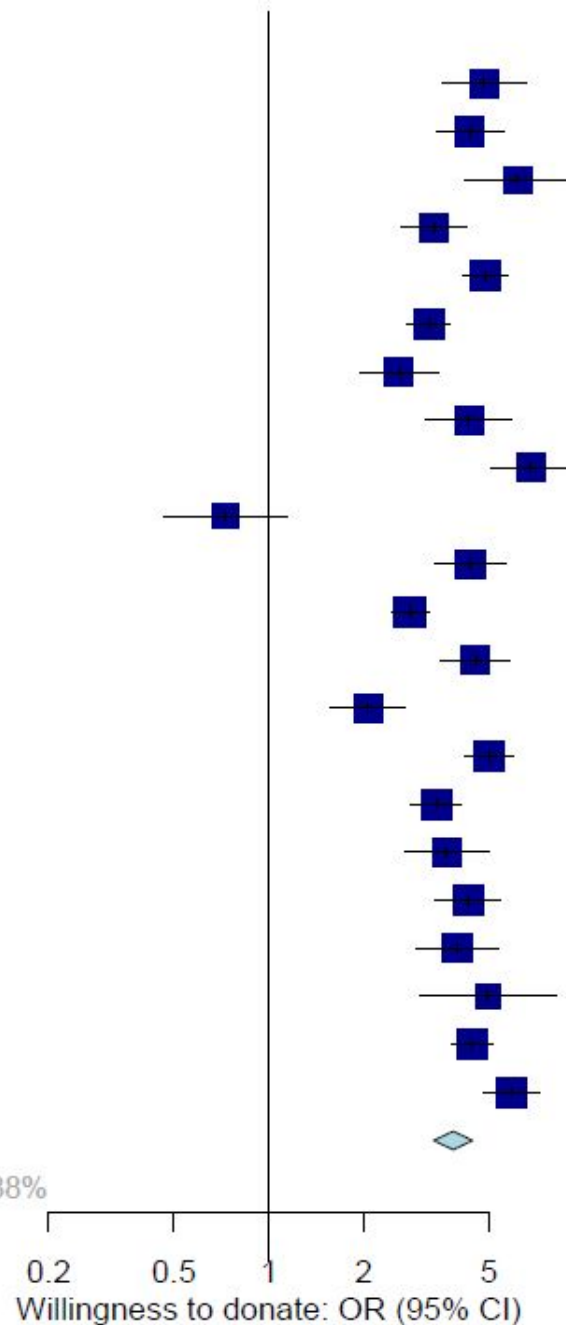
# Trust

## Analysis using UK/USA/Canada/Australia only

Human Genetics
https://doi.org/10.1007/s00439-019-02062-0

ORIGINAL INVESTIGATION

Check for updates

**Trust in genomic data sharing among members of the general public in the UK, USA, Canada and Australia**

Richard Milne[1,2] · Katherine I. Morley[3,4,5] · Heidi Howard[6] · Emilia Niemiec[6] · Dianne Nicol[7] · Christine Critchley[7,8] · Barbara Prainsack[9,10] · Danya Vears[11,12,13,14] · James Smith[15] · Claire Steed[15] · Paul Bevan[15] · Jerome Atutornu[1,16] · Lauren Farley[1] · Peter Goodhand[17] · Adrian Thorogood[18] · Erika Kleiderman[18] · Anna Middleton[1,19] · on behalf of the Participant Values Work Stream of the Global Alliance for Genomics and Health

# 'Trusting participants'

More likely to be

- under 50,
- male,
- with children,
- have personal experience of genetics
- hold religious beliefs
- be from the USA.

(USA, Canada, Australia, UK only)
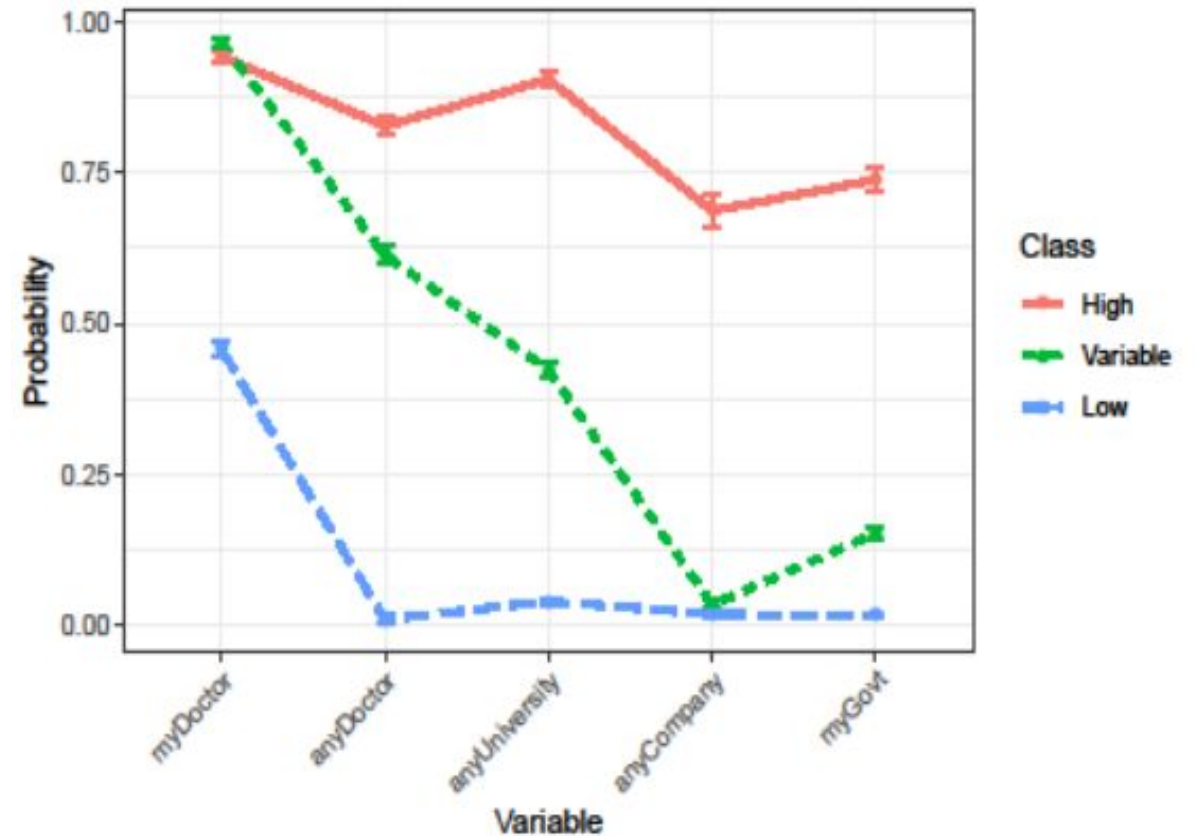
- Willingness to donate strongly associated with high trust group
- Compared with Low Trust group, High trust group less likely to be concerned about government, police, marketing and insurance uses of data
- Negative experiences with data access online most associated with high trust group
- High and variable trust groups most likely to be reassured by laws around donation
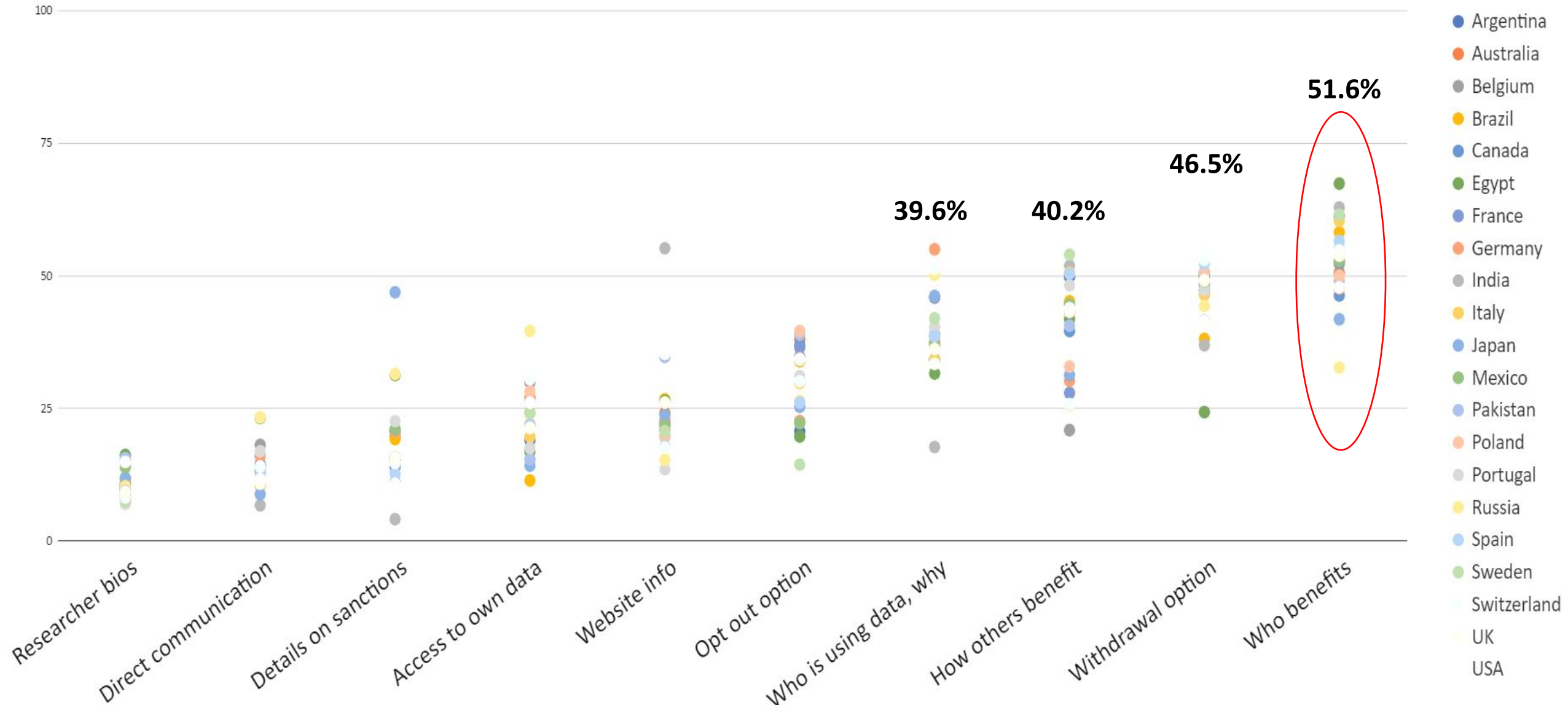
**What is being shared?**

**With whom?**

**Why?**

- Familiarity with genomics
- Is there is something special about genomic data?


- Who the recipient is
- Trust in the recipient


- Building trustworthiness

# Building trustworthy data

"What information would help you to trust the people asking you to donate DNA and/or medical information?"

Across 22 countries, there's variation in who is trusted with data but the **most trusted** is a person's **own doctor**
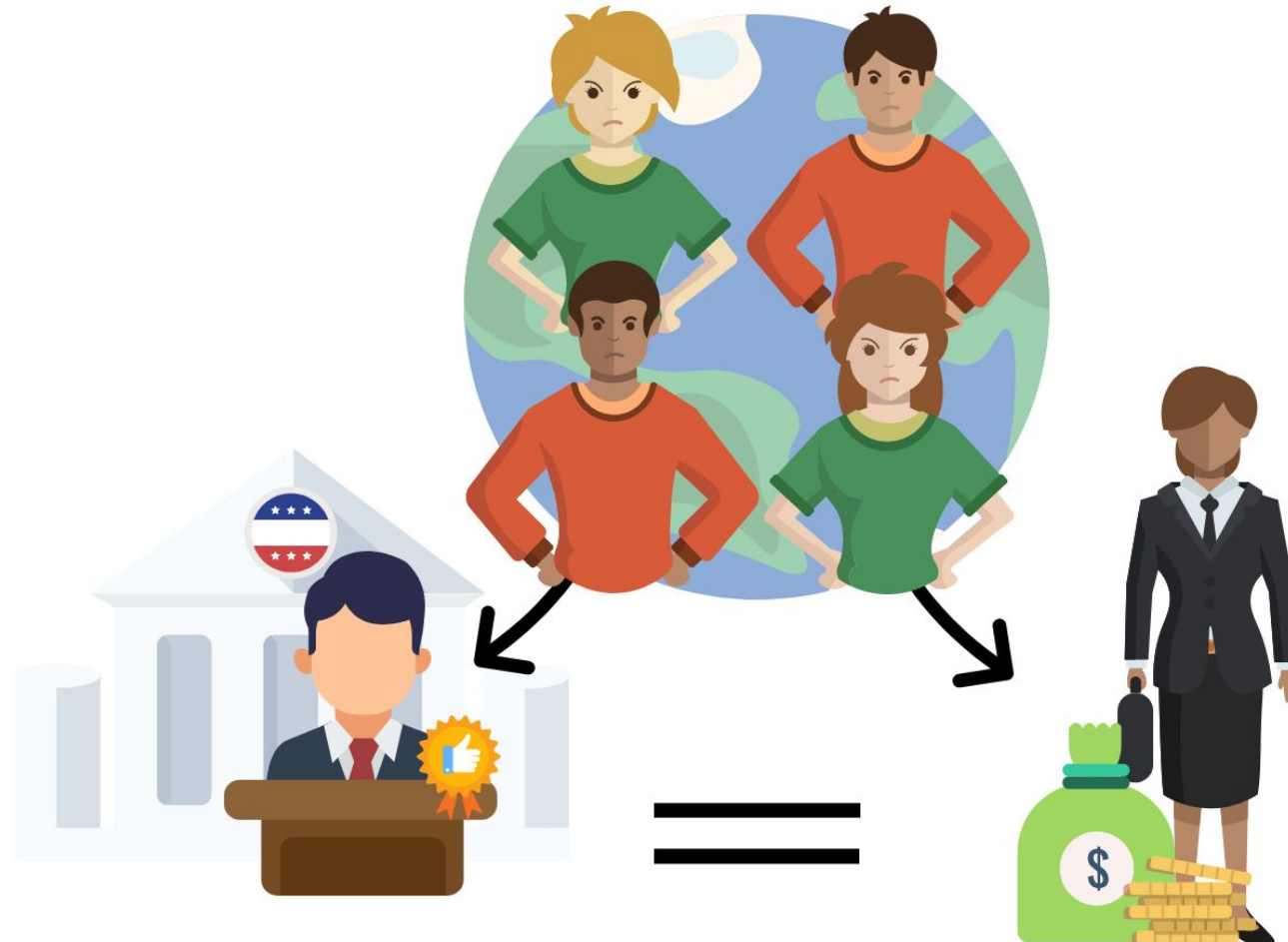
**Doctors** play a **gatekeeping role** in supporting the **development of large-scale data sharing initiatives**

Trust may be increased by **clarity** about **who** will use the data, **for what purpose**, **who will benefit** & **how**

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# Who benefits

Who does and who should benefit?

The 'social contract'

- Reciprocity
- Solidarity
- Altruism

April 2019

## A public dialogue on genomic medicine: time for a new social contract?

Final report

Ipsos MORI    Genomics england    sciencewise    UK Research and Innovation

Department of Health & Social Care

Guidance
**Creating the right framework to realise the benefits for patients and the NHS where data underpins innovation**

Published 15 July 2019

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"participants' preference was clearly for the widest possible public benefit to be felt by all, but they also acknowledged the value in research aiming to primarily benefit vulnerable groups within society"

Original Research Article

**Who benefits and how? Public expectations of public benefits from data-intensive health research**

**Mhairi Aitken, Carol Porteous, Emily Creamer and Sarah Cunningham-Burley**

Mhairi Aitken et al., "Who Benefits and How? Public Expectations of Public Benefits from Data-Intensive Health Research," *Big Data & Society* 5, no. 2 (July 1, 2018): 2053951718816724, https://doi.org/10.1177/2053951718816724.

# Conclusions

Attitudes towards genomic data sharing are relatively consistent across the sample
- But DNA/genomes are not the same for all people

What shapes attitudes towards sharing genomic data?
- familiarity
- how we see DNA
- who is using data
- their trust in these users

What influences trust?
- clear and defined benefits
- ethical and legal protections

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Key challenges relate to:

- Discussing the purpose of partnership between medicine, non-profit & for-profit researchers and industries
- Making clear who benefits and how from the use of data
    - and who is excluded and why
- And who is making these decisions
- Addressing the role of local, trusted actors (own doctor) in global genomic and data-driven medicine

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# "Data are cultural artefacts"



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)

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# **Next steps**

- Implications for policy and genomic medicine

- Understanding data in context

- Learning from/for genomics

# SPACE (Stakeholder Perspectives on social and ethical Aspects of digital Cognitive Evaluation)
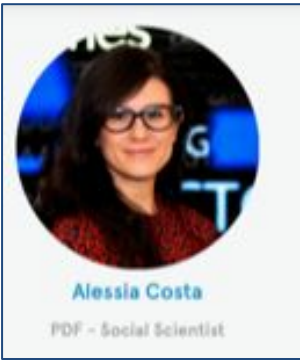
- Work with scientists/clinicians/tech developers

- Work with older adults
  - How do older adults relate to data in everyday life
  - How does their everyday life become data?
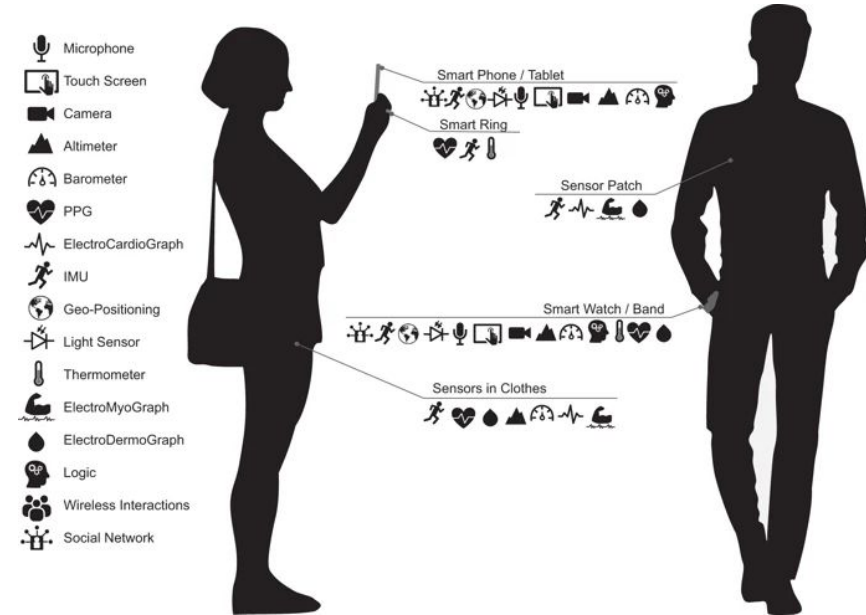
# It couldn't have happened without……

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**Project Management**
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**Data Analysis**
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**Sanger Web team**
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Mandarin: Yali Cong
French: Heidi C Howard
Australian: Christine Critchley, Dianne Nicol
S. African: Shelley Macaulay, Tasha Wainstein, Amanda Krause
German: Barbara Prainsack, Torsten Heinemann
Urdu (Pakistan and India): Q Annie Hassan
Icelandic: Vígdis Stefansdottir
Italian: Deborah Mascalzoni, Virginia Romano, Maria Gnadl
Japanese: Jusaku Minari
Polish: Emilia Niemiec
Portuguese: Álvaro Mendes, Cláudia de Freitas
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Nadia Kovalevskaya
Spanish: Anne West, Maria Cerezo
Swedish: Heidi C Howard, Josephine Fernow
Ghana: Jerome Atutornu

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Chiara Garattini
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Christoph Schickhardt
Danya Vears
Cris Woolston

**REWS**
Adrian Thorogood
Bartha Knoppers

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- 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,

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