

# The underrepresentation of non-European samples in genomic studies: An overview and ethical considerations

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Cancer Genome Analysis course, October 2025

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# Outline

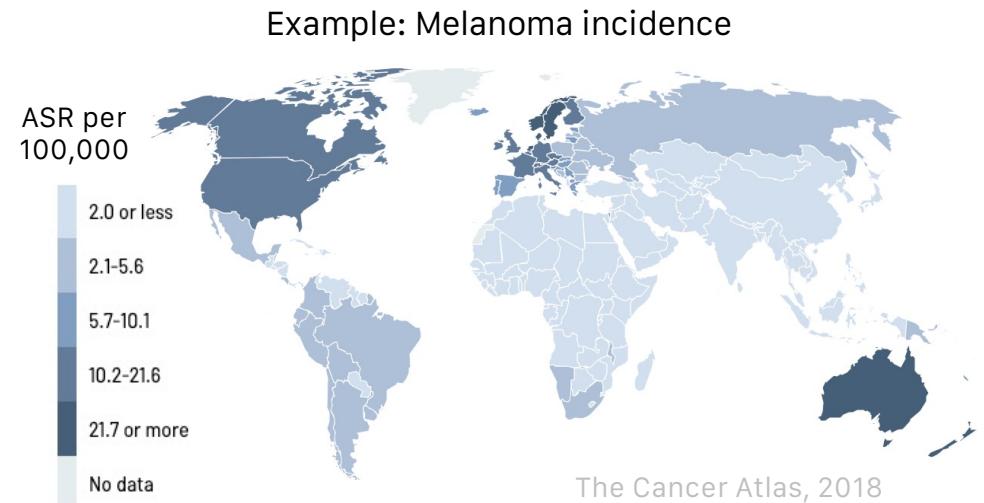
- ❑ Background: The importance of studying diverse populations to understand disease processes
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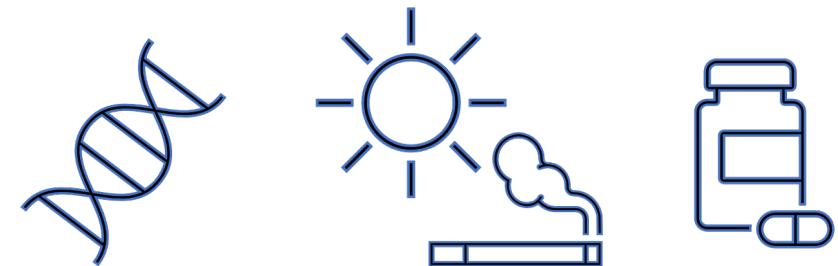
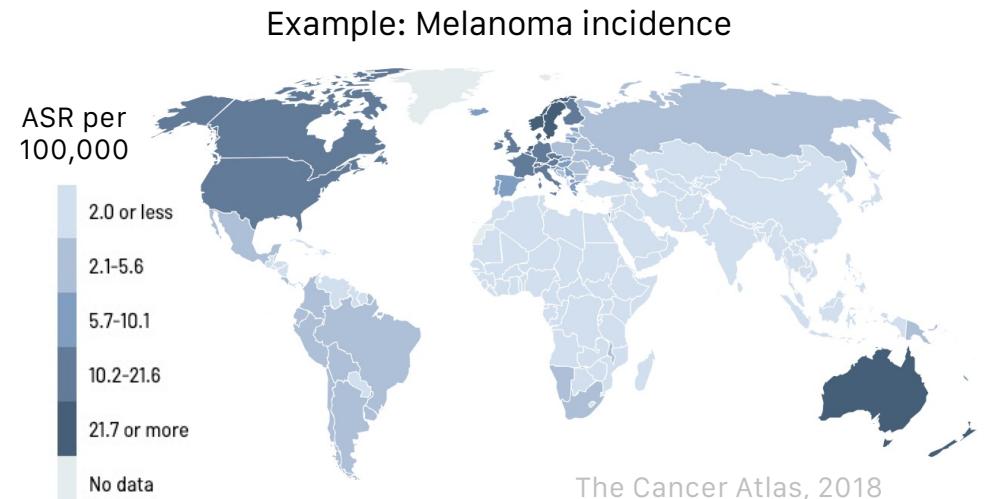
# Why is it important to study disease across different populations? Cancer as an example

- Site-specific cancer incidence rates vary up to 50-fold between geographic areas (The Cancer Atlas)



# Why is it important to study disease across different populations? Cancer as an example

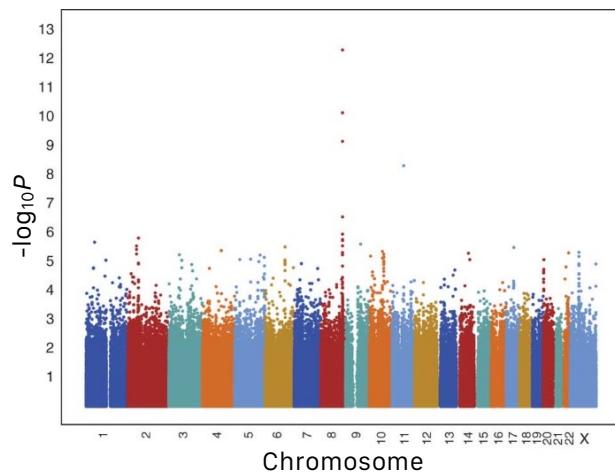
- Site-specific cancer incidence rates vary up to 50-fold between geographic areas (The Cancer Atlas)
- These differences are due to
  - Genetic risk factors
  - Lifestyle and environmental exposure
  - Implementation of effective cancer control measures



Traditional genomic studies have uncovered distinct genetic and environmental risk factors across populations

### Germline predisposition

Prostate cancer in men of African ancestry

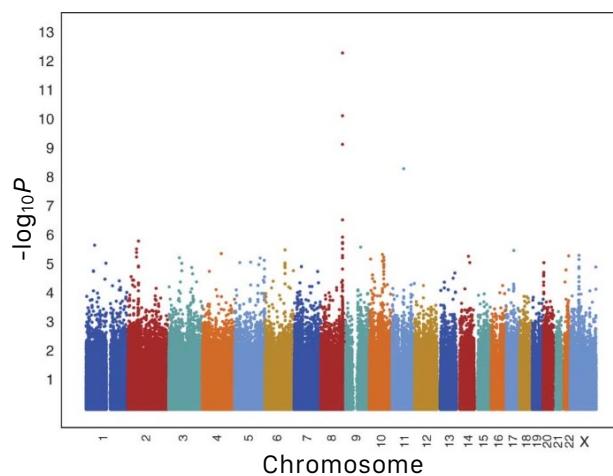


Freedman *et al* (2006)  
Haiman *et al* (2011)

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## Germline predisposition

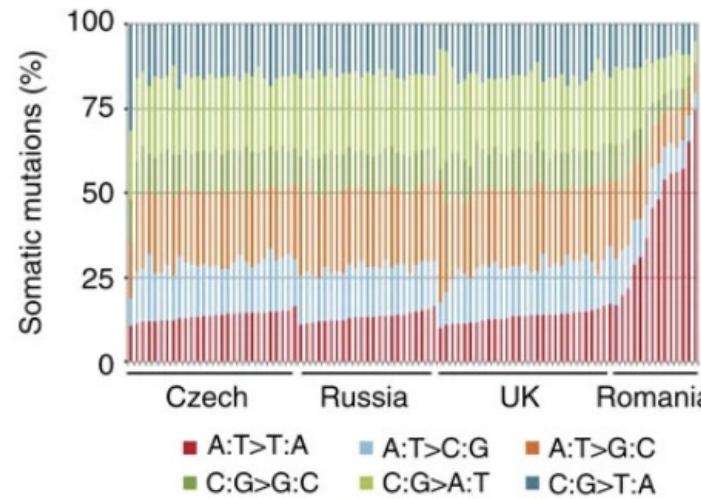
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Freedman *et al* (2006)  
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## Sample genomic profiling

Clear cell renal cell carcinoma in Romania

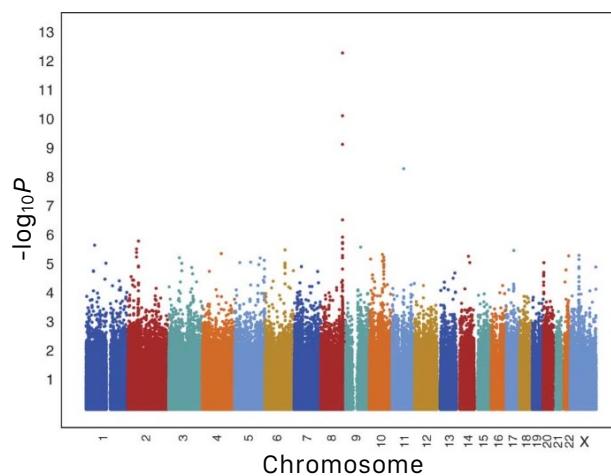


Scelo *et al* (2014)

# Traditional genomic studies have uncovered distinct genetic and environmental risk factors across populations

## Germline predisposition

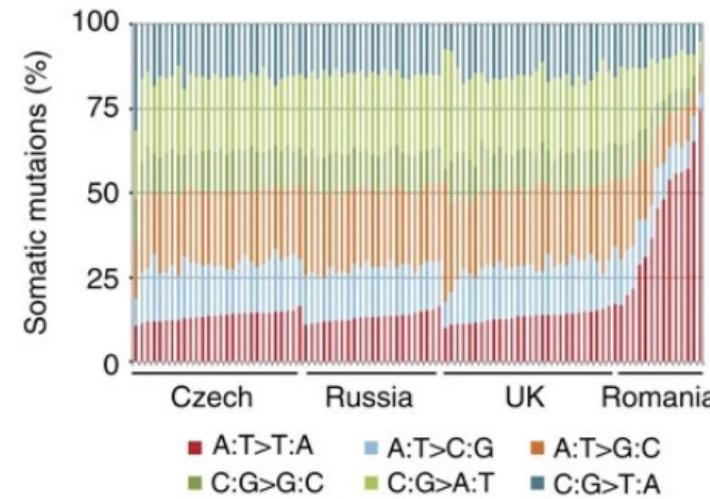
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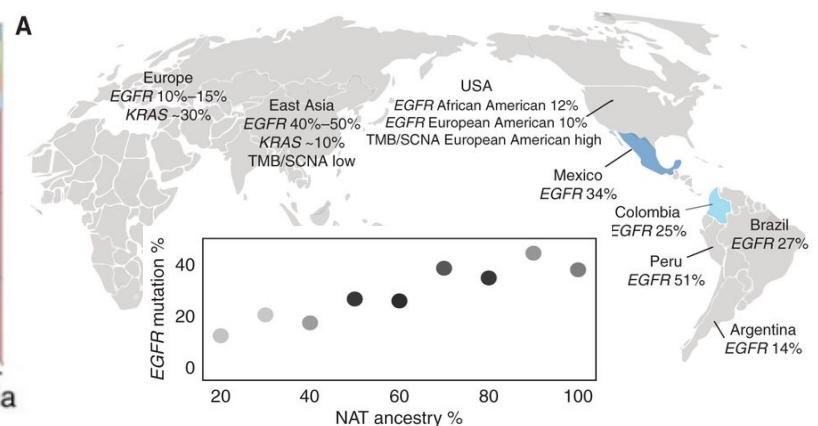
Clear cell renal cell carcinoma in Romania



Scelo *et al* (2014)

## Correlation analyses

Lung cancer in East Asians and Latin Americans

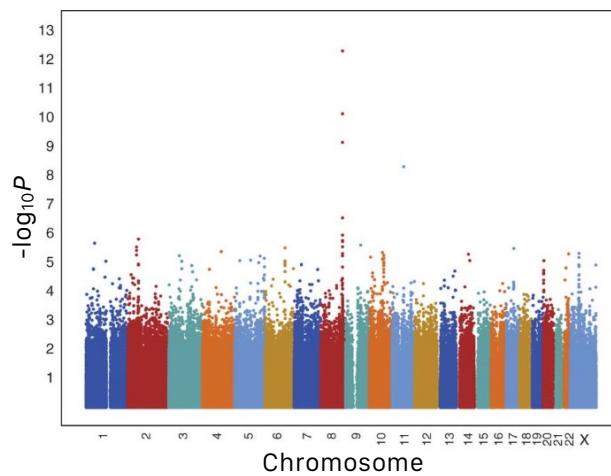


Carrot-Zhang *et al* (2021)

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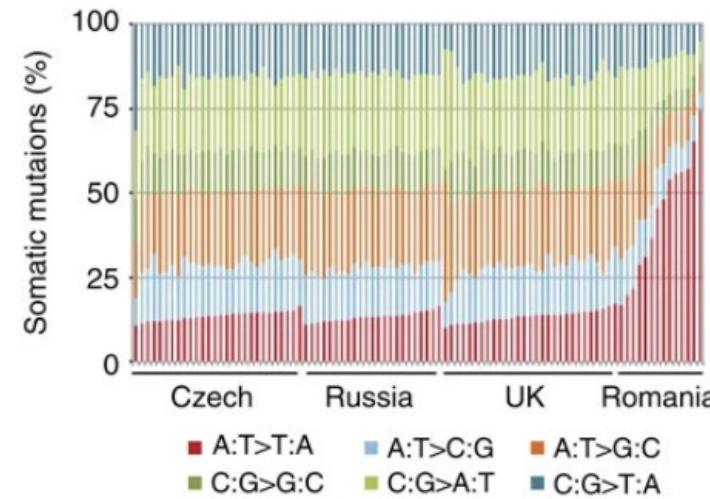
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Ancestry-specific  
genetic risk factors

## Sample genomic profiling

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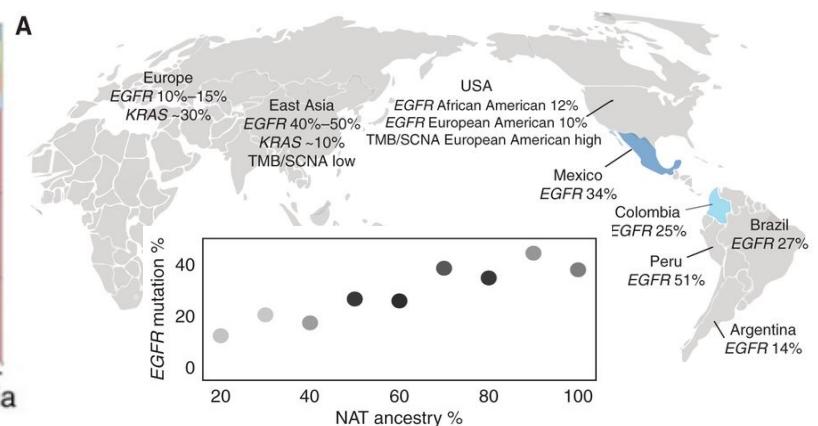
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Differing environmental exposures

## Correlation analyses

Lung cancer in East Asians and Latin Americans



Carrot-Zhang *et al* (2021)



Correlation between ancestry and genomic profile

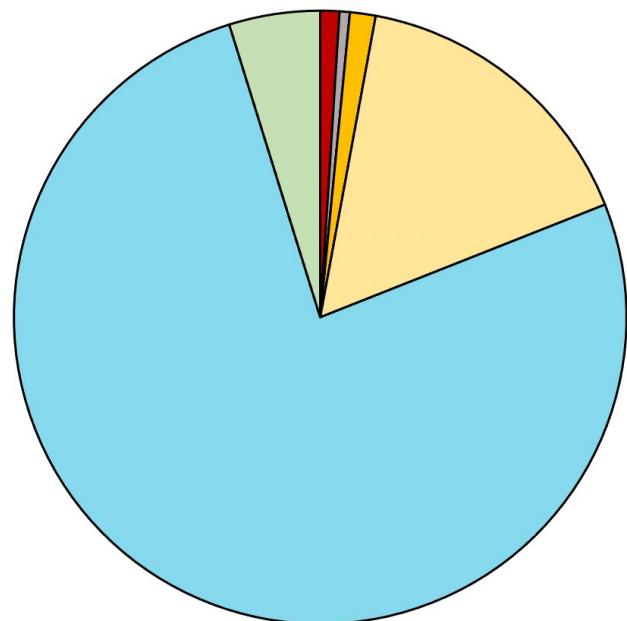
To have a good understanding of disease biology, it is of utmost importance to study individuals from different populations and geographical regions

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**What is the problem?** There is a gross underrepresentation of people of non-European descent in studies of cancer genomics

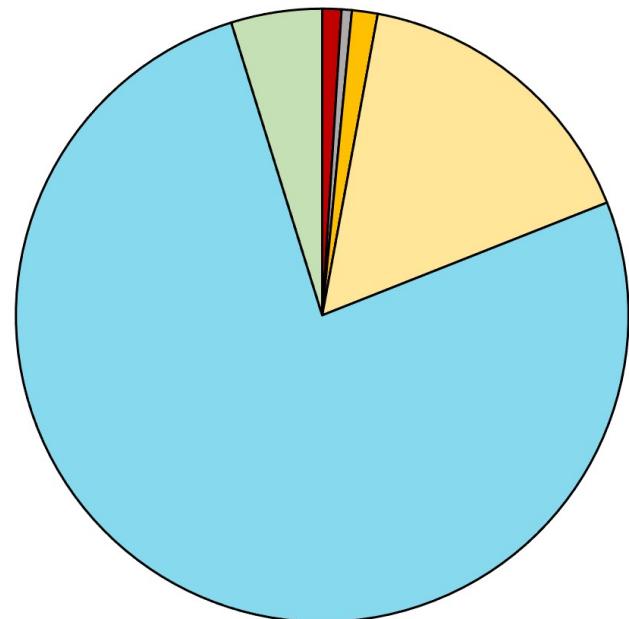
PCAWG (2020)



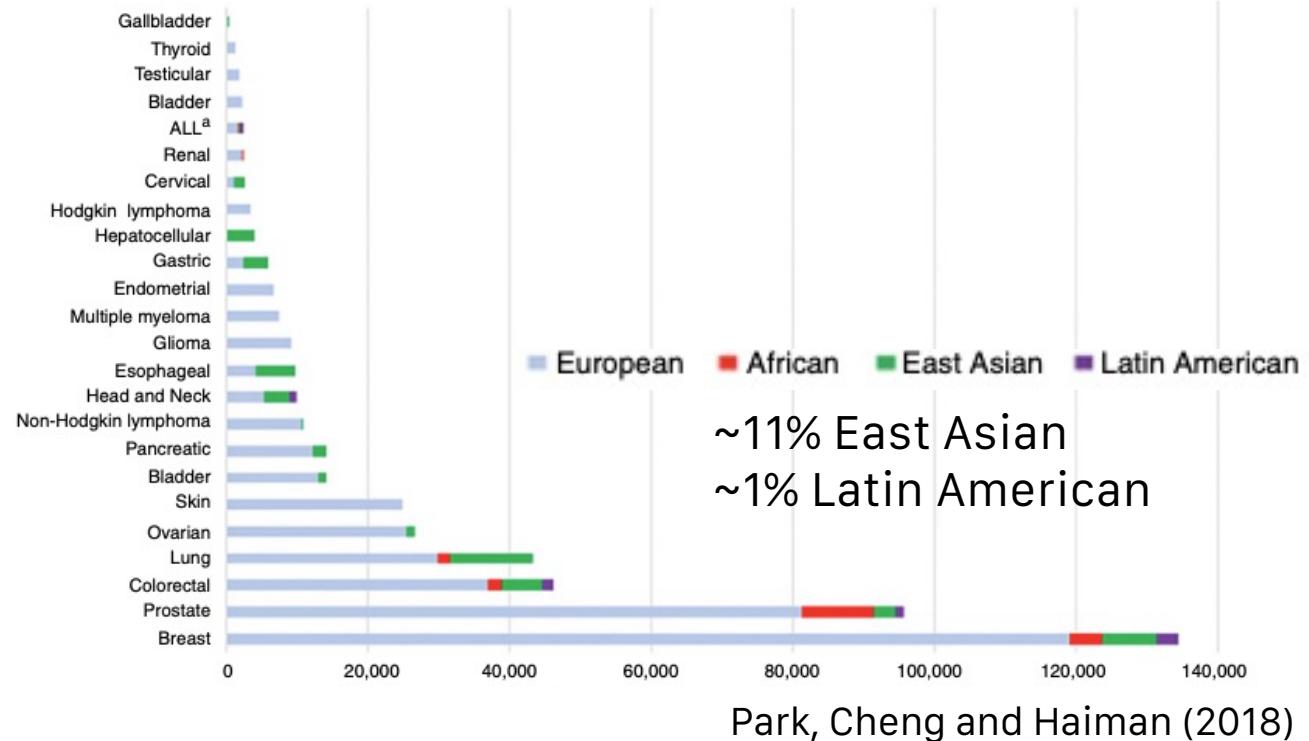
~1%   ■ Admixed American   □ Unknown  
~1%   ■ South Asian   ~17%   ■ East Asian  
■ European              ■ African

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PCAWG (2020)



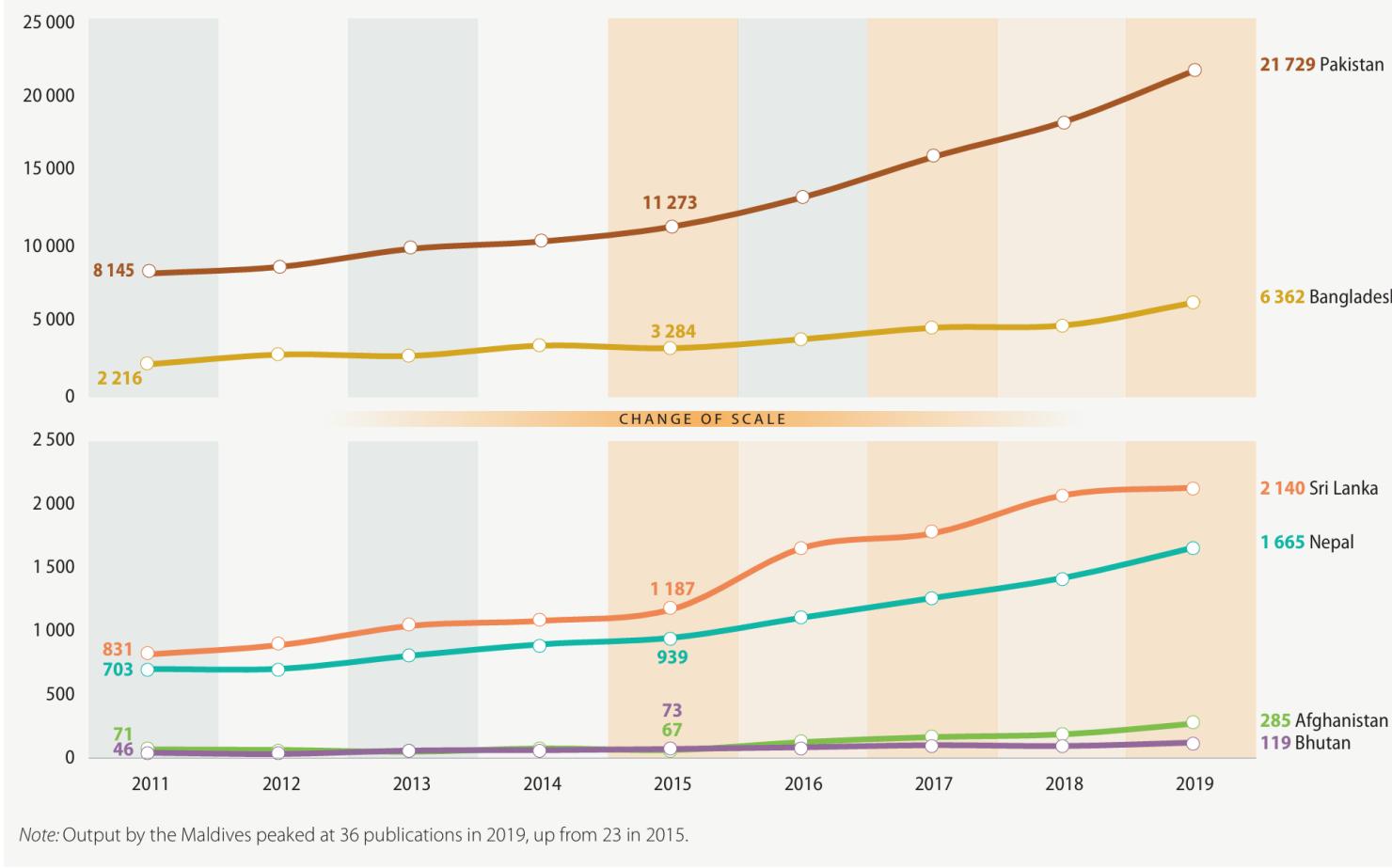
GWAS: number of cancer cases included in the discovery stage of GWAS by ancestral population and cancer site



# What is the problem? Few studies being conducted outside of the Global North (South Asia)

Volume of scientific publications in South Asia, 2011–2019

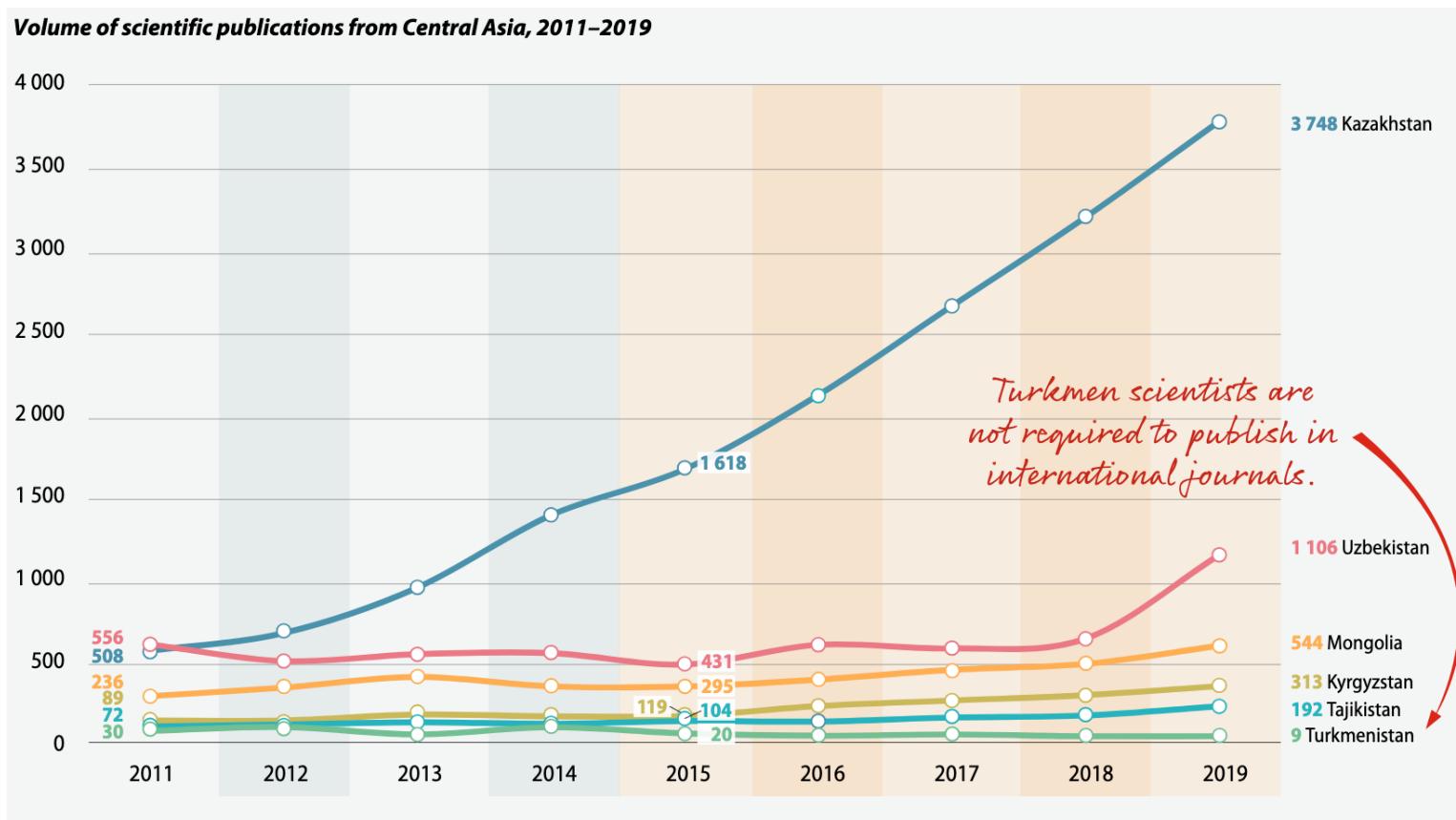
For countries with more than 100 publications in 2019



~160,000 UK  
~160,000 India  
~150,000 Germany  
~100,000 Italy  
~100,000 France  
~80,000 Spain

UNESCO (2021)

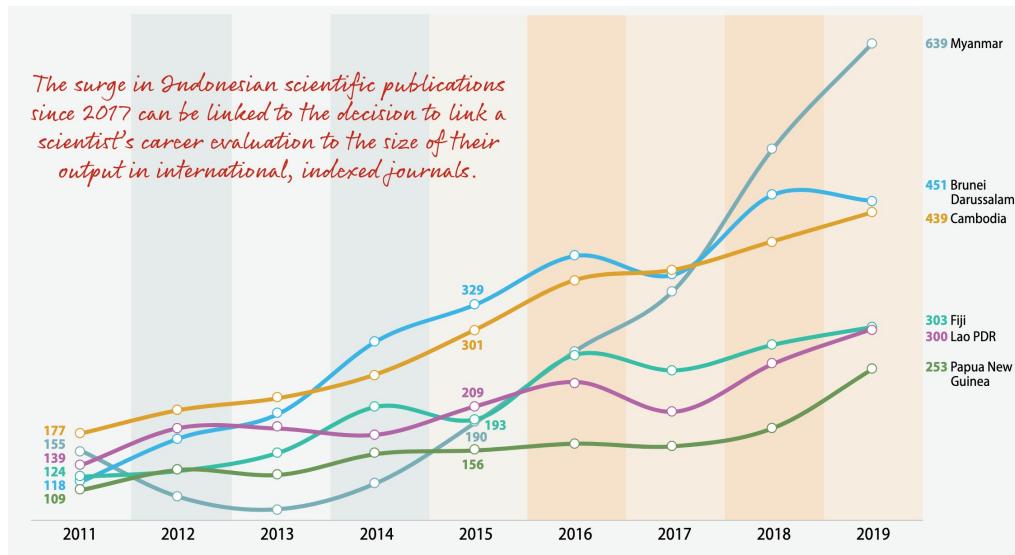
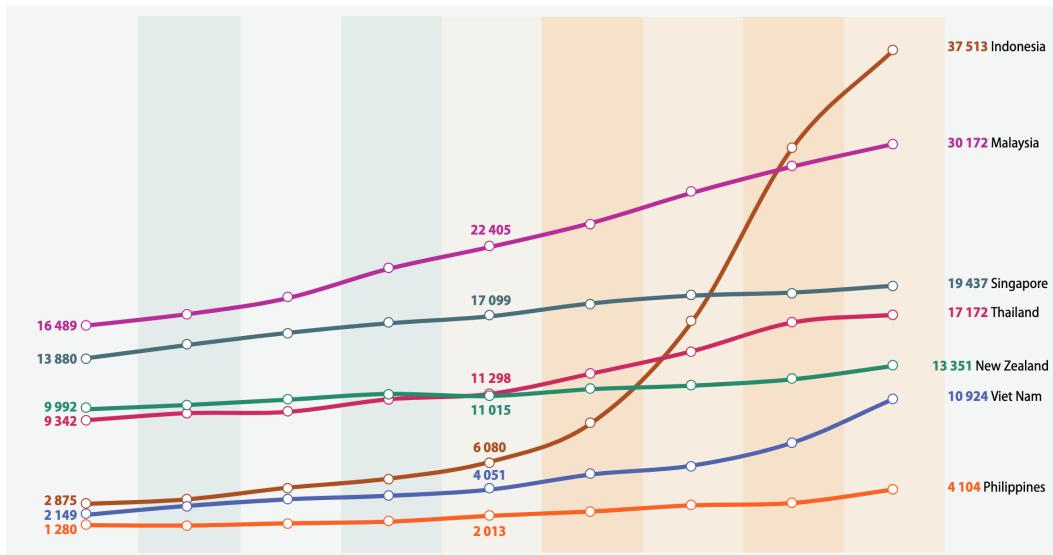
# What is the problem? Few studies being conducted outside of the Global North (Central Asia)



~160,000 UK  
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~100,000 France  
~80,000 Spain

UNESCO (2021)

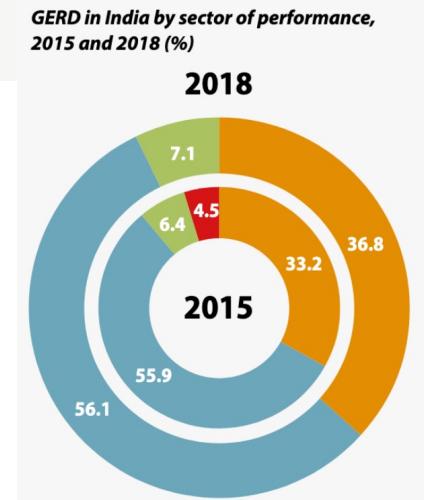
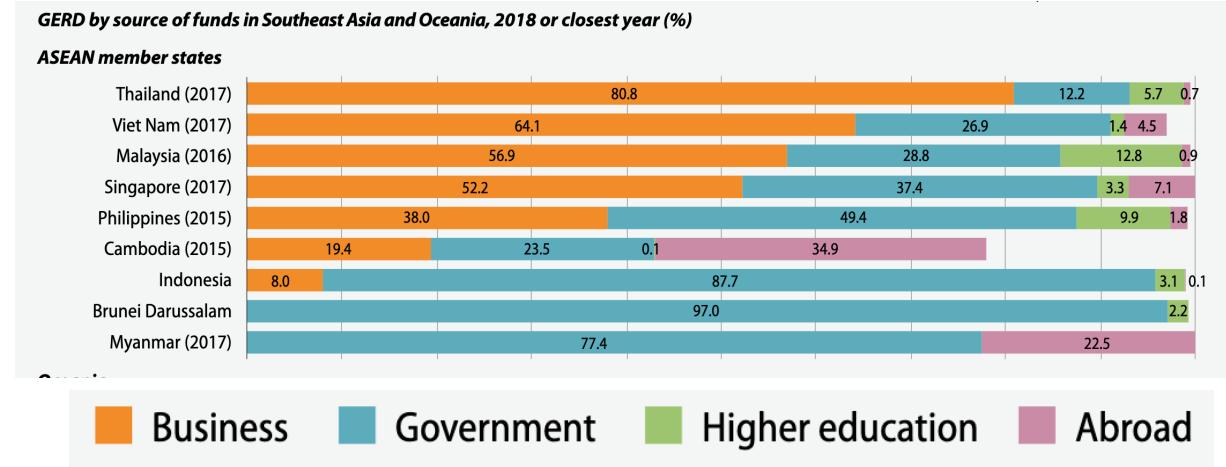
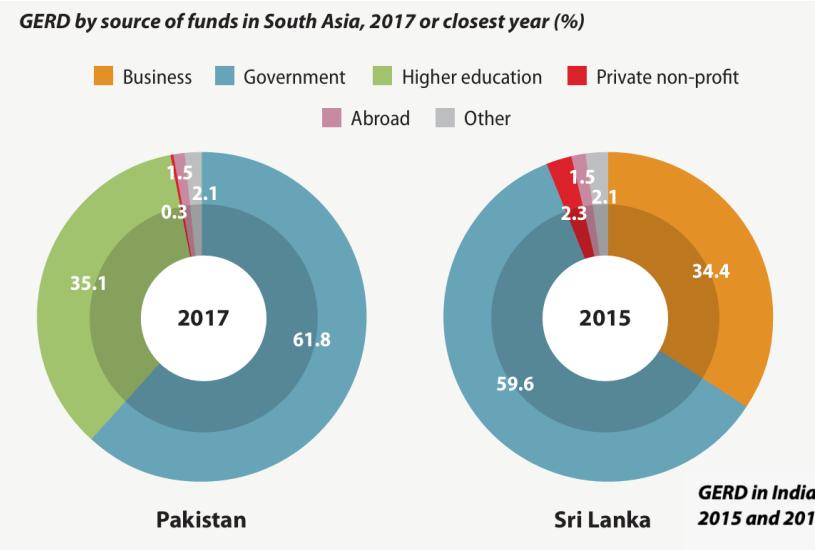
# What is the problem? Few studies being conducted outside of the Global North (South East Asia & Oceania)



- ~160,000 UK
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- ~87,000 Australia
- ~80,000 Spain

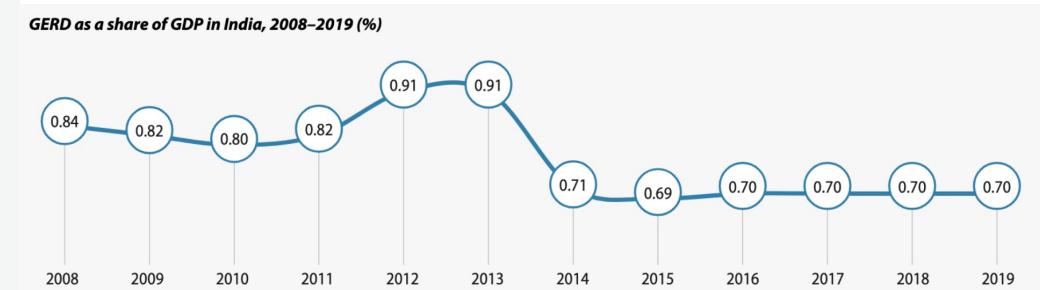
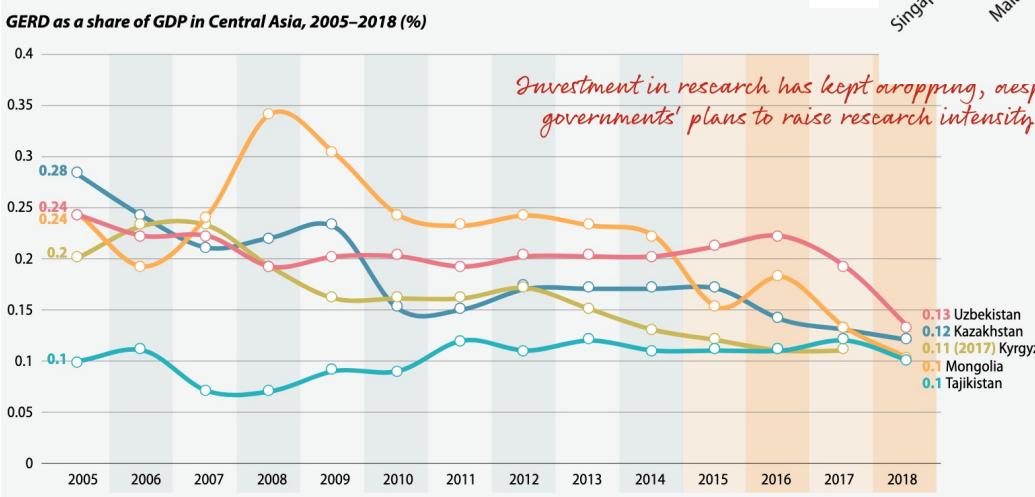
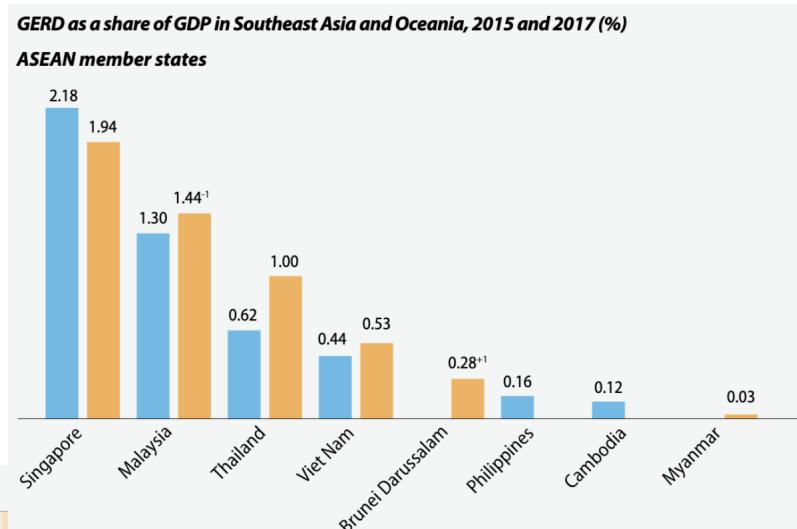
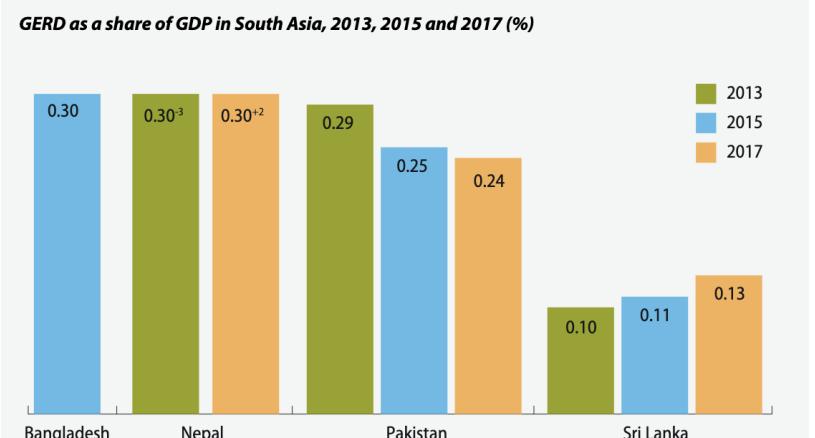
UNESCO (2021)

# What is the problem? Much of the funding for Global South science is from the public sector



UNESCO (2021)

# What is the problem? Generally, there is very little investment in R&D from the public sector in the Global South



UNESCO (2021)

LatAm: <0.70  
Brazil: 1.26

~2.3 EU

There is an underrepresentation of samples of non-European ancestry in public repositories, and fewer studies in general being carried out in the Global South, linked to little available funding for scientific research

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# Why does it exist? Few grants for studies being done in the Global South

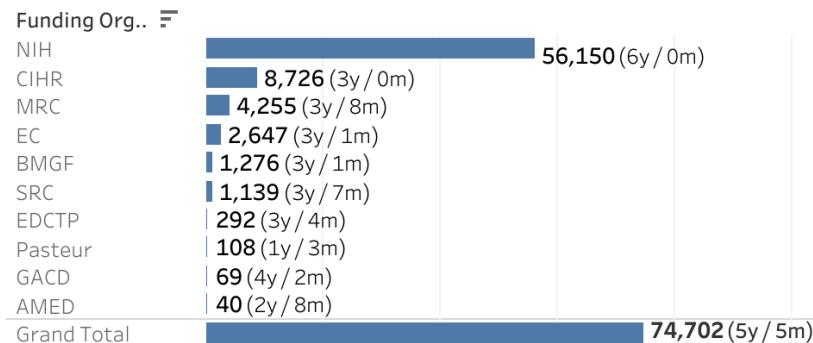
Select disease category

- All
- Neglected tropical diseases
- R&D Blueprint pathogens

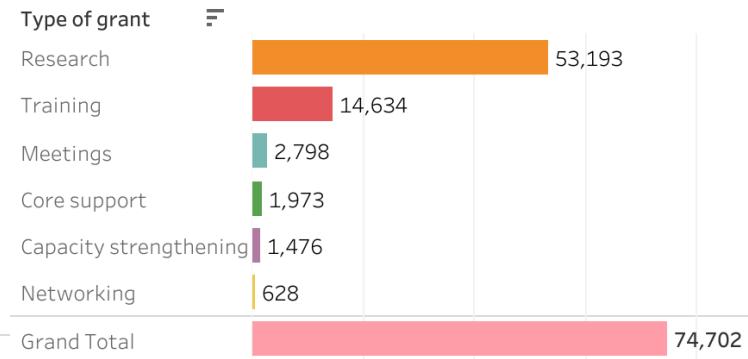
Number of grants for biomedical research in 2020

## A. No. of grants by funder

(Average grant duration in brackets)



## B. No. of grants by type



WHO (2020)

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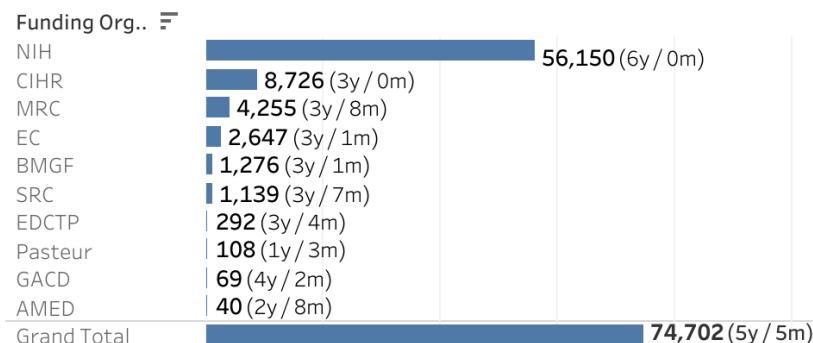
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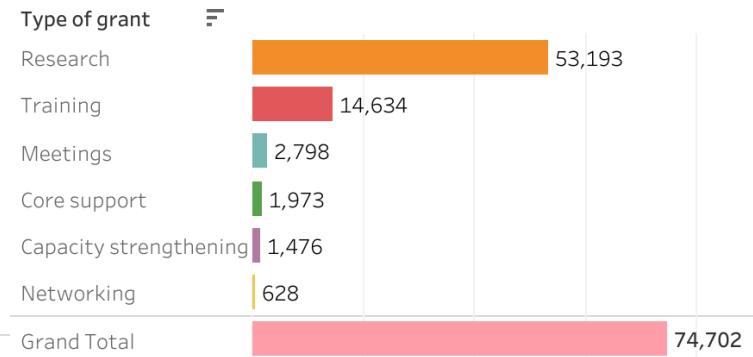
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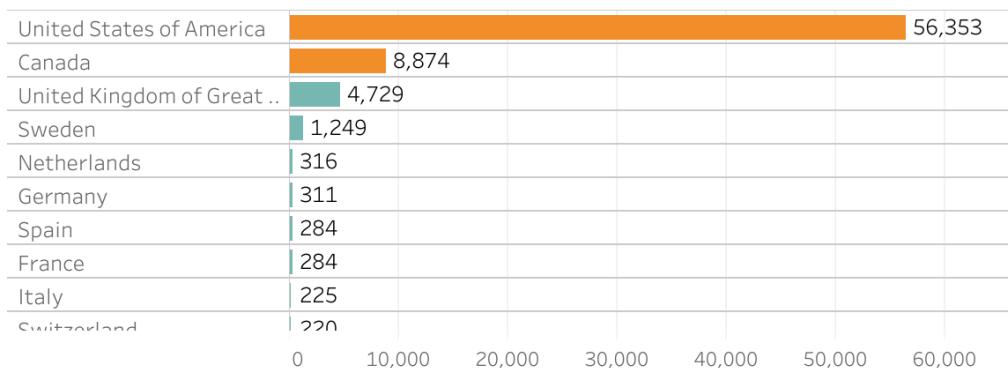
### C. No. of grants by recipient's WHO region and income group

WHO region	High income	Upper middle income	Lower middle income	Low income	Grand Total
Africa		227	208	173	608
Americas	65,242	62	3		65,307
Eastern Mediterranean			34	1	35
Europe	8,441	43	2		8,486
South-East Asia		5	69		74
Western Pacific	129	44	19		192
Grand Total	99%	73,812	381	335	74,702

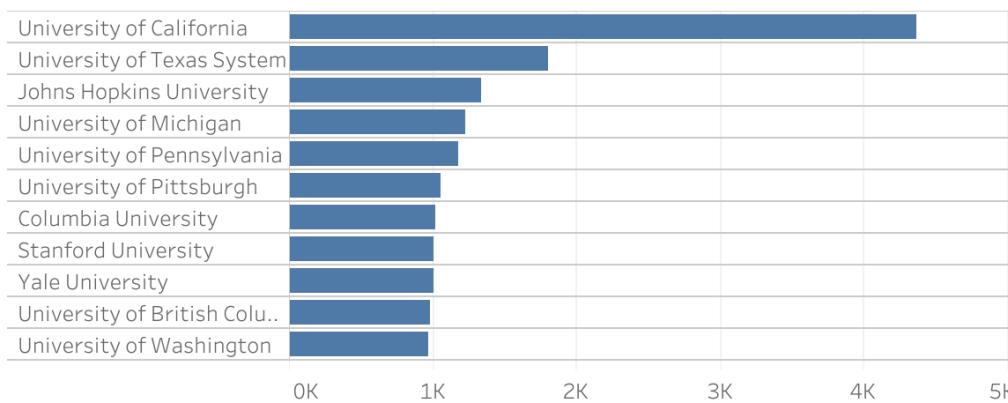
WHO (2020)

# Why does it exist? Few grants for studies being done in the Global South

E. No. of grants by recipient's country or territory



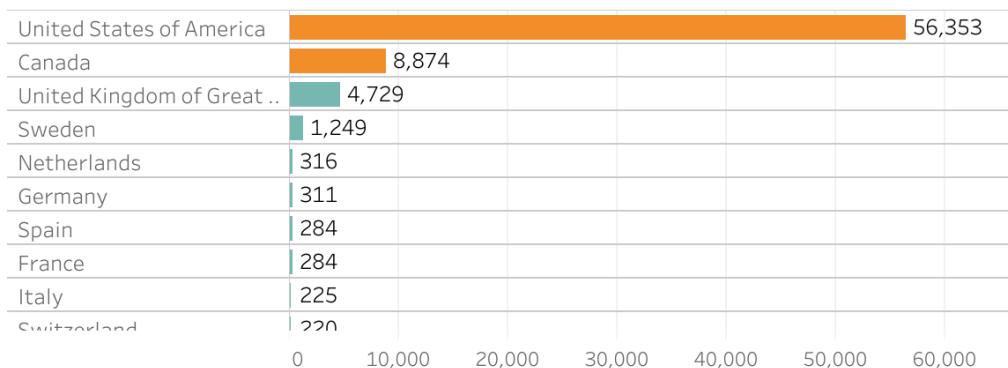
F. No. of grants by recipient organization



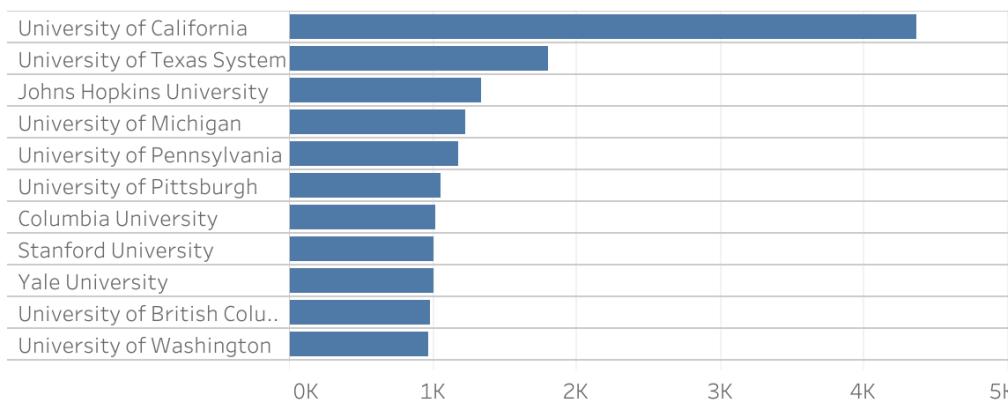
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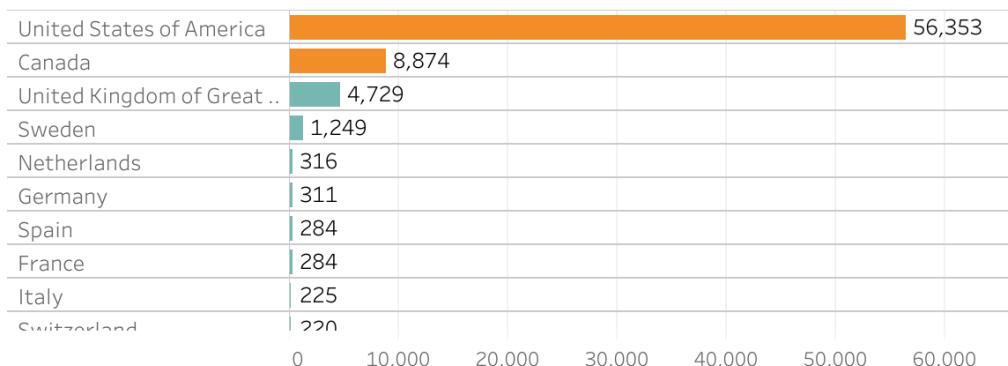


Brain drain  
Job insecurity  
Poor infrastructure

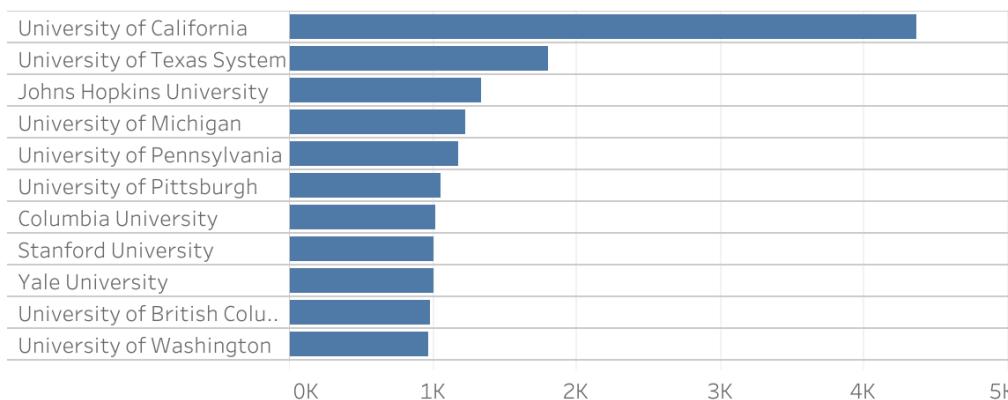
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Scientific partnerships

WHO (2020)

# Scientific partnerships can be great

- Enhance knowledge sharing
  - Complementary expertise → more exciting science!
  - Complementary infrastructure
  - Progress research at a faster pace
  - Cost sharing
  - Opportunities for internships and secondments
- ...



# Collaboration partners

## South Asia

*Top five partners for scientific co-authorship, 2017–2019 (number of papers)*

	<b>1st collaborator</b>	<b>2nd collaborator</b>	<b>3rd collaborator</b>	<b>4th collaborator(s)</b>	<b>5th collaborator</b>
Afghanistan	USA (151)	Japan (94)	Pakistan (82)	India/UK (68)	
Bangladesh	USA (2 132)	Japan (1 513)	Australia (1 505)	Malaysia (1 070)	UK (1 059)
Bhutan	Australia (83)	USA (80)	India (67)	Thailand (46)	Nepal/UK (41)
Maldives	India (30)	UK (22)	Italy (21)	Australia (16)	Nepal (12)
Nepal	USA (932)	India (650)	UK (531)	China (415)	Australia (357)
Pakistan	China (9 216)	Saudi Arabia (5 691)	USA (4 674)	UK (3 444)	Malaysia (3 179)
Sri Lanka	USA (946)	UK (880)	Australia (831)	India (599)	China (577)

Source: Scopus (excluding Arts, Humanities and Social Sciences); data treatment by Science-Metrix

## India

*India's top five partners for scientific co-authorship, 2017–2019 (number of papers)*

	<b>1st collaborator</b>	<b>2nd collaborator</b>	<b>3rd collaborator</b>	<b>4th collaborator</b>	<b>5th collaborator</b>
India	USA (23 628)	UK (9 421)	China (7 244)	Germany (6 825)	Korea, Rep. (6 676)

**Top five partners for scientific co-authorship in Southeast Asia and Oceania, 2017–2019 (number of papers)**

	<b>1st collaborator(s)</b>	<b>2nd collaborator(s)</b>	<b>3rd collaborator(s)</b>	<b>4th collaborator(s)</b>	<b>5th collaborator(s)</b>
Australia	USA (40 958)	China (36 750)	UK (30 121)	Germany (15 730)	Canada (13 185)
Brunei Darussalam	Malaysia (220)	China (160)	UK (152)	Australia (142)	Indonesia (111)
Cambodia	USA (345)	France/Thailand (248)		UK (246)	Australia (222)
Cook Islands	New Zealand (16)	USA (13)	France (11)	Australia (9)	UK (6)
Fiji	Australia (329)	USA (150)	UK (111)	India (82)	New Zealand (74)
Indonesia	Malaysia (3 633)	Japan (3 548)	Australia (1 805)	USA (1 743)	UK (1 395)
Kiribati	Fiji (10)	USA (8)	Australia (7)	UK (6)	Switzerland (4)
Lao PDR	Thailand (240)	UK (163)	USA (160)	Australia (145)	Viet Nam (124)
Malaysia	UK (4 807)	India (3 851)	Australia (3 741)	USA (3 716)	Indonesia (3 633)
Marshall Islands	USA (6)	Micronesia (5)	Australia/Japan/Palau (3)		
Micronesia	USA (54)	Australia (32)	UK (26)	Canada/France/Germany/ India/Japan/Rep. Korea/ Saudi Arabia/Spain/ Sweden (22)	
Myanmar	Japan (321)	China (291)	Thailand/USA (236)		UK (169)
New Zealand	USA (7 149)	Australia (7 087)	UK (5 605)	China (3 643)	Germany (3 017)
Niue	Australia (6)	Samoa/Vanuatu (4)		Solomon Islands/UK (3)	
Palau	USA (40)	Australia (13)	Japan/UK (12)		New Zealand (7)
Papua New Guinea	Australia (313)	USA (157)	UK (96)	France (70)	India (54)
Philippines	USA (1 503)	Japan (961)	China (723)	UK (693)	Australia (658)
Samoa	Australia (45)	USA (32)	New Zealand (24)	Fiji (17)	UK (11)
Singapore	China (15 327)	USA (10 129)	UK (5 486)	Australia (4 257)	Germany (2 578)
Solomon Islands	Australia (108)	UK (41)	USA (39)	Fiji (19)	France (17)
Thailand	USA (5 742)	Japan (3 704)	UK (3 149)	China (2 577)	Australia (1 980)
Timor-Leste	Australia (59)	Indonesia (19)	Portugal (17)	UK (16)	USA (13)
Tonga	Australia (27)	New Zealand (21)	Fiji (12)	USA (11)	France (8)
Tuvalu	France/USA (4)		Brazil/Fiji/Tonga/UK (3)		
Vanuatu	USA (34)	Australia (33)	France (21)	Canada (16)	Japan/UK (11)
Viet Nam	USA (2 462)	Japan (2 327)	Korea, Rep. (2 302)	Australia (1 881)	China (1 841)

# Collaboration partners

Southeast Asia and Oceania

UNESCO (2021)

## Helicopter science and loss of trust

“Neo-colonial research or neo-colonial science, frequently described as **helicopter research**, parachute science or research, parasitic research, or safari study, is when researchers from wealthier countries go to a developing country, collect information, travel back to their country, analyze the data and samples, and publish the results with no or little involvement of local researchers.” – Wikipedia

“In the medical world: "A popular term for a clinical or epidemiologic research **project conducted by foreign scientists who use local contacts to gain access to a population group and obtain samples**" – FreeDictionary.com

# Helicopter science and loss of trust

SCIENCEINSIDER | SCIENTIFIC COMMUNITY

## ‘Helicopter research’ comes under fire at Cape Town conference

Statement presses for equity when scientists from wealthy nations do research in poorer ones

6 JUN 2022 · 1:40 PM ET · BY CATHLEEN O'GRADY

NEWS FEATURE · 13 APRIL 2021

## Parachute science falls to earth

Researchers say more needs to be done to correct past injustices.

Clare Watson

PALEONTOLOGY

## Paleontology has a ‘parachute science’ problem. Here’s how it plays out in 3 nations

Legacies of colonialism can stifle science. But there are ways to rein it in

By Helen Thompson

JUNE 21, 2023 AT 9:00 AM

## Helicopter science and loss of trust

Examples in oncology:

- ❑ Clinical trials with exploitative practices have been run in low- and middle-income countries (LMICs), including Latin America
- ❑ Cancer drug approval in LMICs that have hosted clinical trials is much slower than that in HICs,
- ❑ In many cases, these drugs are also too expensive for patients in LMICs to access

Grover et al., 2016; Gyawali et al., 2022, Fundytus et al., 2021; Miller et al., 2021

## Considerations: Ethics for sample collecting

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- The human body and its parts should not give rise to direct financial gain
- Information provided to potential donors should be understandable and helpful

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- Research involving human biological material should undergo independent ethical review, with very limited exceptions
- Researchers should treat as confidential all personal and medical information
- Researchers should keep up to date with all ethical, legislative, regulatory and governance requirements

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- Should data be made public? What would be the benefits and risks?

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- What happens if discoveries from patient samples lead to commercial products or patents?
- Beyond individual consent, should community-level consultation or agreements be sought?

There is scarce funding going into research and development activities in the Global South. Scientific partnerships with HICs can be a great solution, but we must also be careful - 'helicopter science' practices can lead to loss of trust

# Outline

- ❑ Background: The importance of studying diverse populations to understand disease processes
- ❑ What is the problem?
  - ❑ Very little representation of non-European samples in public repositories
  - ❑ Very few studies being conducted outside de Global North
- ❑ Why does it exist?
  - ❑ Lack of support for studies being conduced in the Global South
  - ❑ ‘Helicopter’ science
- ❑ How can we solve it?
  - ❑ More equitable scientific partnerships
  - ❑ Facilitating access to funding
  - ❑ Expanding representation in scientific boards, conferences, etc
- ❑ Conclusions

## **How can we solve it?**

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Global South PIs are now eligible for many international funding calls

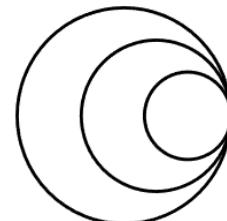


Chan  
Zuckerberg  
Initiative

UKRI

Melanoma  
Research Alliance

Medical  
Research  
Council



**wellcome**  
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**science**

The **Academy of**  
**Medical Sciences**

wellcome  
**sanger**  
institute



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# How can we solve it?

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WORLD CONFERENCES  
ON RESEARCH INTEGRITY  
Cape Town Statement



Joint commitment for action on  
inclusion and diversity in publishing

## The TRUST Code

A Global Code of Conduct for Equitable Research Partnerships

BMJ Global Health



Research  
Fairness  
Initiative

Bridging research integrity and global  
health epidemiology (BRIDGE)  
statement: guidelines for good  
epidemiological practice

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- ❑ Facilitate publishing of Global South-led research
  - ❑ Journal discounts
  - ❑ Language
- ❑ Increase representation of Global South scientists at all levels (in research labs, in publishing, in conferences)

## How can we solve it?

- Channel more funding to Latin American-led teams
- Promote (and demand) equitable research partnerships
  - Capacity building
  - Training opportunities both ways
  - Recognition of experience and value
- Support Latin American (and LMIC) partners so they can meet data availability requirements
- Facilitate publishing of Latin American (and LMIC)-led research
  - Journal discounts
  - Language



A higher representation of Global South science at all levels (project leadership, published studies, scientific publishing, research conferences) will result in a higher representation of non-European samples in public repositories

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## Conclusion

Supporting Global South researchers will increase the contribution that they make to research outputs, which will, in turn, increase the study of samples from these regions and thus the diversity of data hosted in large repositories.

This should be done in collaboration with international partners from the beginning and considering the needs and benefits for everyone involved

**Thank you!**