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SOCIETY+
ETHICS
RESEARCH



“GenomEthics”

Prof Anna Middleton

Head of Society and Ethics Research Group

We must bridge the gap between technology and policymaking. Our future depends on it



nature

International weekly journal of science

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NATURE | COLUMN: WORLD VIEW



Integration of social science into research is crucial

Social scientists must be allowed a full, collaborative role if researchers are to understand and engage with issues that concern the public, says

[Ana Viseu](#).

Overview

- Principles of Bioethics
- Current ethical issues
 - What to do with incidental findings from research?
 - Genomic data sharing – privacy and concerns
 - Embryo editing
 - ABC versus St Georges NHS trust
- Policy and governance creation

Principles of Bioethics

- Autonomy (individual choice)
 - Non-maleficence (do no harm)
 - Beneficence (do good)
 - Justice (apply equitably)
-
- Beauchamp and Childress from 1982 -



Examples

What to do with incidental findings from research?
(autonomy, justice)

Genomic data sharing – privacy and concerns
(non-maleficence)

Embryo editing (beneficence versus non-maleficence)

ABC versus St Georges NHS Trust (whose autonomy?)

ABC Versus St George's NHS trust

NEWS

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Huntington's disease: Woman who inherited gene sues NHS

**Fergus Walsh**

Medical correspondent

@BBCFergusWalsh

🕒 18 November 2019



Share



ABC vs St Georges

- 2009 genetic testing confirmed that the patient had HD.
- Insisted he did not want his daughters, one of whom was pregnant, to be informed about HD status
- Daughter accidentally informed of HD diagnosis after birth of her son
- Daughter tested and found to carry HD gene, decided to take legal action against medical team who chose not to disclose diagnosis



The image shows a podcast cover for BBC Radio 4's 'Inside the Ethics Committee'. The top section features the BBC Radio 4 logo and the title 'Inside the Ethics Committee' in white text on a dark blue background. Below the title is a navigation bar with links for 'Home', 'Episodes', 'Clips', 'Podcast', and 'Contact Us'. The main visual is a black and white photograph of a family (a pregnant woman, a man, and a child) walking on a beach at sunset, with their silhouettes reflected in the wet sand. A 'Listen now' button with a speaker icon is overlaid on the bottom left of the image. Below the image, the title 'Sharing Genetic Information' is displayed in white, followed by 'Series 12 Episode 3 of 3'. A short description of the episode is provided: 'Medical information is personal, and DNA perhaps the most personal of all. But what about a life-threatening genetic fault your family might share? Do they have a right to know?'. The episode is marked as 'Available now' and has a duration of '43 minutes'.

Sharing Genetic Information
Series 12 Episode 3 of 3

Medical information is personal, and DNA perhaps the most personal of all. But what about a life-threatening genetic fault your family might share? Do they have a right to know?

Available now
⌚ 43 minutes

<https://www.bbc.co.uk/programmes/b07nrxd4>

the grandmother

2008



the mother

2012



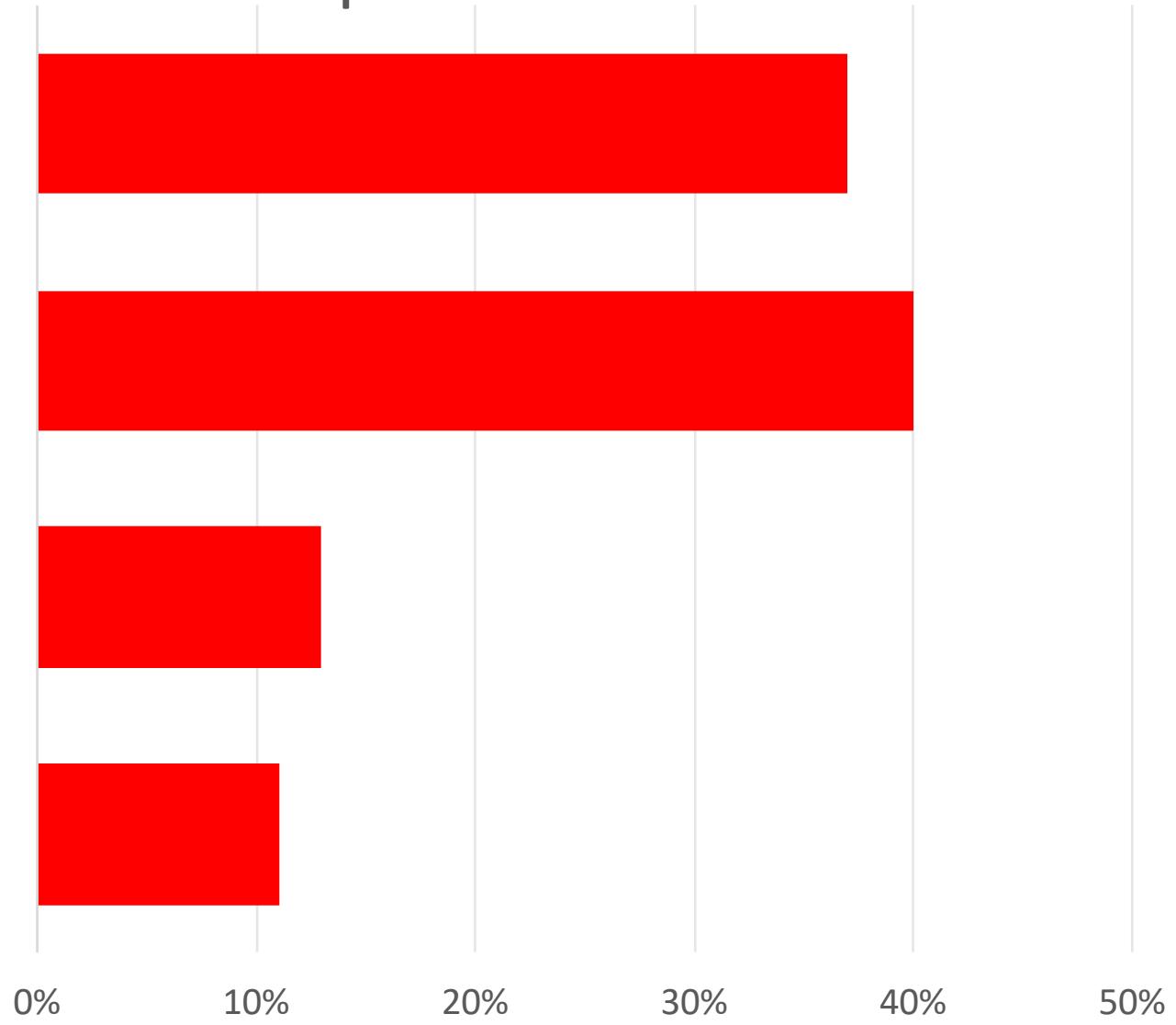
Should health professionals have a legal duty to share genetic information with a patient's relatives?

Yes, but only if there is agreement from an affected relative

Yes, even if there is no agreement from an affected relative

No

I don't know



Disclosing genetic and other shared information

- In the UK, confidentiality is not absolute
- Disclosure can occur if
 - the patient refuses to inform others,
 - an identifiable person (relative) is at serious risk of harm
 - such harm might be prevented by disclosure. (GMC)
- In *ABC*, Court of Appeal ruled doctors may have legal duty of care to inform relatives of risk
- Clinicians need to weigh the potential harms of disclosure against the potential benefits

Embryo editing



CNN



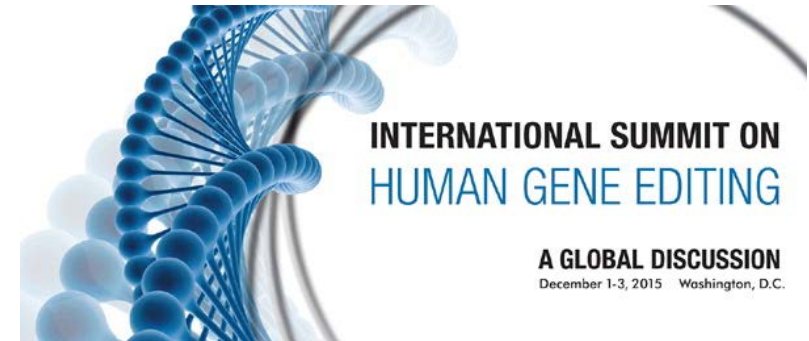


CHINA 24

...s, download CGTN APP, and follow @CGTNOfficial on Twitter, @CGTN on Facebook, YouTube, Weibo, Wechat, Tumblr

International Summit on Human Gene Editing

- “It would be **irresponsible** to proceed with any clinical use of germline editing unless and until
 - (i) **the relevant safety and efficacy issues have been resolved ... and**
 - (ii) **there is broad societal consensus” about the appropriateness of the proposed application**



Next steps

- How to ensure the effectiveness of international ethical and scientific positions?
- What to do in cases of violation of these norms (on the part of scientists, publishers, funders, governments)?
- Balancing concerns and hopes of different publics – what counts as ‘societal consensus’

What to do with incidental findings from research?



- ✓ Questions about you
- ➔ Sharing of Pertinent Findings
 - Sharing of Incidental Findings
 - Categorizing Incidental Findings
 - Relations with Risk
 - Raw data
 - Duty of Genomic Researchers
 - Filter of Genomic Information
 - Consent for genomic research
 - Last few questions about you

Sharing of Pertinent Findings

- Should Pertinent Findings from genome studies be made available to research participants?
 - Research participants should be able to receive pertinent findings if they want them
 - I don't think pertinent findings from research projects should be available
 - I don't know

« Previous

Next »



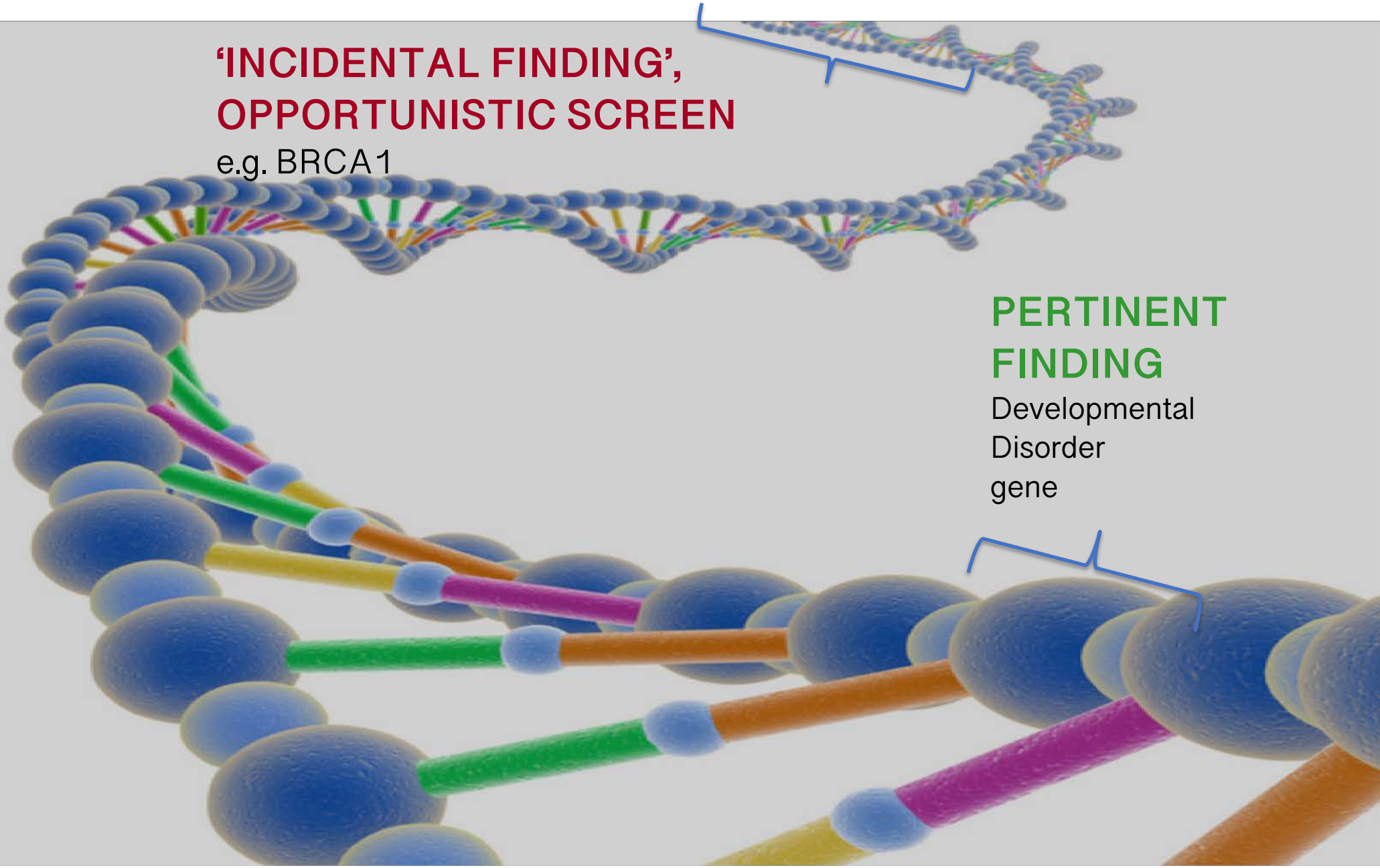
4
News

**‘INCIDENTAL FINDING’,
OPPORTUNISTIC SCREEN**

e.g. BRCA1

**PERTINENT
FINDING**

Developmental
Disorder
gene



Positions on IFs

Techno-enthusiasts

- Return all potentially actionable results

Genomic libertarians

- Let people have what they want

Genomic Fabians

- Gradual introduction of return and evaluation

Luddites

- No to wider use of genomic tests

Clarke, A. J. (2014). Managing the ethical challenges of next-generation sequencing in genomic medicine. *British medical bulletin*, 111(1), 17-30.



Drawn from Life by an Officer

THE LEADER OF THE LUDDITES

Pub. May 1812 by Messrs. Walker and Knight, Sweeting Alley, Royal Exchange.

PCGF2

G→A Chr17:36,895,854



Public = 4961



Genomic
researchers = 607



Genetic health
professionals = 533

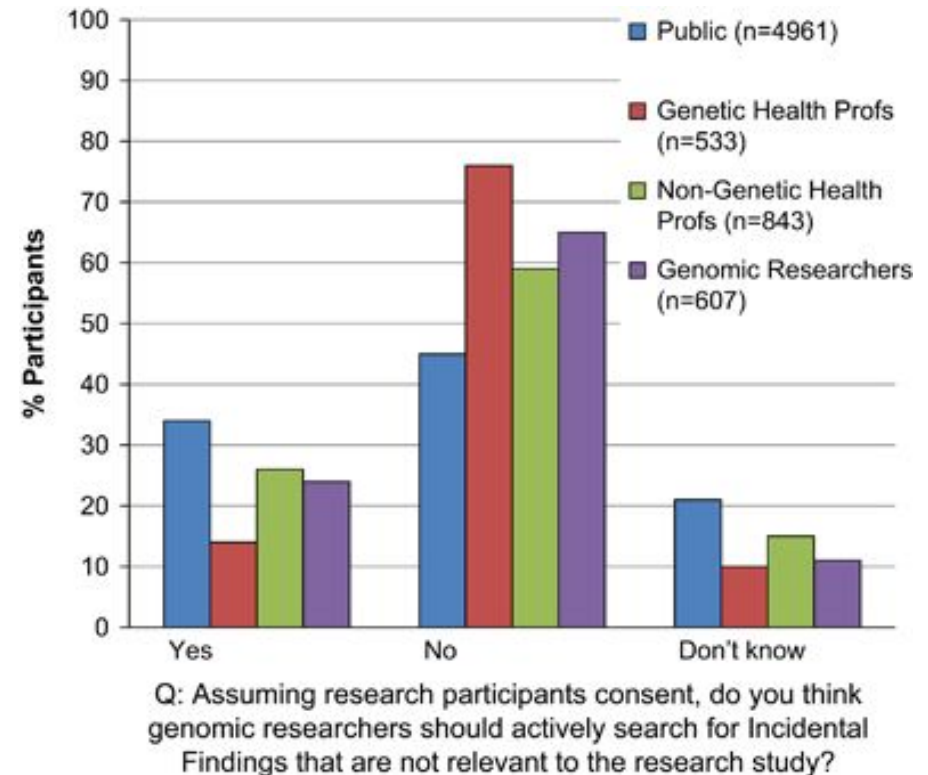
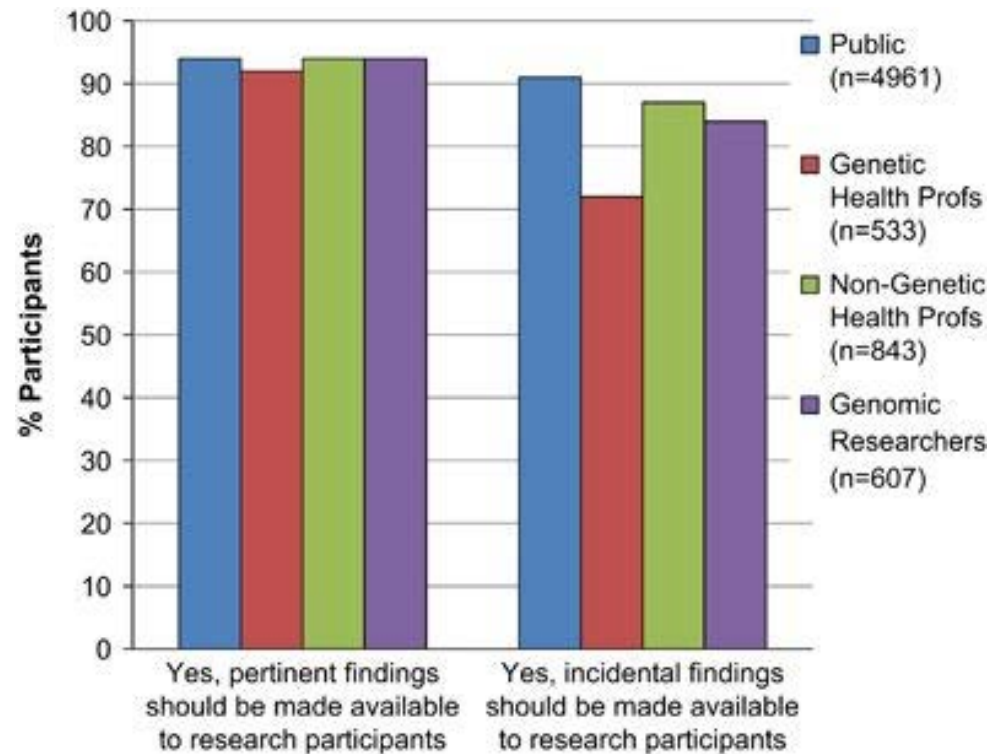


Other health
professionals = 843



Q: What influences attitudes the most?

A: Our professional background rather than the country we are from



Three key messages

1. On the whole, all stakeholders would be interested in receiving IFs
2. Actionability is important
3. Genetic health professionals are more conservative

Original 100,000 Genomes list (returned with consent)

- Hereditary non-polyposis colorectal cancer (HNPCC)/ Lynch syndrome
 - (genes: mismatch repair genes MLH1, MSH2, MSH6, PMS2) - adult onset**
- Familial adenomatous polyposis (FAP) (gene: APC)
- MYH-associated polyposis (MAP) (gene: MutYH)
- Hereditary, breast and ovarian cancer (genes: BRCA1 and BRCA2) - adult onset
- Von Hippel-Lindau syndrome (gene: VHL) - child and adult onset
- Multiple endocrine neoplasia type 1 (gene: MEN1) - child and adult onset
- Multiple endocrine neoplasia type 2 (gene: RET) - child and adult onset
- Familial medullary thyroid cancer (FMTC) (genes: RET and NTRK1) – child and adult onset
- Retinoblastoma (gene: RB1) – child onset
- Familial hypercholesterolaemia gene: LDLR- child onset- and also APOB and PCSK9 - child and adult onset

Genomic Data Sharing



Premise

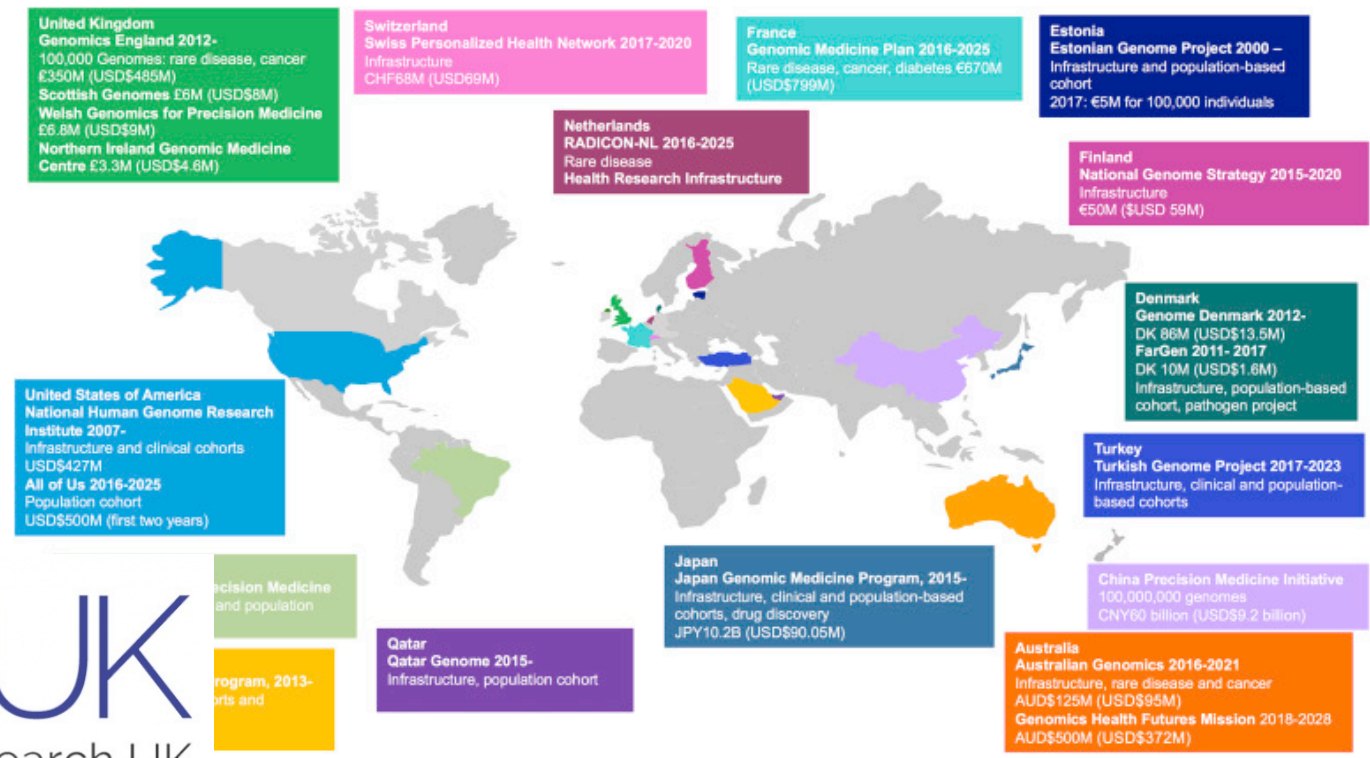
- Data sharing endeavors rely on people willing for their donated data to be shared (i.e. 'data donors')
- Even if we aren't personally donating, we'll be related to someone who is, i.e. the ethical issues linked to data sharing are relevant to global publics
- Exemplary practice involves listening to what potential donors want, believe and fear and consider policy implications
- This work is part of GA4GH strategic roadmap

Diversity in genomic data

- Universal Declaration of Human Rights. guarantees the rights of every individual in the world “to share in scientific advancement and its benefits”
- Existing genomic datasets are dominated by white European ancestry populations
- Increases the likelihood non-EA individuals will receive inconclusive or, erroneous interpretations of genomic variants.



Genomic and health data sharing



HDRUK
 Health Data Research UK



Stark et al. (2019)

Medical Information about Insects

Heart Disease DNA Information

Cancer DNA Information

Database on health information

Database on Environmental
Giles Medical Information about Insects

Heart Disease DNA Information
DNA
0110010110101

Cancer DNA Information

DNA Information Healthy People
GCTAC

Database on Environmental

Gene Storage Database
Database on health information

Gene Storage Database
Cancer DNA Information

DNA Information Healthy People
DNA
Book II Cancer DNA Information
Medical Information about Insects

Medical Information about Insects
Database on health information
The word
Heart Disease DNA Information

Index of gene storage information
Gene Storage Database
Heart Disease DNA Information

Medical Information about Insects

Database on health information

Database on Environmental

Gene Storage Database

あなたのDNA、あなたの意見



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これることができます。

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 »

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

Pitt erfðæfni, 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

آپ کا ڈی این اے، آپ کا کہنا

خوش آمدید



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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۔ فلٹوں کو دیکھنے اور چند سوالات کا جواب دیجیے۔ فلٹیں آپ کو سب جان کاری دے گی۔ اپنے نام سے ان الفاظ کو دیکھیں جن کے کچے این سے ہے اور آپ کو ایک فہرست نظر آئے گی۔

سروے کوئی بھی اور سمجھی بھر سکتے ہیں، لہذا آگے بڑھیں۔ آپ کی شرکت مکمل طور پر گمنام ہے اور ہم دیکھ کر جینوم کیسے، کیجیبرج، برطانیہ میں تمام جو بات محفوظ رکھیں گے۔

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

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

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

reset answers and start again

الحمض النووي الخاص بك

- ✓ اہلاً وسهلاً
- ✓ البصمة الإلكترونية
- ✓ إمكانية الوصول للتبليغات من قبل آخرين
- ✓ إمكانية الوصول من قبل الأطباء المختصين
- ✓ إمكانية الوصول من قبل الباحثين غير علمي الريح
- ✓ إمكانية الوصول من قبل الباحثين الهادفين للريح
- الأضرار المتوقعة
 - المتوقع للمعلومات
 - الثقة
 - آخر الأسئلة المتعلقة بك
 - إرسال إجاباتك



films can be skipped, paused or watched again

الأضرار المتوقعة

لتفرض أنه قد أصطبت الخيار للتعرج بالحمض النووي الخاص بك و بمعلوماتك الطبية بحيث تكون مجهول الهوية وذلك من أجل أن تستخدم من قبل أي مما يلي:

- الأطباء المختصين (مثال: من أجل مساعدتهم في تشخيص الحالات المرضية و القيام بنشر الأبحاث الطبية)
- الباحثين الغير علمي الريح (مثال: من أجل القيام بأبحاث طبية و أيضا للحصول على تمويل جديد)
- الشركات الهادفة للريح (مثال: من أجل تطوير الأدوية والحصول على مال من وراء ذلك)

ضمن السيلزبيوعات الثلاثة المذكورة أعلاه، لتفرض أيضا أن هناك فرصة بأن يتم التعرف عليك شخصيا من قبل أحد الباحثين القادرين على الوصول الي قاعدة بياناتك. نريد معرفة ماذا تعتقد ...

● إن أقوم بالتعرج بمعلومات عن الحمض النووي الخاص بي و معلوماتي الطبية.

● سوف أقوم بالتعرج بمعلومات عن الحمض النووي الخاص بي و معلوماتي الطبية لئتم استخدامها من قبل ... (أكثر من الاجابات التالية بالضغط على المربع)

الأطباء المختصين

إن أقوم بالتعرج	سأقبل بنسبة 1% أن يتم التعرف علي	سأقبل بنسبة 10% أن يتم التعرف علي	سأقبل بنسبة 50% أن يتم التعرف علي	سأقبل بنسبة 90% أن يتم التعرف علي	لا اظن
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الباحثون الغير علمي الريح

إن أقوم بالتعرج	سأقبل بنسبة 1% أن يتم التعرف علي	سأقبل بنسبة 10% أن يتم التعرف علي	سأقبل بنسبة 50% أن يتم التعرف علي	سأقبل بنسبة 90% أن يتم التعرف علي	لا اظن
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الباحثون الهادفون للريح

إن أقوم بالتعرج	سأقبل بنسبة 1% أن يتم التعرف علي	سأقبل بنسبة 10% أن يتم التعرف علي	سأقبل بنسبة 50% أن يتم التعرف علي	سأقبل بنسبة 90% أن يتم التعرف علي	لا اظن
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تطبيقات

«الذاتي»

«السابق»

أكلنا

Global public survey

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



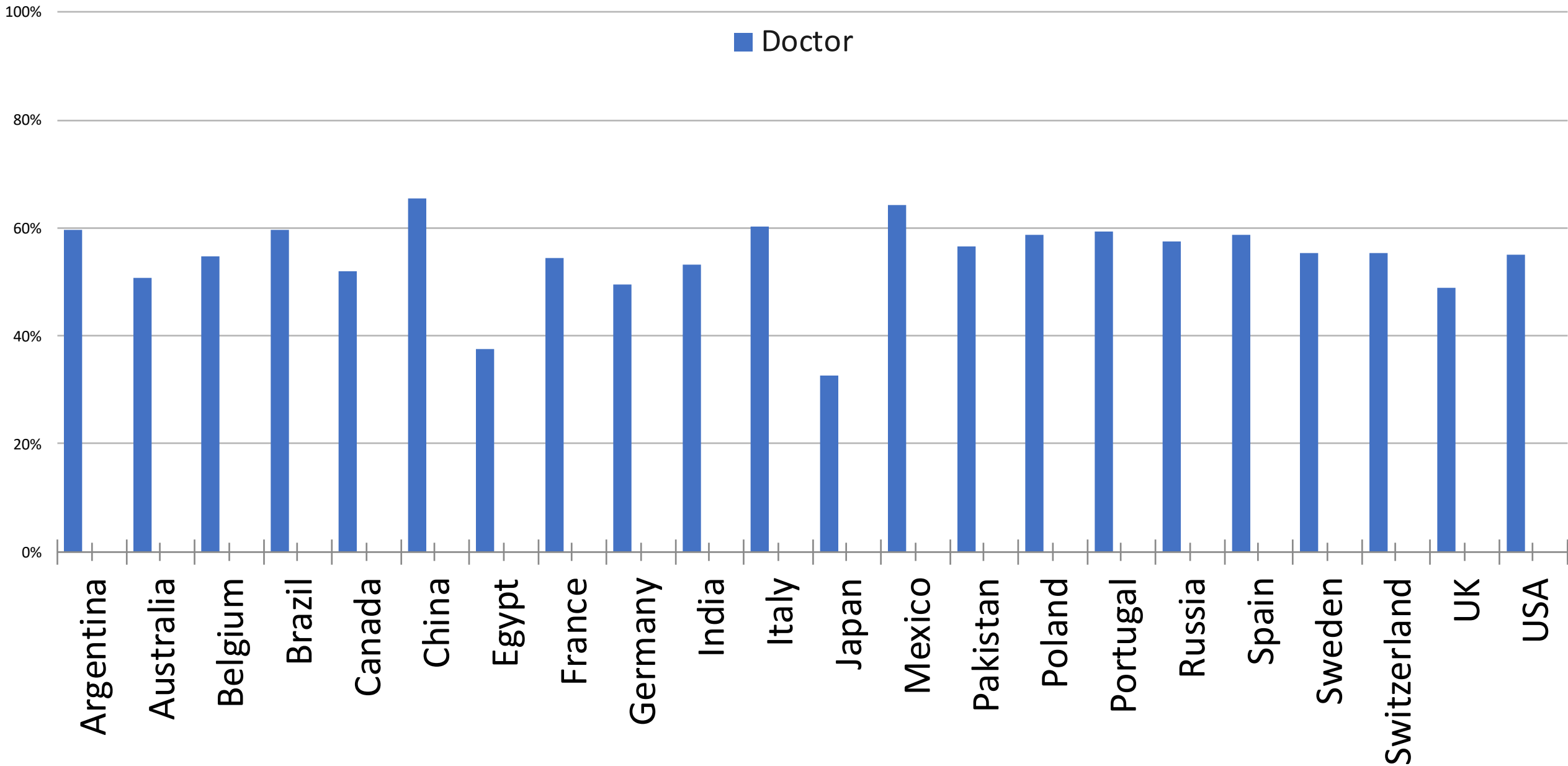
Currently available
Coming soon

Willingness to donate one's
genomic data

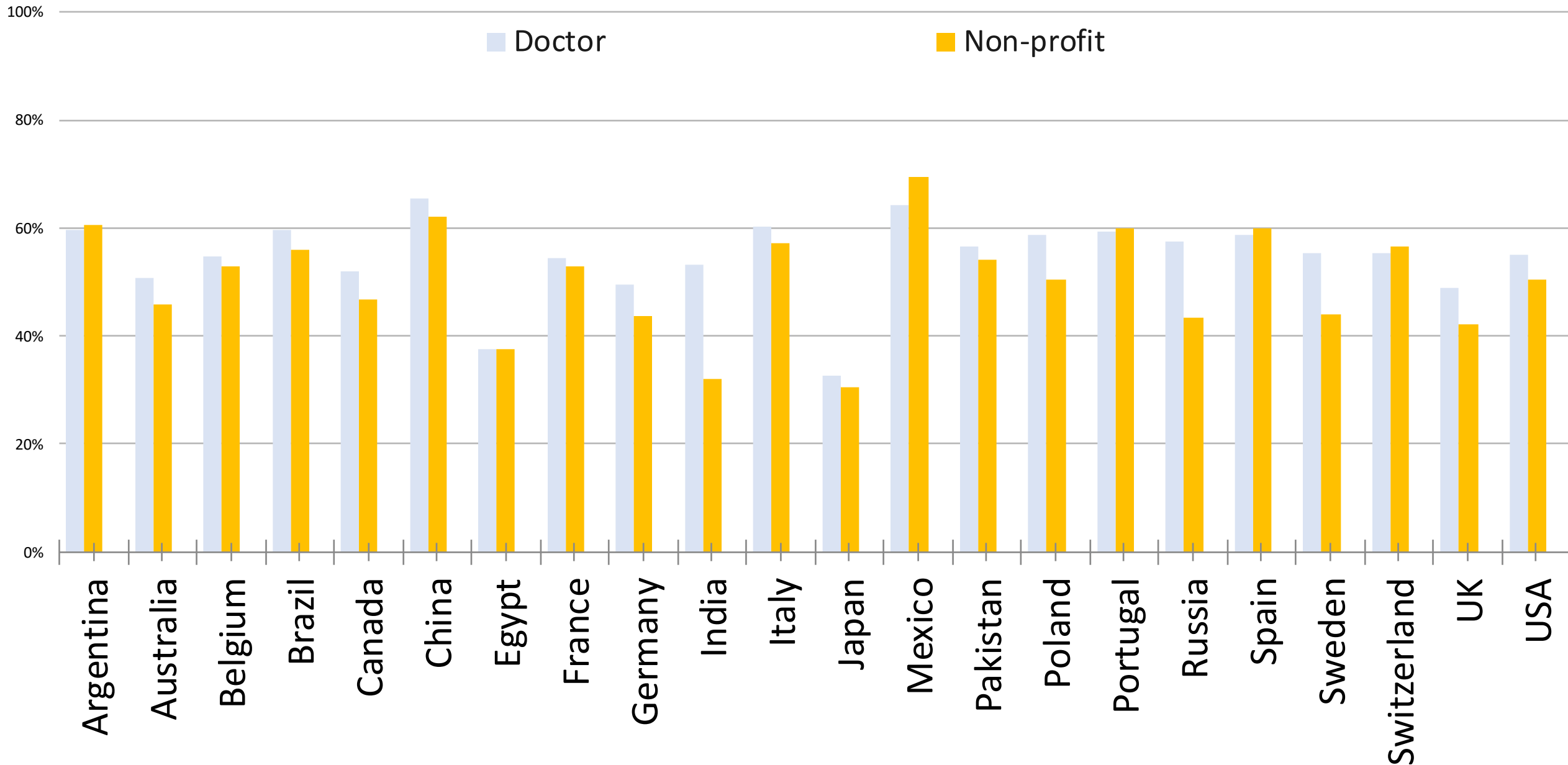
Would you donate your anonymous DNA information and medical information for use by:

- Medical doctors
- Non-profit researchers
- For-profit researchers

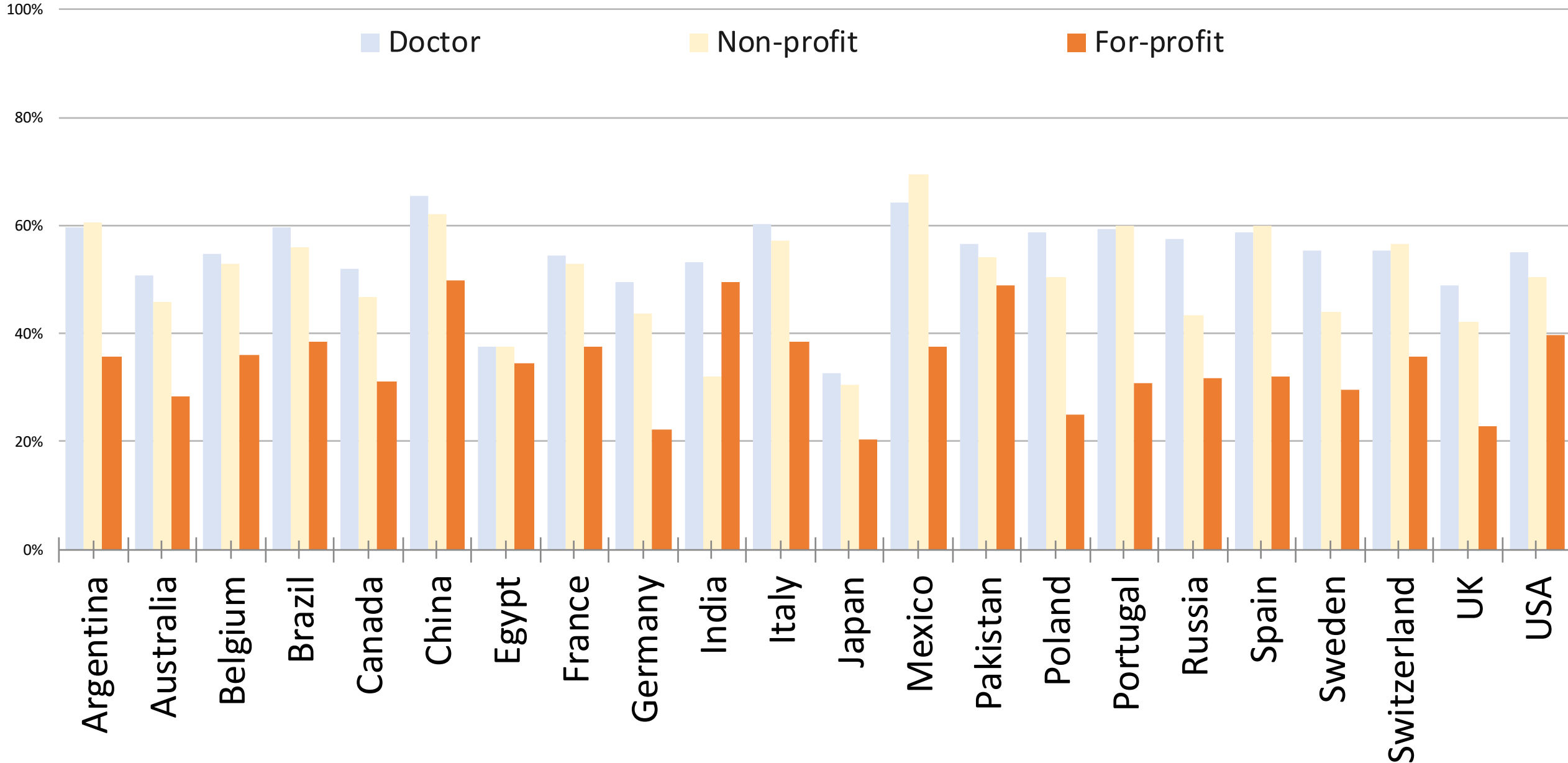
Willingness to donate DNA and health information



Willingness to donate DNA and health information



Willingness to donate DNA and health information



Across 22 countries, **global publics** are more comfortable with their data being used by **doctors** than **for-profit companies**



What affects willingness?

- Who the recipient is
- Familiarity with genomics
- Perception that there is something special about genomic data that warrants donation
- Trust in the recipient
- Perceived harms from re-identification
- Ability to receive raw data back
- Reassurance of legal protections in place

Trust and Data Sharing are thought to go hand in hand

Trinidad et al, 2010; Eckstein et al 2018; Lawler et al 2018; Shabani et al 2014,
Nuffield Council on Bioethics, 2015

Trust is thought to shape attitudes towards genomics and intention to participate in research

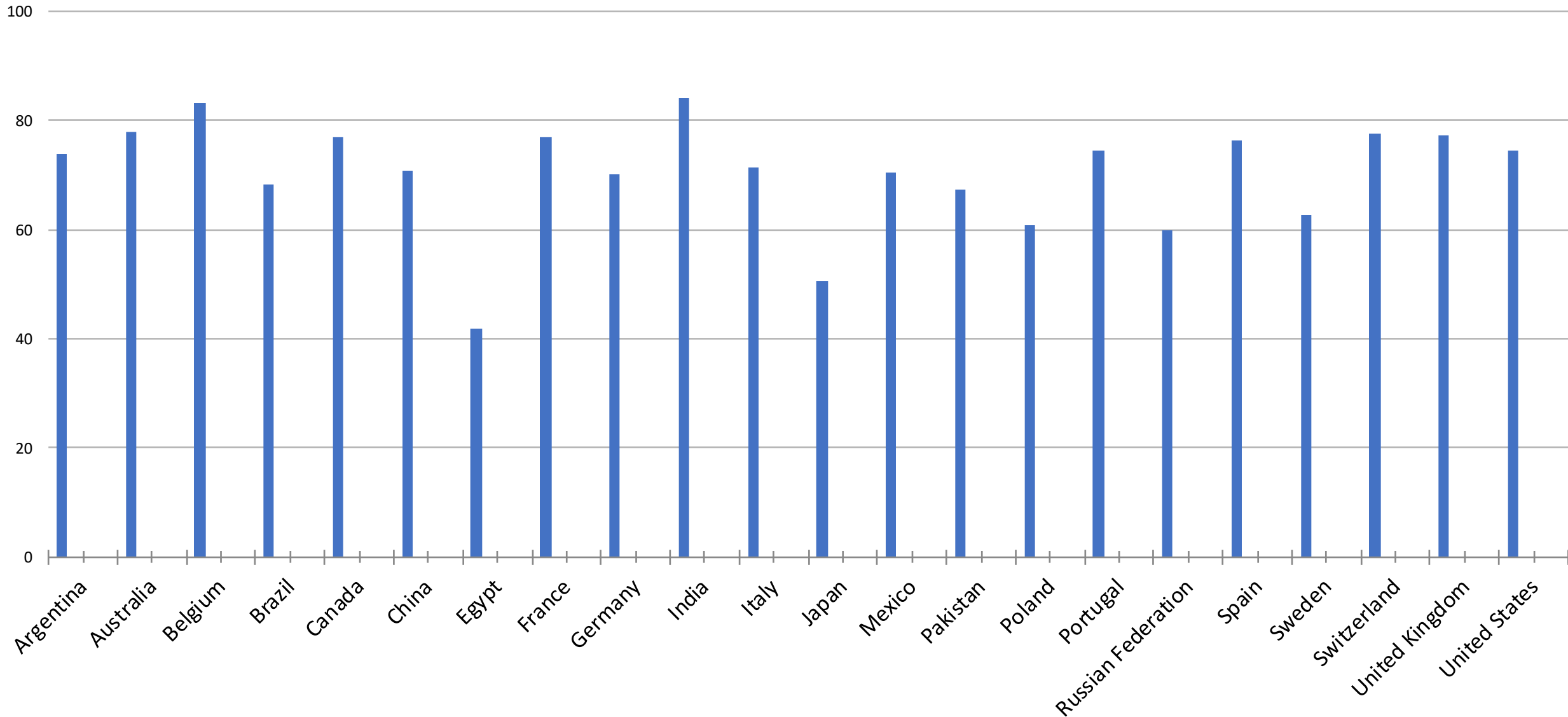
(Lipworth, et al 200; Critchley et al 2015; Nicol et al 2016; Lawler et al 2018)

Where trust is absent, the social license and mandate of researchers and clinicians to obtain and distribute data may be lost

(Carter et al 2015)

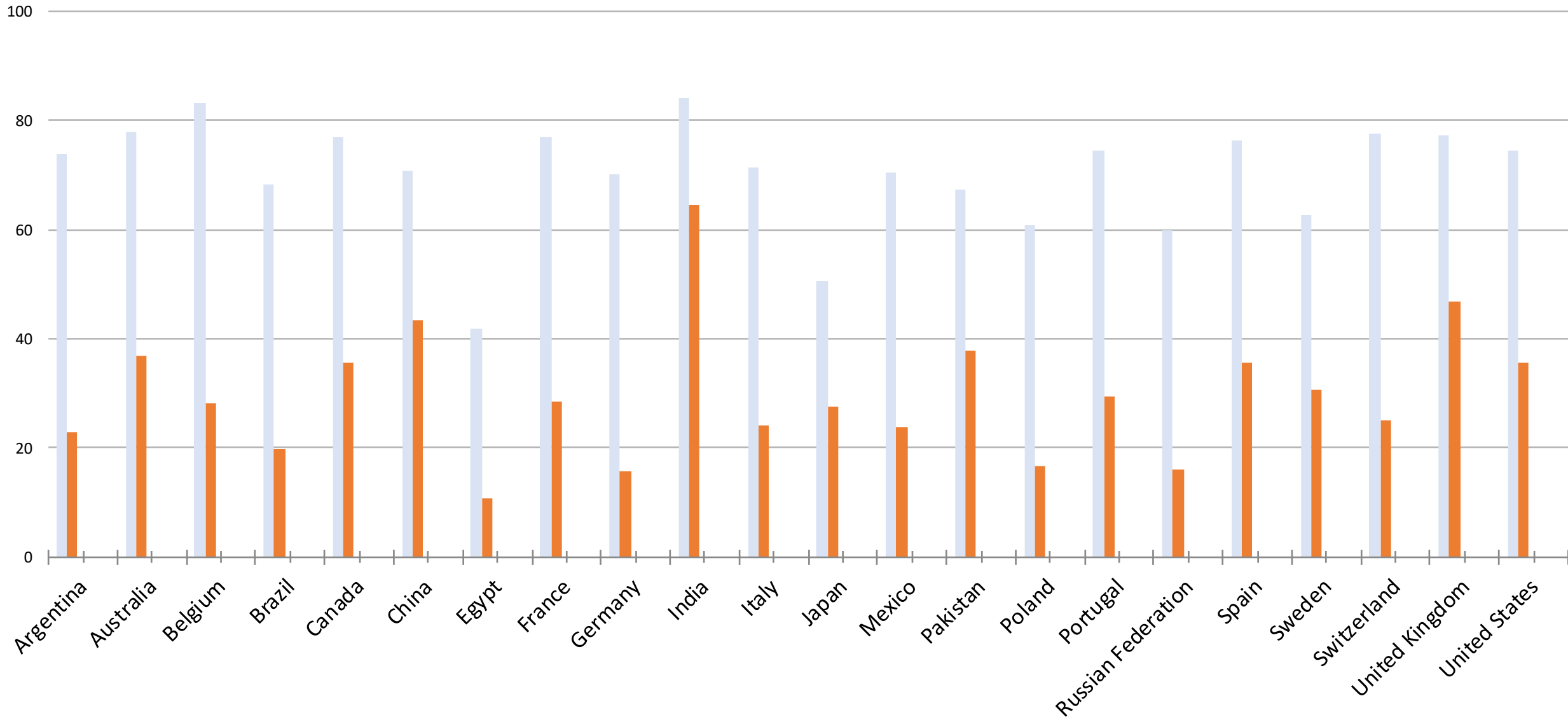
Total percentage trusting each organisation or individual with DNA and health information (global meta-analysis)

■ Own doctor



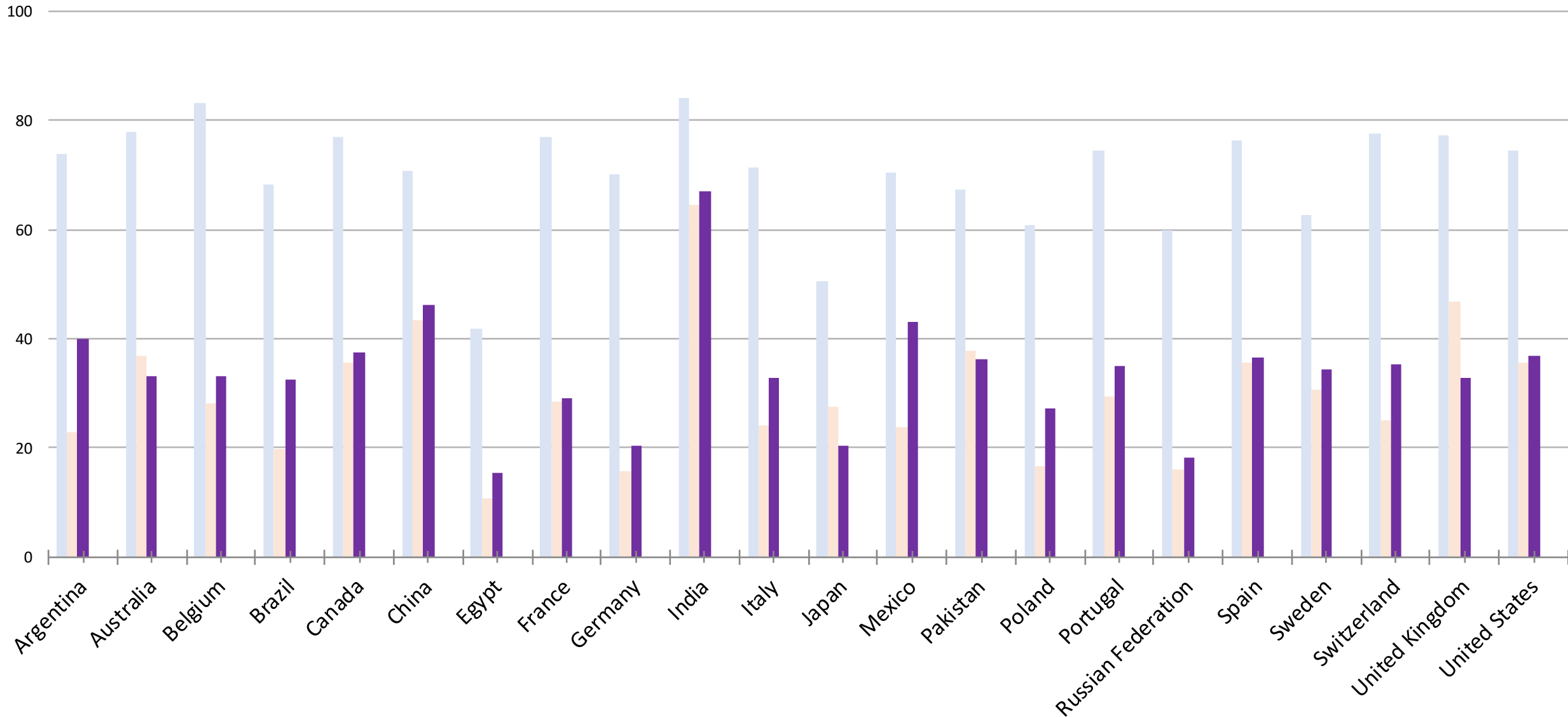
Total percentage trusting each organisation or individual with DNA and health information (global meta-analysis)

Own doctor Any doctor in country



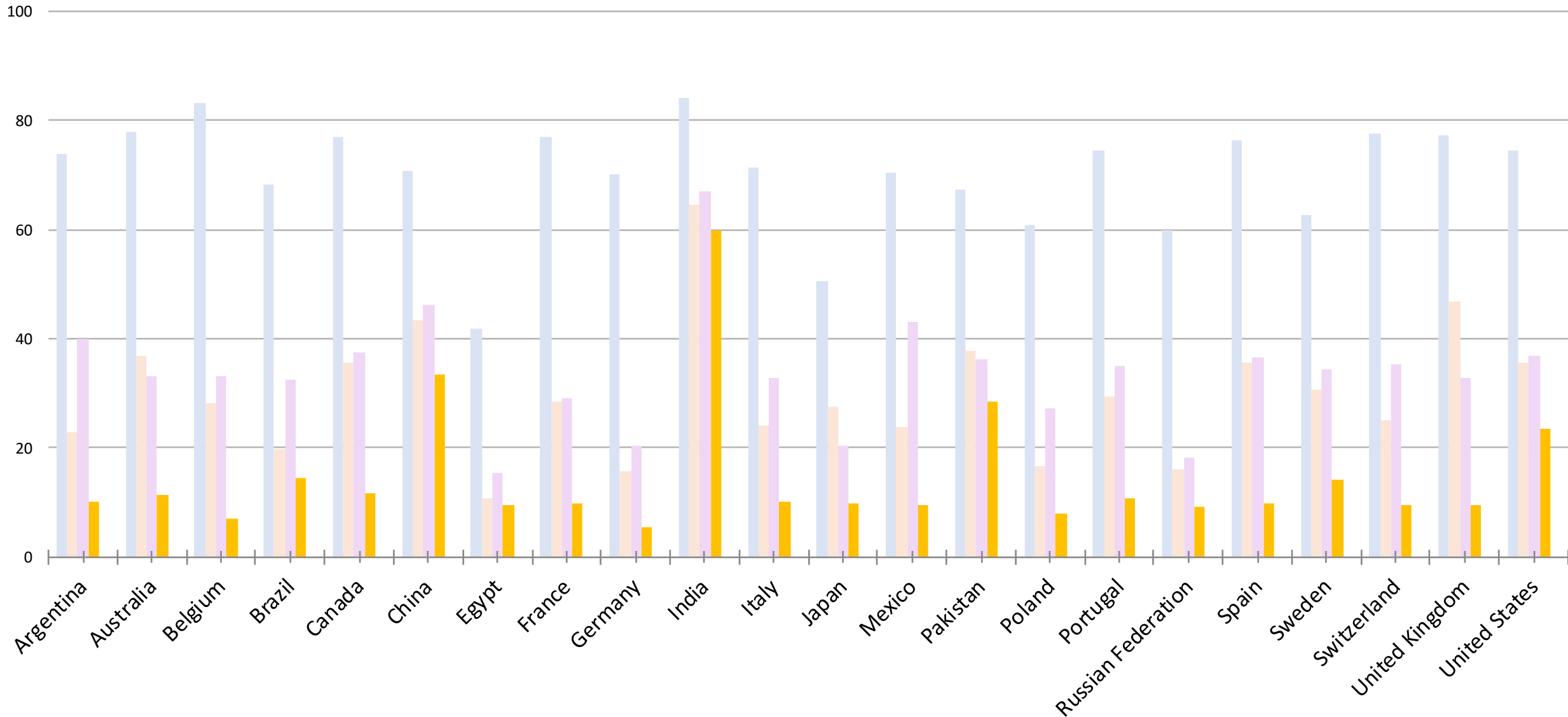
Total percentage trusting each organisation or individual with DNA and health information (global meta-analysis)

Own doctor Any doctor in country Non-profit researcher in my country

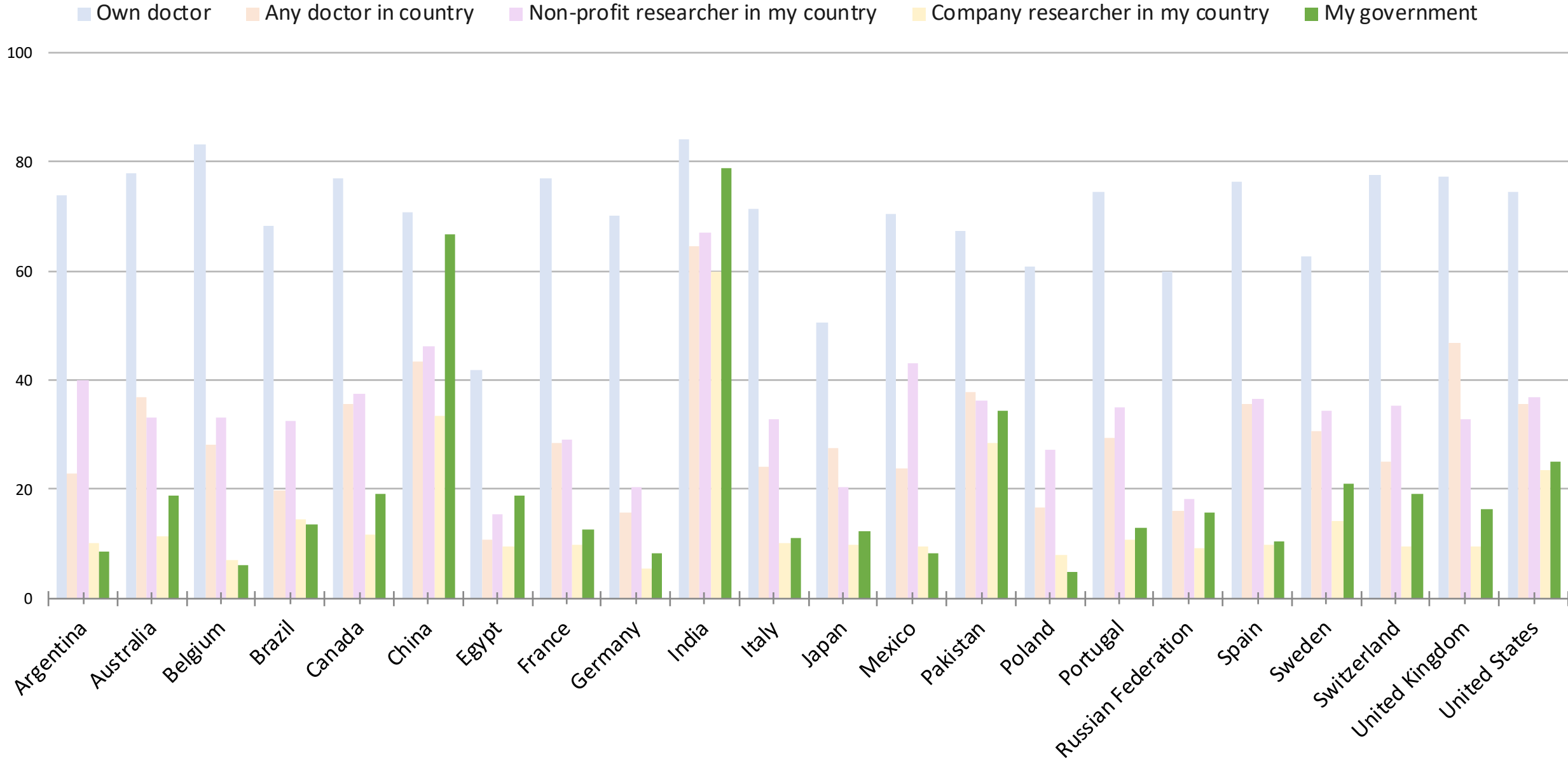


Total percentage trusting each organisation or individual with DNA and health information (global meta-analysis)

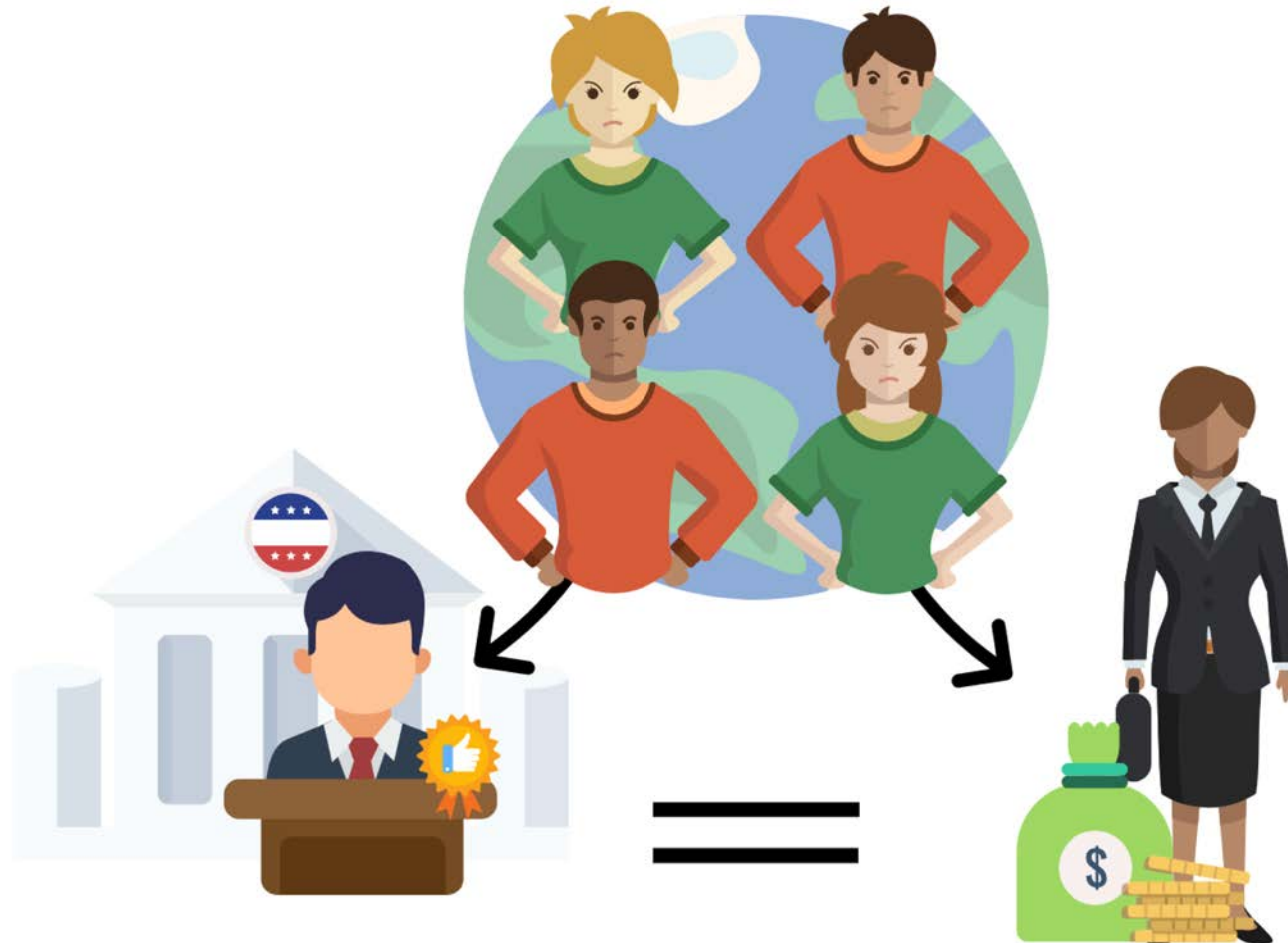
Own doctor Any doctor in country Non-profit researcher in my country Company researcher in my country



Total percentage trusting each organisation or individual with DNA and health information (global meta-analysis)



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**

Deeper Analysis using English speaking data only

English speakers were first to be recruited, and also each set of country data will be analysed by each collaborator

Profile of the most 'trusting participants'

- (USA, Canada, Australia, UK only)
 - More likely to be under 50, male, with children, hold religious beliefs, have personal experience of genetics and be from the USA. This profile are the most likely to be willing to donate data for any reason
 - Milne et al 2019 Trust paper

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 Howard⁶ · Emilia Niemiec⁶ · Dianne Nicol⁷ · Christine Critchley^{7,8} · Barbara Prainsack^{9,10} · Danya Vears^{11,12,13,14} · James Smith¹⁵ · Claire Steed¹⁵ · Paul Bevan¹⁵ · Jerome Atutornu^{1,16} · Lauren Farley¹ · Peter Goodhand¹⁷ · Adrian Thorogood¹⁸ · Erika Kleiderman¹⁸ · Anna Middleton^{1,19}  · on behalf of the Participant Values Work Stream of the Global Alliance for Genomics and Health

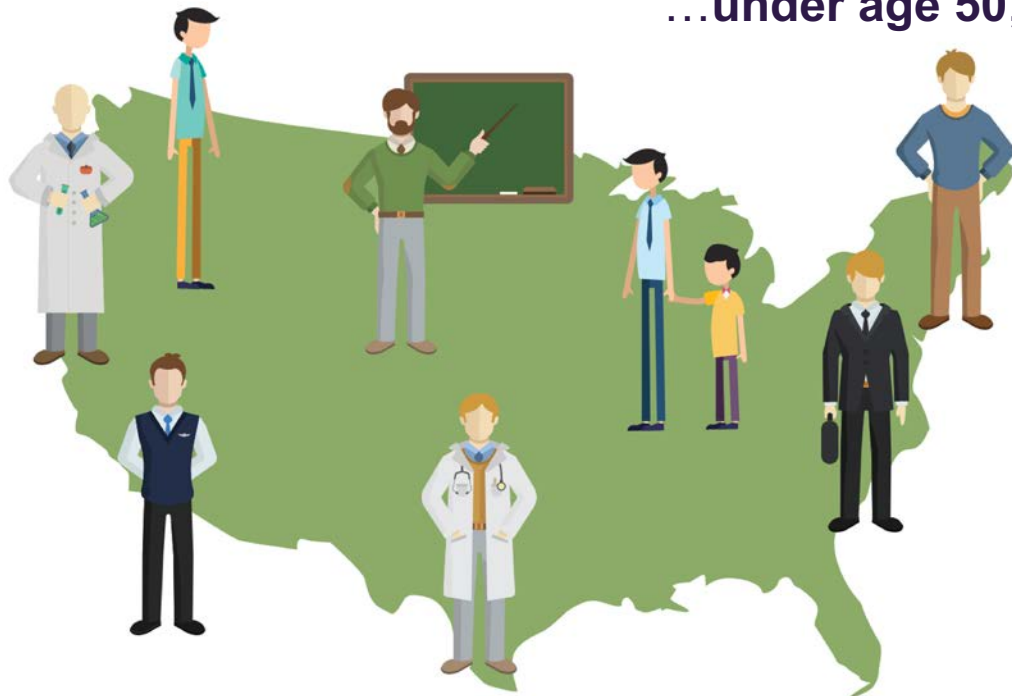
To support **responsible data sharing** practices we need to:



Ensure **diversity** amongst the professionals who **create data sharing policies, governance structures and legislation**

This is because the **profile of people** who are the most **trusting of the data sharing process** is **very similar** to the **profile of the decision makers** in genomics...

...under age 50, male, with personal **experience of genomics** and from the **USA**



The aim of Society and Ethics Research is to.....

- Explore the voice of publics to enable further discussion, debate, evaluation, ethical review
- Feed this work into
 - Policy
 - Governance
 - Regulation

It couldn't have happened without.....

PI

Anna Middleton

Project Management

Lauren Robarts

Data Analysis

Richard Milne

Kate Morley

Sanger Web team

James Smith

Paul Bevan

Claire Stead

Films

Tim Pope

Loudcity

Funding

Wellcome: Audrey Duncanson

Collaborators for Each Country/Translation

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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

Russian: Vera Izhevskaya, Elena Baranova, Alena Fedotova, Nadia Kovalevskaya

Spanish: Anne West, Maria Cerezo

Swedish: Heidi C Howard, Josephine Fernow

Ghana: Jerome Atutornu

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Danya Vears

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Madeleine Murtagh

Connecting Science: Julian Rayner

GA4GH: Peter Goodhand

