

Participants' perspectives and the evolution of genomic data sharing policies

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& HEALTH POLICY

- I receive support for work on genetic/genomic data sharing policy from the U.S. National Cancer Institute Grant R01 CA237118
- The views expressed are my own

- Evolution of genomic data sharing policies
- Participant perspectives on data sharing
 - Sources of diversity
 - Sampling of empirical studies
 - Consent/willingness to participate
 - Strategies to build trust and significance of trust
 - Return of results
 - Governance
 - Indigenous groups

Policy Evolution



Policies

Events

Bermuda Principles

1990s

Autism, PXE biobanks

GC Data Release and Resource Sharing

2005

Nuu-chah-nulth case

Havasupai case

GWAS/dbGaP

GSR to controlled access

Homer et al: can identify from pooled data

GA4GH

Genomic Data Sharing

2015

Precision Medicine Initiative

Cambridge Analytica

Equifax Breach

Most GSR back to unrestricted access

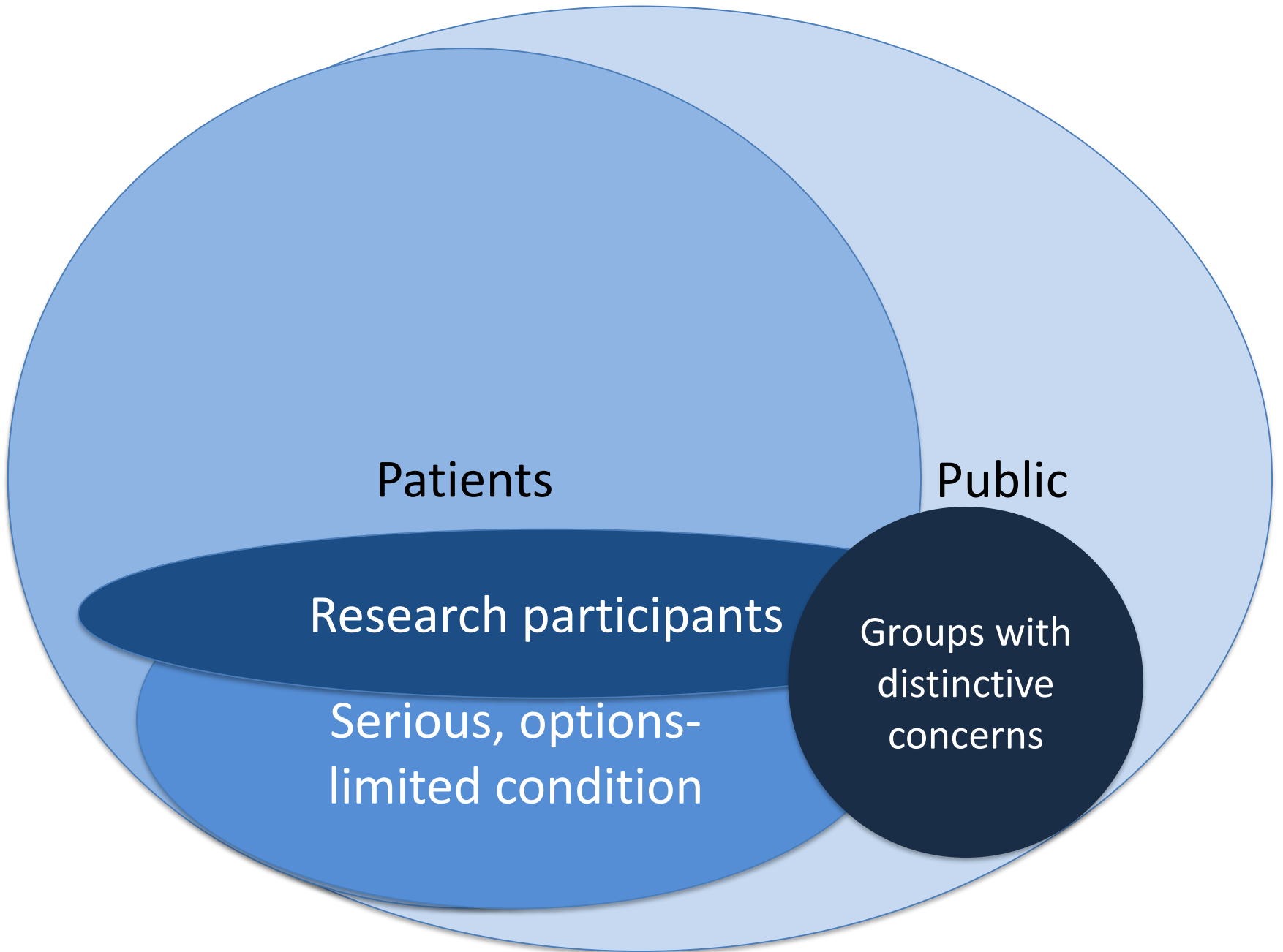
Golden State Killer case

Data Management and Sharing

2020

Tri-Agency Research Data Management

Sources of Diversity



Patients

Public

Research participants

Groups with
distinctive
concerns

Serious, options-
limited condition

Perspectives: A Sampling

Consent/Willingness to Share

Open

A systematic literature review of individuals' perspectives on broad consent and data sharing in the United States

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Purpose: In 2011, an Advance Researcher Data Repository in biobanks only if patients provide consent. The American Society of Human Genetics (ASHG) and the National Human Genome Research Institute (NHGRI) require broad consent from donors.

Methods: We conducted a systematic review of the literature toward biobanking, broad consent, and data sharing. Databases included MEDLINE, Embase, and GenETHX. Study screening was performed by two reviewers.

Results: The final 48 studies (n = 8), mixed methods (n = 1), surveys (n = 2), study quality was fair (n = 27), and poor (n = 2).

Vast amounts of genomic data are generated for many types of research. These data are often placed in databases which may exist at both the decentralized or centralized sites, and Phenotypes. These data have one purpose—whether for research or clinical care—frequently can be used for other purposes. These facts raise two distinct, but related questions: what conditions data can be shared in order to increase what can be learned from them. The second is whether data can and should be shared with other investigators in academic institutions, the government, and the commercial sector.

Currently, regulations for the protection of research participants and the Health Information Technology for Economic and Clinical Health Act amendments to the Health Insurance Portability and Accessibility Act Privacy Rule¹ permit the sharing and repurposing of data under certain conditions

While the majority often expressed support for broad consent when that was the only choice offered, only a minority of respondents favored broad consent when other options, such as tiered or study-by-study consent, were offered... Willingness to give broad consent increased if data were de-identified. While individuals were generally willing for data or biospecimens to be shared with other academic researchers, individuals were less willing for their data to be shared in federal databases or with commercial enterprises.

Nonetheless, questions remain about the ethical and practical desirability and acceptability of broad consent for research and data sharing. Approaches to obtain permission for use of genomic samples and data include no consent, opt-out, opt-in, case-by-case, tiered or categorical,⁴ and broad or blanket consent. Many have argued that blanket consent for unanticipated future research uses is unethical⁵ or unworkable,⁶ whereas others argue that such consent is acceptable as long as additional protections are in place,⁷ especially since broad data sharing

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Submitted 3 May 2015; accepted 1 September 2015; advance online publication 19 November 2015. doi:10.1038/gim.2015.138

Public Opinion about the Importance of Privacy in Biobank Research

David J. Kaufman,^{1,*} Juli Murphy-Bollinger,¹ Joan Scott,¹ and Kathy L. Hudson¹

Concerns about privacy requires understanding willingness to participate conducted. Ninety per information, and 37% Nearly half (48%) would provide consent for ea tively, would grant acc only when responden Among respondents w

Nearly half (48%) would prefer to give permission once...for all research approved by an oversight panel. Slightly fewer (42%) wanted to be asked permission for each research project separately, and 10% preferred to select categories of research.... 81% agreed that [being asked for consent] would make them feel 'respected and involved'...

ingness. Survey respondents valued both privacy and participation in biomedical research. Despite pervasive privacy concerns, 60% would participate in a biobank. Assuring research participants that their privacy will be protected to the best of researchers' abilities may increase participants' acceptance of consent for broad research uses of biobank data by a wide range of researchers.

Introduction

Large, prospective cohort studies that use DNA samples annotated with varying amounts of medical, lifestyle, and environmental information are becoming standard research tools for examining the effects of genes, environment, and lifestyle on common complex diseases,¹⁻⁵ but

The National Institutes of Health (NIH) and other federal agencies have contemplated the creation of a large biobank that would recruit a nationwide representative sample of at least 500,000 people. A proposed study design¹² would establish recruitment sites across the country for the collection of biospecimens and the performance of a comprehensive baseline exam on each participant. Hospital

“I Would Allow These Researchers to Use My Samples and Information for Research.”

“If I Could Not Be Identified, I Would Be Willing to Have My Information and Research Results Available on the Internet to Anyone.”

Academic or Medical Researchers in the United States

Government-Funded Researchers

Pharmaceutical-Company Researchers

Aagree^a p Value

Aagree^a p Value

Aagree^a p Value

Aagree^b p Value

Household Income

\$0–24,999	89%	0.004	77%	0.02	72%	0.47	49%	0.91
\$25,000–49,999	90%		76%		75%		47%	
\$50,000–74,999	94%		80%		75%		48%	
\$75,000+	95%		88%		77%		54%	

Education

Bachelor's degree or higher	95%	0.01	87%	0.0004	74%	0.40	53%	0.39
No bachelor's degree	90%		77%		75%		48%	

Race or Ethnic Group

Black, non-Hispanic	85%	0.004	71%	0.06	71%	0.07	49%	0.13
Hispanic	89%	0.47	78%	0.48	69%	0.04	46%	0.33
White, non-Hispanic	93%	reference	81%	reference	76%	reference	50%	reference

Example: Perspectives into Policy

- NIH Genomic Data Sharing Policy requires consent for genomic research with specimens/cell lines created or collected after January 25, 2015:
 - Even if de-identified
 - Can be broad consent
 - Exception for “compelling scientific reasons”

The reason the Policy expects consent for research for the use of data generated from de-identified clinical specimens and cell lines...is because the evolution of genomic technology and analytical methods raises the risk of re-identification. Moreover, requiring that consent be obtained is respectful of research participants, and it is increasingly clear that participants expect to be asked for their permission to use and share their de-identified specimens for research.

Clinical Trial Participants' Views of the Risks and Benefits of Data Sharing

Michelle M. Mello, J.D., Ph.D., Van Lieou, B.S.,
and Steven N. Goodman, M.D., Ph.D.

ABSTRACT

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BACKGROUND

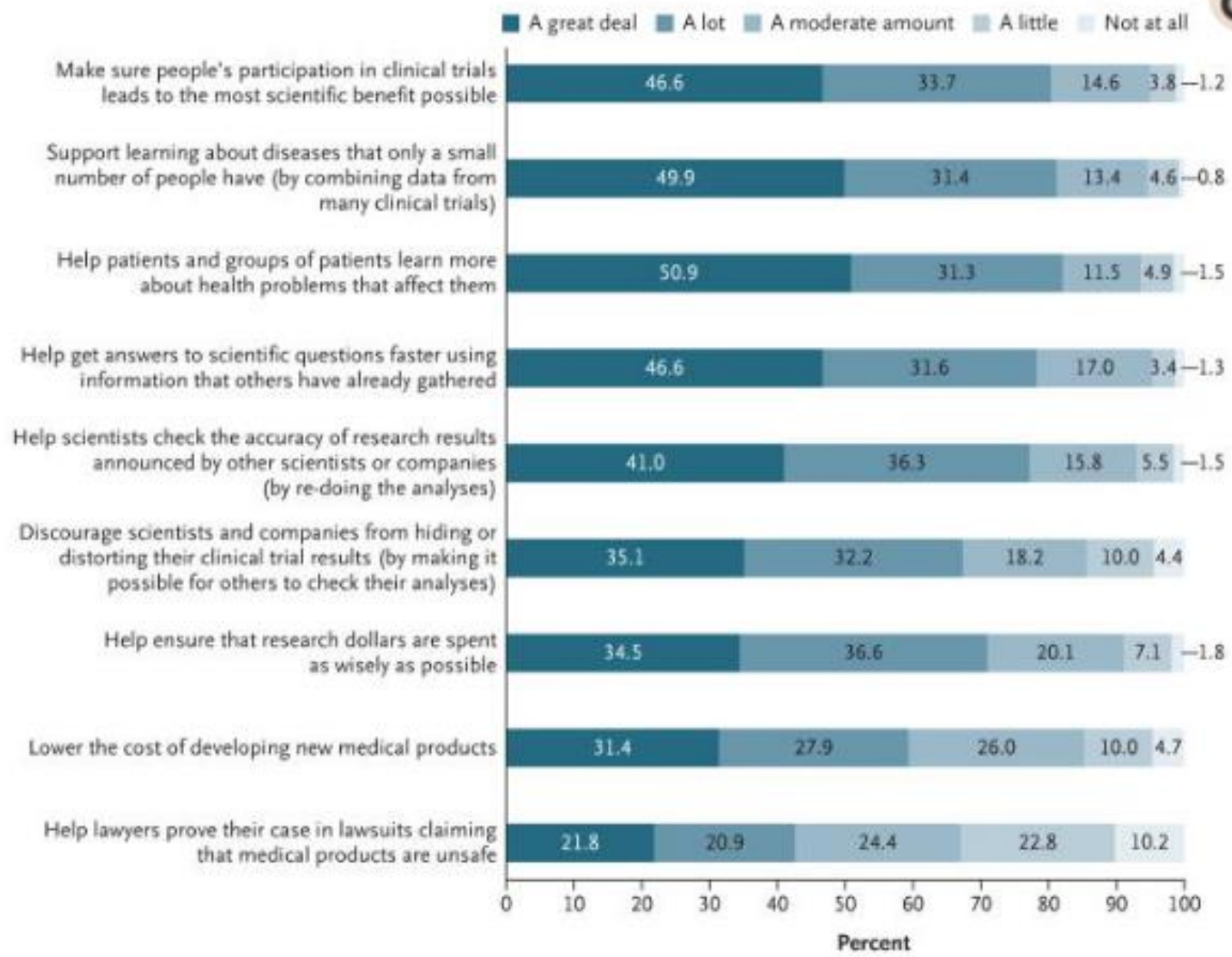
Sharing of participant-level clinical trial data has potential benefits, but concerns about potential harms to research participants have led some pharmaceutical sponsors and investigators to urge caution. Little is known about clinical trial participants' perceptions of the risks of data sharing.

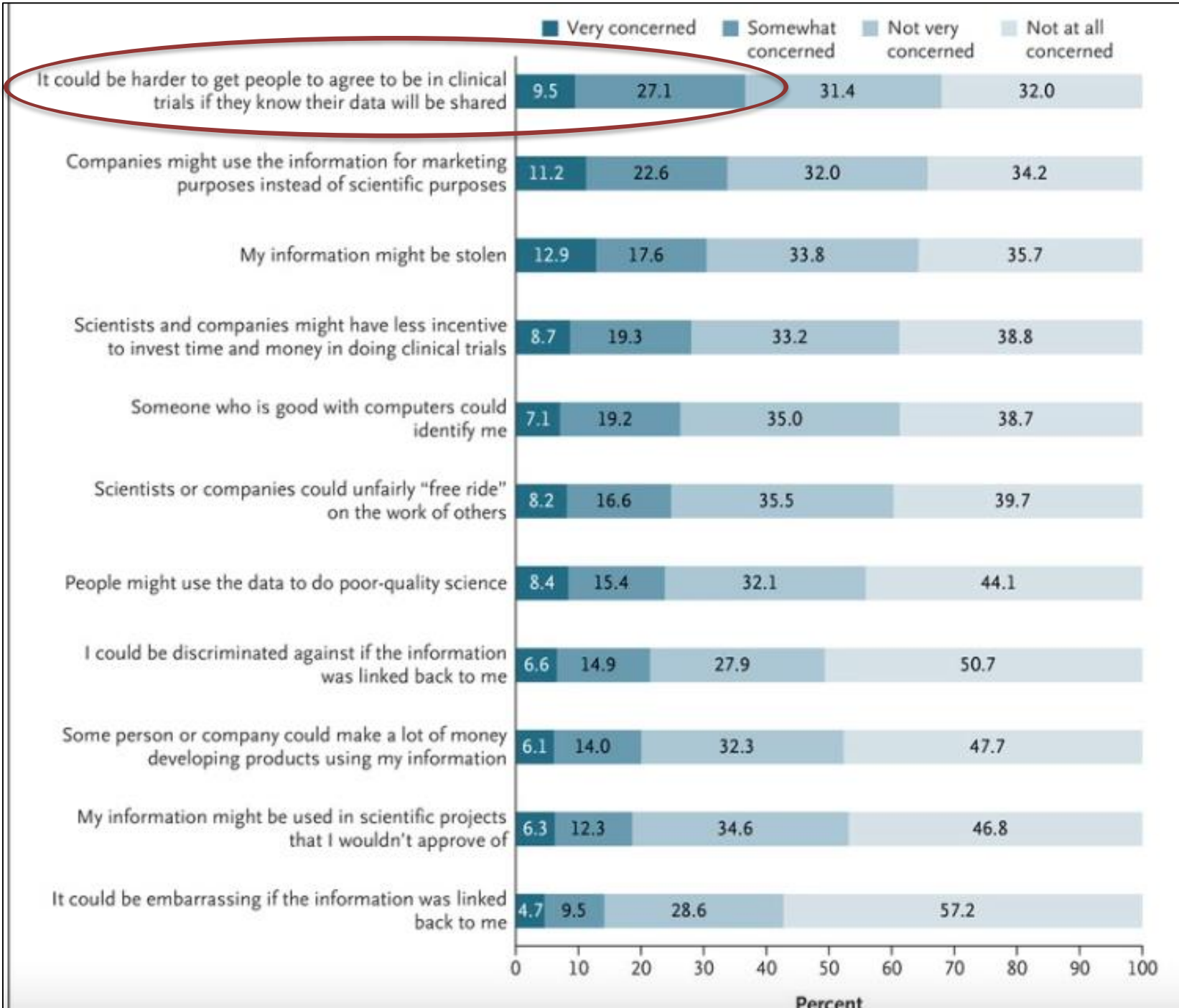
METHODS

We conducted a structured survey of 771 current and recent participants from a diverse sample of clinical trials at three academic medical centers in the United States. Surveys were distributed by mail (350 completed surveys) and in clinic waiting rooms (421 completed surveys) (overall response rate, 79%).

RESULTS

Less than 8% of respondents felt that the potential negative consequences of data sharing outweighed the benefits. A total of 93% were very or somewhat likely to allow their own data to be shared with university scientists, and 82% were very or somewhat likely to share with scientists in for-profit companies. Willingness to share data did not vary appreciably with the purpose for which the data would





Global Public Perceptions of Genomic Data Sharing: What Shapes the Willingness to Donate DNA and Health Data?

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Summary

Analyzing genomic data across populations is central to understanding the role of genetic factors in health and disease. Successful data sharing relies on public support, which requires attention to whether people around the world are willing to donate their data that are then subsequently shared with others for research. However, studies of such public perceptions are geographically limited and do not enable comparison. This paper presents results from a very large public survey on attitudes toward genomic data sharing. Data from 36,268 individuals across 22 countries (gathered in 15 languages) are presented. In general, publics across the world do not appear to be aware of, nor familiar with, the concepts of DNA, genetics, and genomics. Willingness to donate one's DNA and health data for research is relatively low, and trust in the process of data's being shared with multiple users (e.g., doctors, researchers, governments) is also low. Participants were most willing to donate DNA or health information for research when the recipient was specified as a medical doctor and least willing to donate when the recipient was a for-profit researcher. Those who were familiar with genetics and who were trusting of the users asking for data were more likely to be willing to donate. However, less than half of participants trusted more than one potential user of data, although this varied across countries. Genetic information was not uniformly seen as different from other forms of health information, but there was an association between seeing genetic information as special in some way compared to other health data and increased willingness to donate. The global perspective provided by our "Your DNA, Your Say" study is valuable for informing the development of international policy and practice for sharing genomic data. It highlights that the research community not only needs to be worthy of trust by the public, but also urgent steps need to be taken to authentically communicate why genomic research is necessary and how data donation, and subsequent sharing, is integral to this.

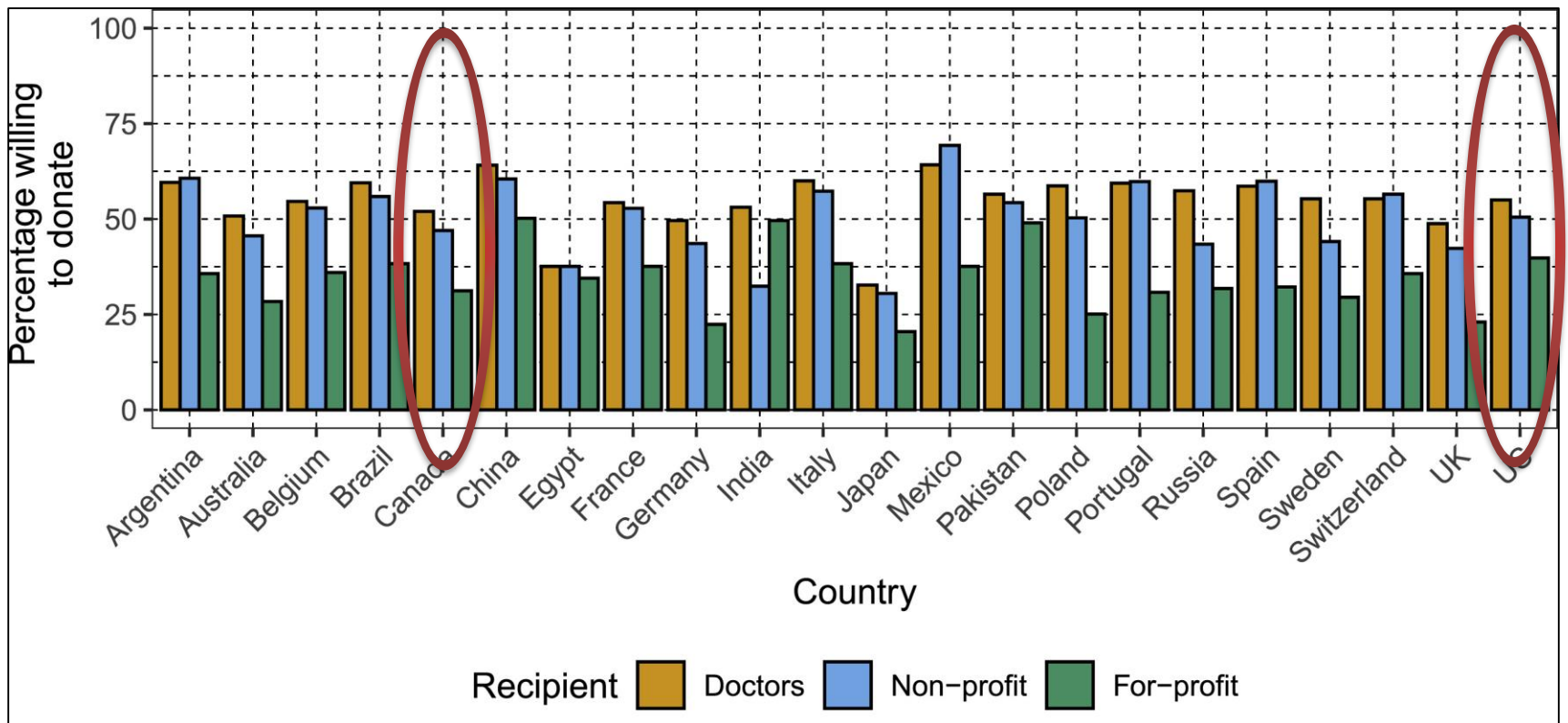
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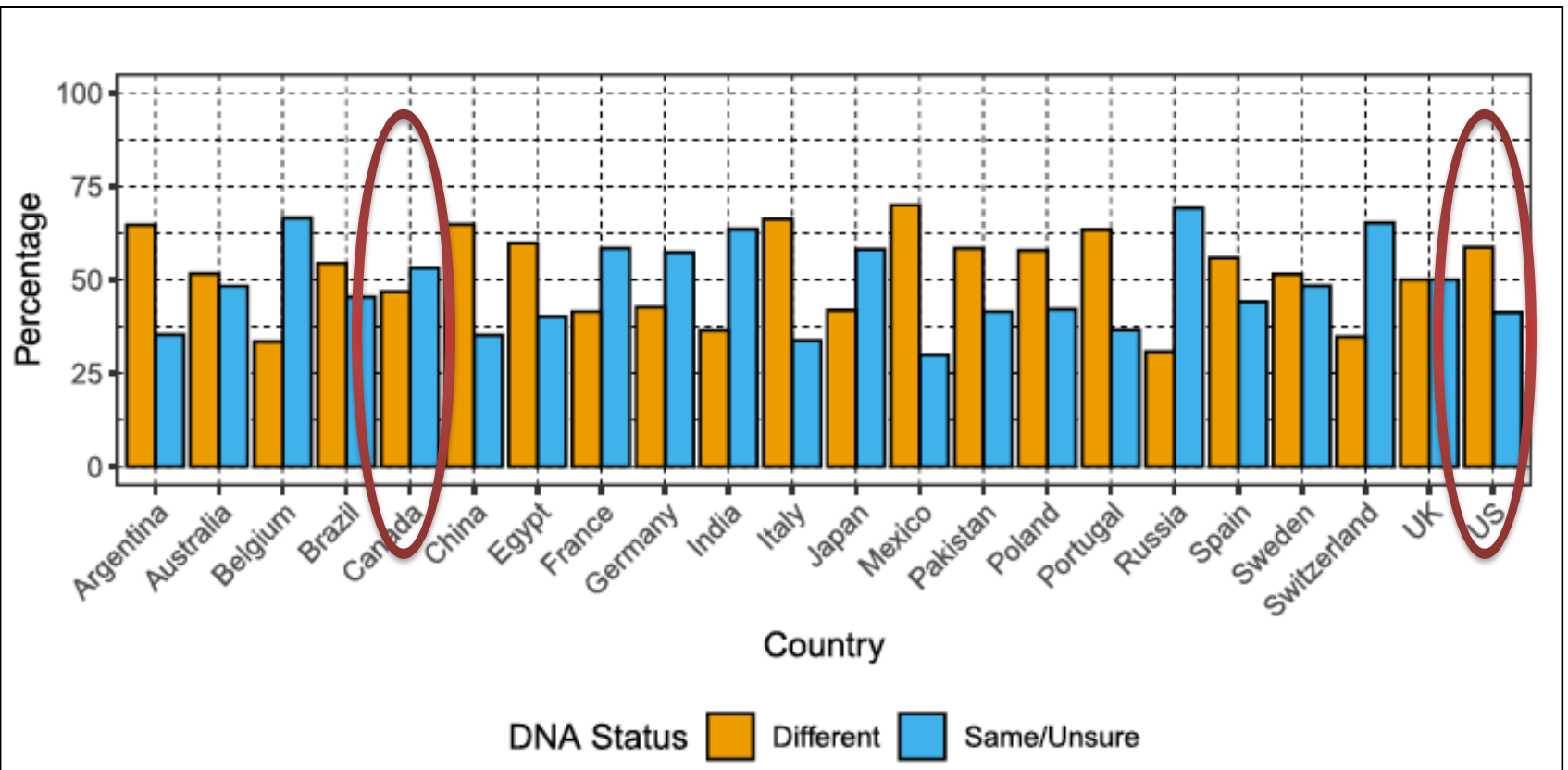
*Correspondence: am33@sanger.ac.uk

<https://doi.org/10.1016/j.ajhg.2020.08.023>

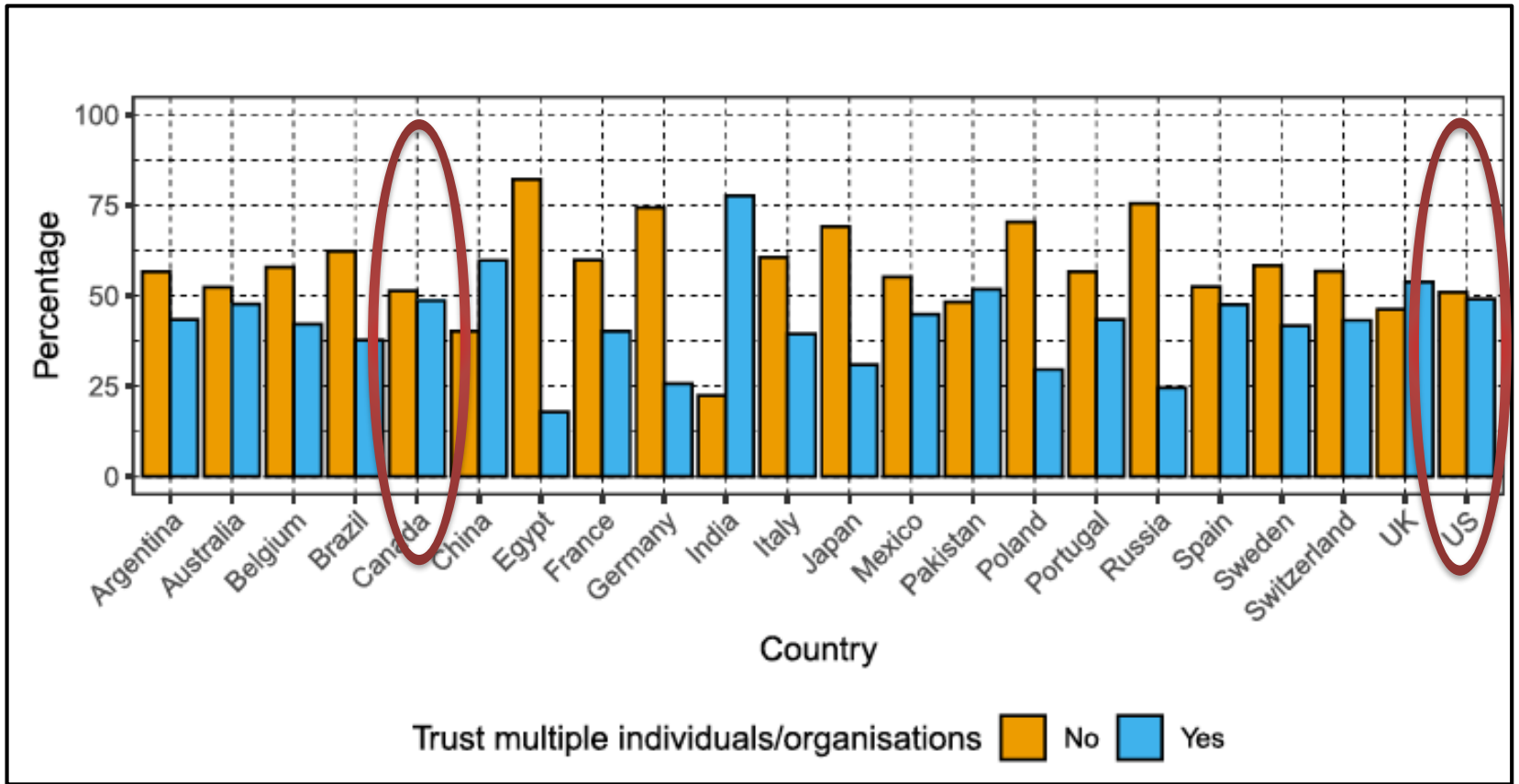
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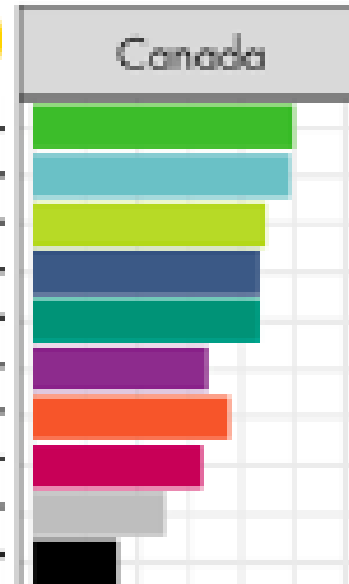


(Building) Trust

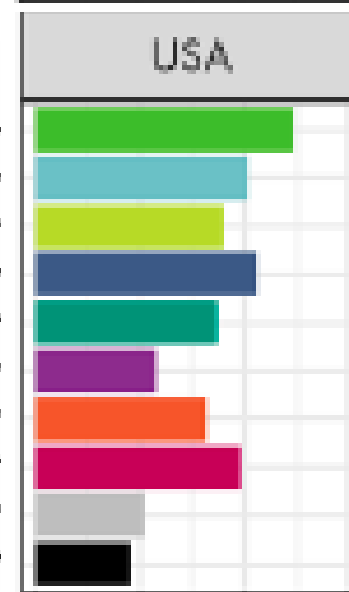


Q: What information would help you to trust the people asking you to donate DNA information and/or medical information? (choose all that apply)

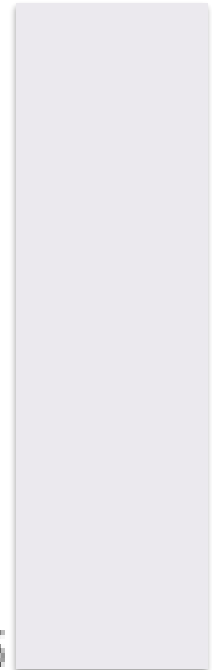
- Information about who will benefit
- The option to withdraw data
- Who is using your data and the purpose
- How others will benefit from data access
- Ability to opt out of access by others
- Details about sanctions for data misuse
- Access to own DNA/medical data
- Website explaining pros and cons of data access
- Direct communication with data gatekeepers
- Biographies of researchers who would access data



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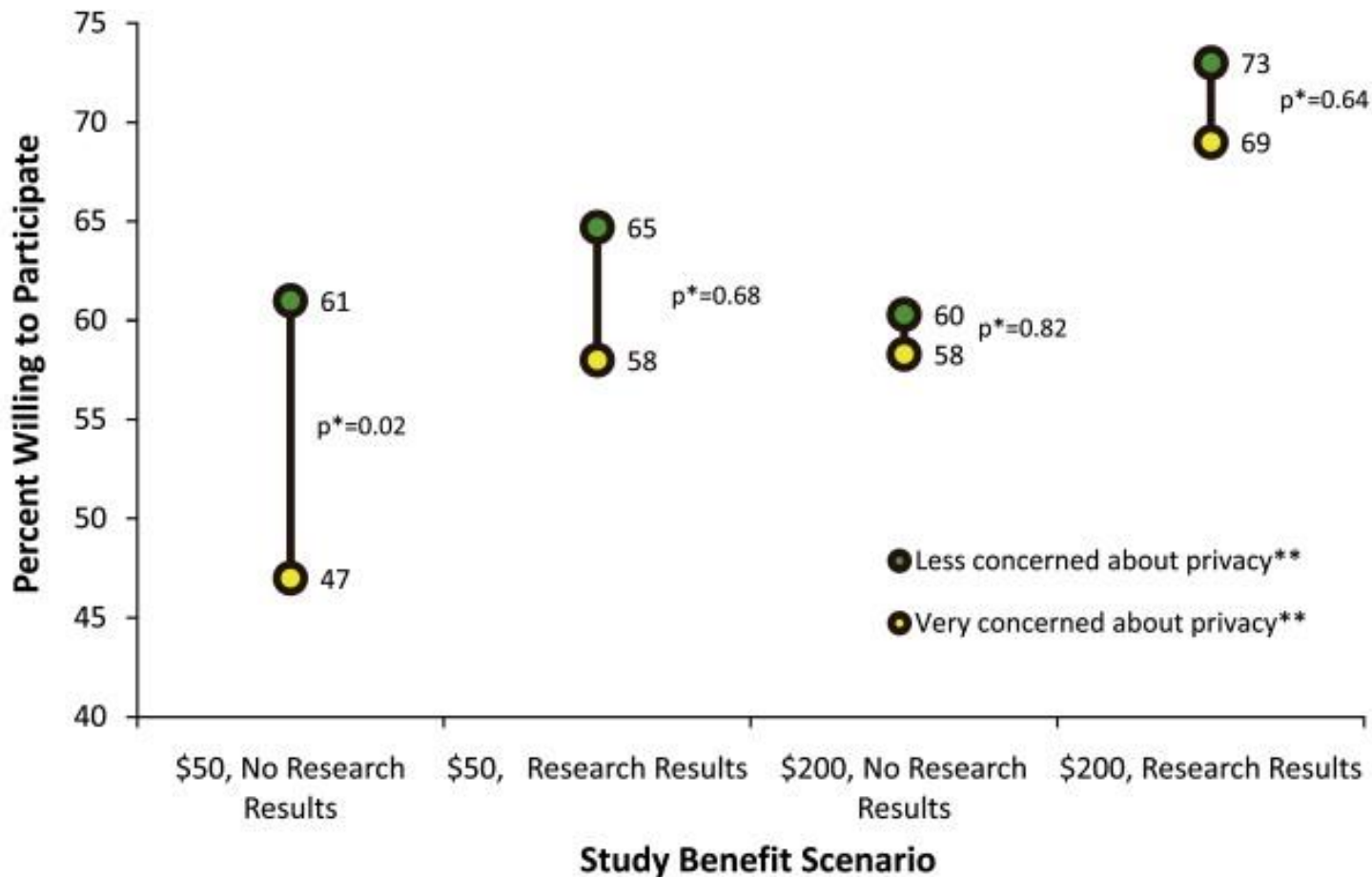


0 25 50 75



Theme	Occurrence (n = 22)	Representative quotes
Trust in the Health System	9	"I think with the reputation and just being attached to [the university] somehow, I think there is a pretty good chance that it's secure. I am really not concerned about that" (p. 4540).
Sharing Information with the Biobank is Low Risk	8	"I know there's certain DNA that's in blood that they could match. But other than that, I mean my blood sample, I don't see that they would be able to take anything out of the blood without matching it with something else to know that it's me" (p. 4982).
Assume Sharing Will be Limited to Trusted Entities	14	"I would assume it would be for their purposes, that it wouldn't be like somewhere in Illinois is asking for you to send my information. [...] It would stay within [the health system] or the university" (p. 4788).
De-identification Means Safety is Guaranteed	12	"Even if they have access to the medical history, it's more in an anonymous fashion that's not going to be able to easily identify me." (p. 5179)

Return of Results



*p values are for the comparison of the odds that those less concerned with privacy would participate to the odds that those very concerned would participate, adjusting for age, gender, race and ethnicity, household income, and education.

**These categories represent those who said they would be "very concerned" about "protecting my privacy" if they were participating in the study and those who said they would be somewhat concerned, a little concerned, or not at all concerned

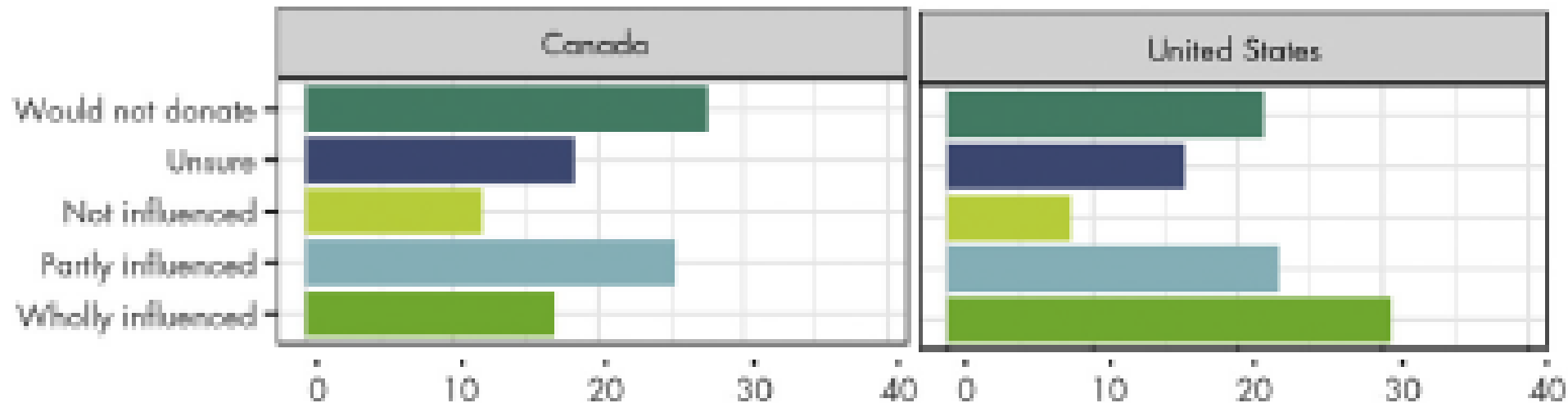
Value of items that could be returned from a study, by educational attainment of respondents to the Return of Value survey, 2018

Item	Educational attainment				
	All participants (N = 2,549)	High school or less (n = 569)	Some college (n = 685)	College graduate (n = 688)	Advanced degree (n = 597)
How I may respond to some medications based on my genetics ^a	6.30	5.78	6.40	6.50	6.44
How my genetics affect my risk of getting a medical condition ^a	6.28	5.74	6.41	6.44	6.46
How my lifestyle affects my risk of getting a medical condition ^a	5.98	5.62	6.08	6.12	6.06
Information about clinical trials near me ^a	5.81	5.43	6.05	5.92	5.80
Information about how researchers are using my information ^a	5.77	5.53	5.78	5.84	5.92
My ancestry ^a	5.70	5.42	5.91	5.74	5.69
Monetary compensation for taking part in the study	5.64	5.60	5.64	5.67	5.64
Basic information about me (my lab results, survey responses, height, weight, etc.)	5.39	5.46	5.38	5.36	5.37
Information from my medical record ^a	5.35	5.50	5.54	5.28	5.09
How my health and behaviors compare to others'	5.31	5.18	5.47	5.32	5.25
My genetic traits	5.29	5.38	5.39	5.20	5.23
How to connect with others like me in the study ^a	4.08	4.52	4.22	3.89	3.76

SOURCE Authors' analysis of Return of Value survey data from 2018. **NOTES** Ratings used a scale from 1 ("not valuable") to 7 ("very valuable"). A fuller version of the exhibit is available in appendix exhibit A6 (see note 16 in text). "Some college" and "advanced degree" are explained in the notes to exhibit 1. Ten participants did not provide their educational attainment levels. ^aBonferroni corrected $p < 0.000055$ (p values are from an F test for analysis of variance for differences in means).

Let's assume you were asked to consider donating your DNA information for research.

Would being offered a DNA readout influence your decision to donate?



Your DNA, Your Say:
Milne F et al. *Genet Med* 2022

Governance

Indigenous Groups

Should Navajo Nation moratorium on genetic research be lifted?

Not sure	316 (46%)
Yes	251 (36%)
No	122 (18%)

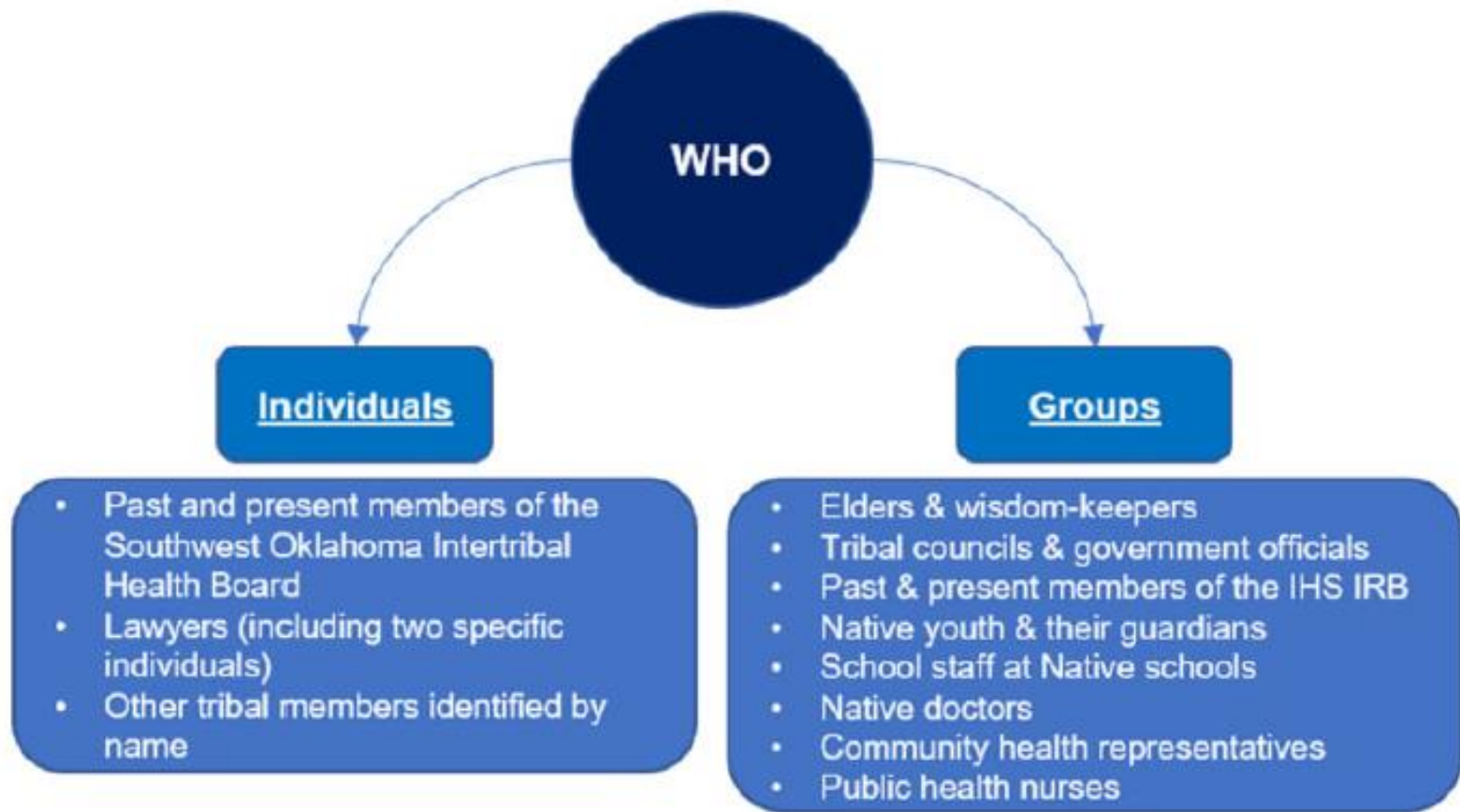
Comment Examples

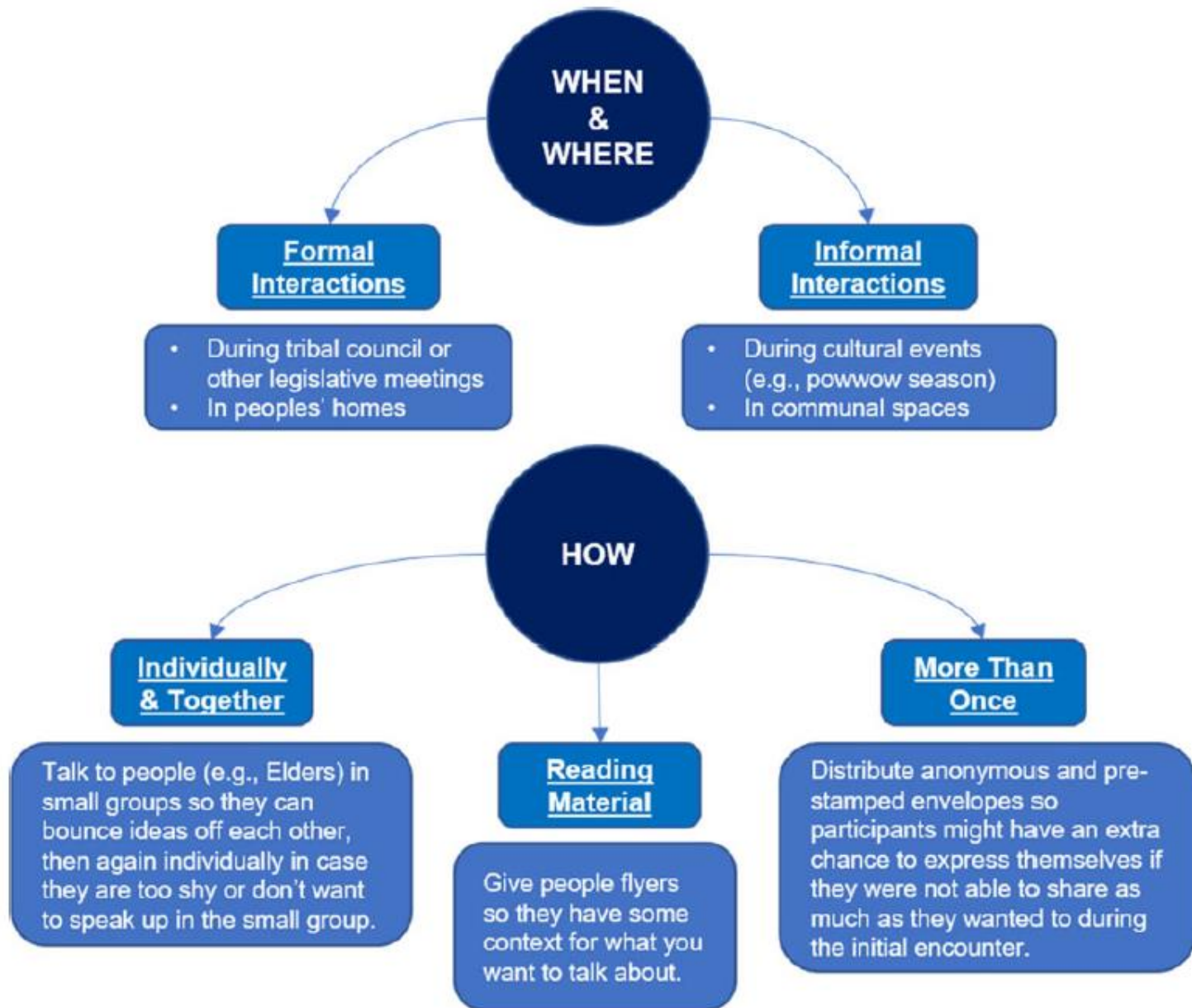
“We need to have more information on the subject.”

“This isn’t a simple question of ‘yes, it should be lifted’ or ‘no, it should not be lifted.’ The NN need to have the proper staff, resources, policies, procedures, and infrastructure in place to exercise appropriate oversight and to protect our people.”

Majority rated “Very important”:

- Data sharing protections in place
- Research benefits to Navajo tribe
- Inclusion of cultural knowledge









<https://fnigc.ca/ocap-training/>

Summary

Concerns/Consent

- Data hoarding violates the expectations and wishes of many participants
- Most participants want to be asked and prefer to be given choices, have reservations about sharing with for-profits, government
 - But in practice, most willing to consent to broad data sharing
- Not accommodating *all* preferences in policies ≠ violating rights BUT

Context

- Steps can be taken to increase comfort/trust, demonstrate respect, and establish trustworthiness (e.g., deidentification, return of value, care re access rules and other aspects of governance including participant voice, vigilance re privacy and security)
- Especially important if aiming for more representative data resources

Cautions

- Groups with cause for greater concern, sensitive research: special measures to involve and protect warranted
- Requires different mindset (e.g., communal focus, much longer time horizon, ceding control)

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