Race, Ancestry, Identity, and Genetics
Part 2

Learning Goals

1. Describe societal perceptions of social identity (race, ethnicity, and nationality) and genetic ancestry, including racial essentialism.
2. Examine the consequences that these societal perceptions of social identity and genetic ancestry have on individuals and society broadly.
3. Critique scientific uses of social identity (race, ethnicity, and nationality) and genetic ancestry

Ground norms

- Be respectful of conflicting opinions
- Commit to learning and growing
- Compassionate listening
- No talking over anyone
- Make space, take space
- What is said in the room, stays in the room (confidentiality)
- Use "I"statements for sensitive topics
- Don't equate people with stereotypes
- Don't rush to judge others

Race, genetics, and medicine

[Dorothy Roberts Video](#)

10 minutes - Small group discussion (groups of 4)

- What and/or how were you taught about racial health disparities?
- Was this education on racial health disparities focused more biological explanations versus social explanations?
- How does this connect to what we discussed in class last week?

5 minutes - Large Group Discussion/Sharing

Discussion questions to be considered:

- What and/or how were you taught about racial health disparities?
- Was this education on racial health disparities focused more biological explanations versus social explanations?
- How does this connect to what we discussed in class last week?

Review - Social Identity and Genetic Ancestry

Definitions

Race: socio-political mechanism for classifying individuals; often tied to status and power; relies on ideology or inequality

Ethnicity: cultural construct often linked to community, religion, language, etc.

Genetic Ancestry: biological inheritance of DNA, can be traced through the genome using genotype data

Nationality: place of legal organization, ownership, citizenship, or lawful permanent residence (or equivalent immigration status to live and work on a continuing basis) of suppliers of commodities and services.

Social Identity (Theory): social categorization and self-conception that explains how people represent social categories and their associated attributes govern how they behave and how they conceive of themselves.

Popejoy, A. (2020, September). Information Disparities and Implications for Clinical Genetics. Lecture conducted from at Stanford University, United States.

Cornell Law (2023)

Hogg, M. A., & Rinella, M. J. (2018). Social identities and shared realities. Current opinion in psychology, 23, 6–10.

Recap on genetic ancestry

1. There are small, subtle shifts in the frequencies of genetic variants across geographic distances
2. It is possible to cluster individuals into groups using genetic information
3. The majority of genetic variation comes from differences between individuals and not between populations or races

Discussion of ancestry testing "surprises"

Sigrid Johnson Was Black. A DNA Test Said She Wasn't.

Questions to consider

- How is genetic ancestry determined in this setting? How is it related to social identity and how is it distinct?
- What questions or points of confusion arise for you?
- Do you think social identities can be determined by the results of genetic ancestry tests? What about the social identities you personally hold?

Activity timing

- 10 min independent reading
- 5 min small group discussion
- 5 min large group share-out

Population classification in genetic research

Historical population classification was consistent with the European belief that race is a biological reality

1737: Carl Linnaeus' Systema Naturae

I. QUADRUPEDIA.

Corpus hirsutum.   Pedes quatuor.   Feminæ viviparæ., lactiferæ.

ANTHROPO-MORPHA. *Dentes* primores 4. utrinque: vel nulli.	Homo.	Nosce te ipsum.	H { Europæus albesc. Americanus rubesc. Asiaticus fuscus. Africanus nigr.
	Simia.	ANTERIORES. POSTERIORES. *Digiti* 5. . . . . 5. Posteriores anterioribus similes.	Simia cauda carens. Papio. Satyrus. Cercopithecus. Cynocephalus.
	Bradypus.	*Digiti* 3. vel 2. . . 3.	Ai. *Ignavus.* Tardigradus.

1919: one of the earliest studies comparing genetic variation between groups

SEROLOGICAL DIFFERENCES BETWEEN THE BLOOD OF DIFFERENT RACES.

THE RESULT OF RESEARCHES ON THE MACEDONIAN FRONT.*

BY DR. LUDWIK HIRSCHFELD,
DOZENT AT THE UNIVERSITY OF ZURICH;

AND

DR. HANKA HIRSCHFELD,
OF THE CENTRAL BACTERIOLOGICAL LABORATORY, ROYAL SERBIAN ARMY.

English ...
French ...
Italians ...
Germans...
Austrians
Serbs... ...
Greeks ...
Bulgarians
Arabs ...
Turks ...
Russians...
Jews... ...
Malagasies
Negroes (Senegal)
Annamese
Indians ...

Inconsistencies and uncertainties in population usage persist in modern-day research

" persistent and indiscriminate blending of classification schemes… has led the practical definition of 'population' to become more ambiguous rather than standardized over time. "
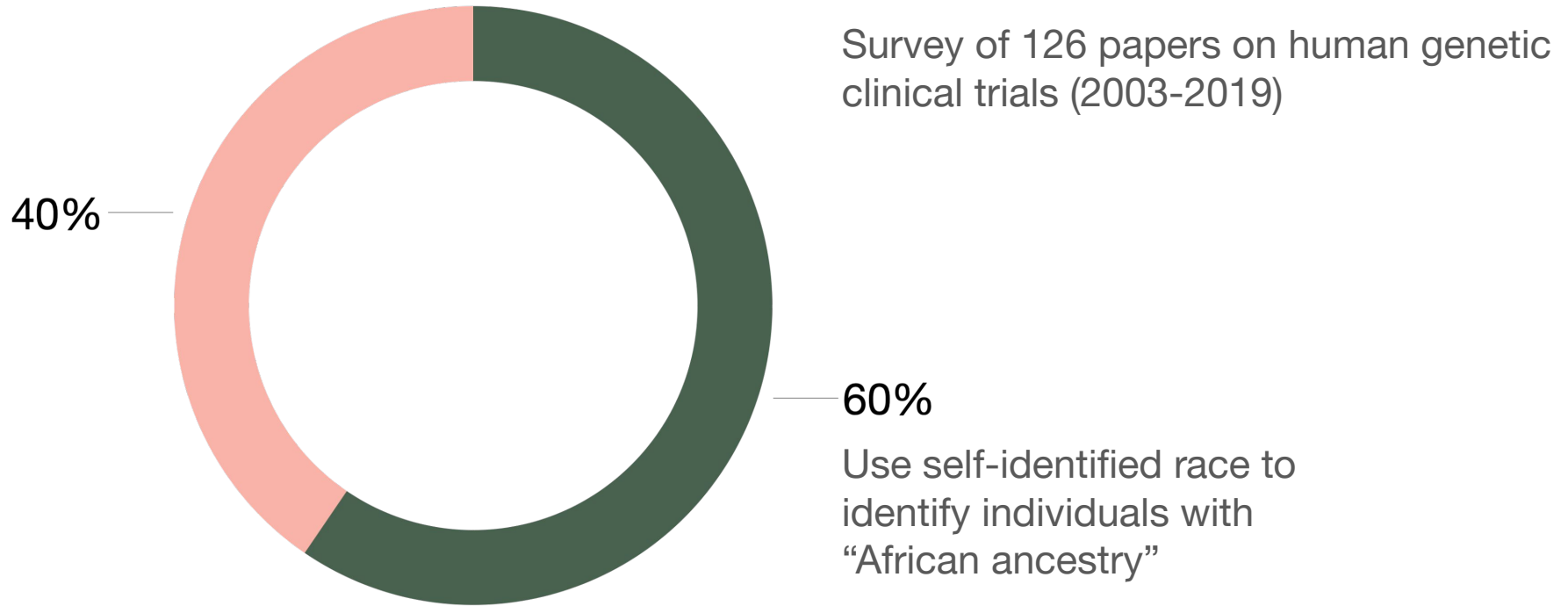
Panofsky and Bliss 2017

Ambiguity in classification often risks reifying the concept of biological race
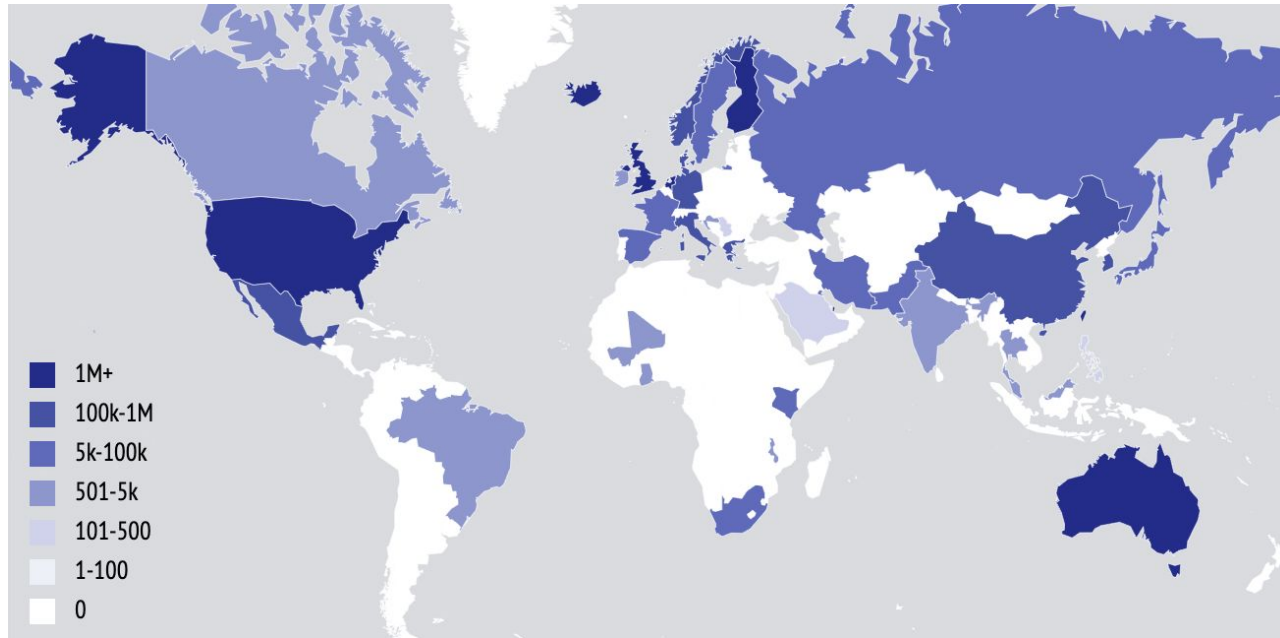
Table 1. Classification Systems and Codes

Code	Classification System	Examples
1	U.S. Census (Race)	White or Caucasian, Black or African ... etc. (Note that the U.S. Census syste... categories. We consider all of these ... classification system.)
2	Continent	European, African
3	Continental region	Northern European, West African
4	Country	Netherlands (or Dutch), Japanese
5	Country region	Western United States, Sicilian, Austr...
6	Ethnicity	Bedouin, Han
7	Language	Bantu speakers
8	Other	Usually religion: Jewish, Druze, Amish...

Ethnic group	Self-reported ethnic background
White	
	British
	Any other white background
	Irish
	White
Asian or Asian British	
	Indian
	Pakistani
	Any other Asian background
	Bangladeshi
	Asian or Asian British
Black or Black British	
	Caribbean
	African
	Any other Black background
	Black or Black British
Chinese	
	Chinese
Mixed	
	Any other mixed background
	White and Asian
	White and Black Caribbean
	White and Black African
	Mixed
Other/Unknown	
	Other ethnic group
	Not stated

UK Biobank

)17

Ambiguity in classification often risks reifying the concept of biological race

40%

Survey of 126 papers on human genetic clinical trials (2003-2019)

60%

Use self-identified race to identify individuals with "African ancestry"

Duello et al. 2021

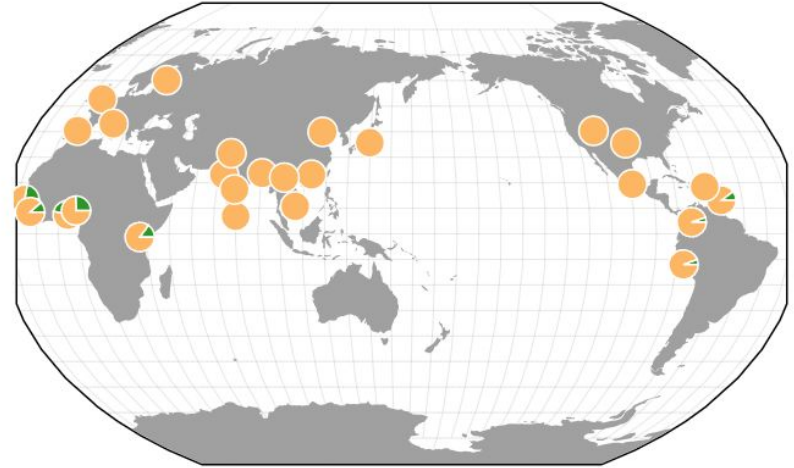# Diversity of individuals in genetic studies is generally pretty abysmal

Why is diversity in genetic datasets important for research?
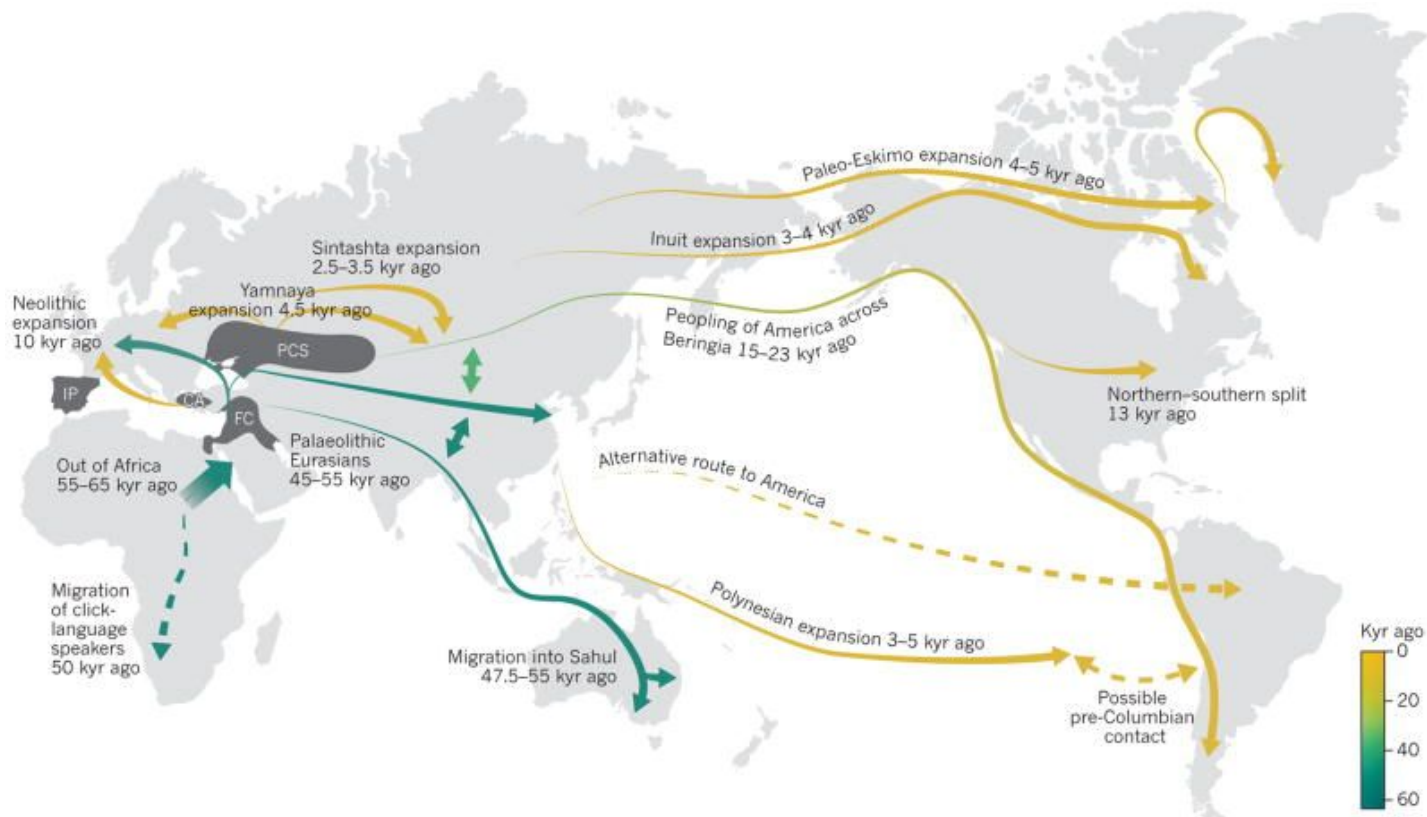
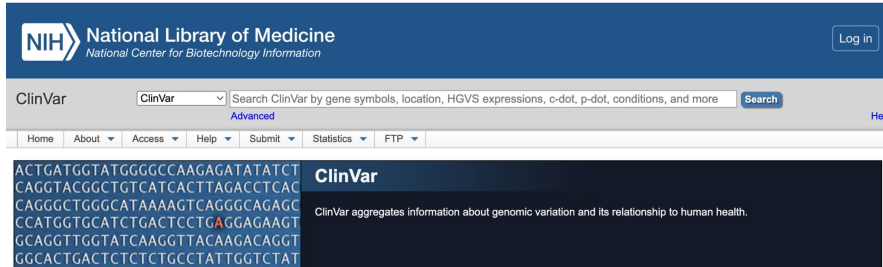# Why is diversity in genetic datasets important for research?

Variants are hard to identify as disease-causing when they are **rare**.

Because frequencies of genetic variants vary across populations, we are less able to identify the most relevant disease-causing variants in understudied populations.

Marcus & Novembre 2016
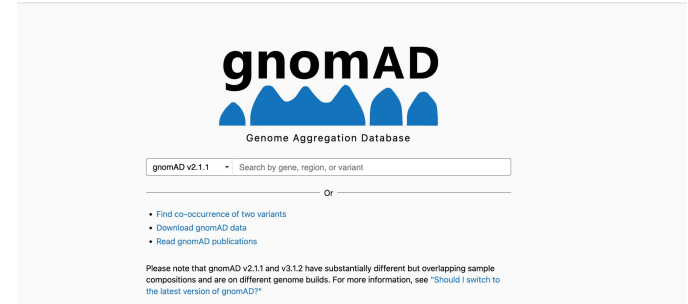
Why is diversity in genetic datasets important for research?

Lack of representation of minority populations in clinical databases

The majority of clinical population databases have genetic data from white populations.

Middle Eastern and Pacific Islander populations are not represented in these databases, making it hard to classify these genetic variants in relation to disease.

Why is diversity in genetic datasets important for research?

Most large-scale genetic studies (GWAS) identify variants that are **correlated** with a disease.

Many of these variants are not disease-causing – they are simply correlated with the actual disease-causing variants.

Patterns of correlations vary across geographic space.

This means genetic studies in Europeans will only find variants that are really good proxies for disease risk **in Europeans**.

Europe

non-Europe

Takeaway: there are no easy answers

1. "Populations" are societally constructed labels.
2. Treating "populations" as biologically objective plays into a long history of race science.
3. But… genetic similarity and dissimilarity matters. Diversity in research datasets matters.
4. In some cases researchers might need to use "populations" as an imperfect proxy for genetic similarity. In most cases they should try to focus on genetic similarity without ascribing societally constructed labels.

We should constantly be interrogating how populations were defined and why they are being studied.

Discussion of population classification in scientific articles

Association Between Genetic Risk for Psychiatric Disorders and the Probability of Living in Urban Settings

Jessye M. Maxwell, MSc; Jonathan R. I. Coleman, PhD; Gerome Breen, PhD; Evangelos Vassos, MD

Questions to consider

- Can you figure out which 'populations' were included in this analysis? Which were excluded?
  - Why? Was there a justification provided?
- How did they define 'populations' (or did they?)
- How easy was it to find this information?
- What impact might these inclusions/exclusions have on the reader?

Activity timing

- 10 min independent reading
- 3 min partner discussion
- 7 min large group discussion

Discussion of race and genetics in society

Why White Supremacists Are Chugging Milk (and Why Geneticists Are Alarmed)

Article:
https://www.nytimes.com/2018/10/17/us/white-supremacists-science-dna.html

Activity timing

- 10 min independent reading
- 10 min small group discussion
- 10 min large group discussion

Questions to consider:

- What diversity measures are relevant to this case study (e.g., race, ethnicity, ancestry)?
- What are (potential) misconceptions about race and ancestry that emerged in this case example?
- Why could these (potential) misconceptions be harmful? What are the risks this case example raises?
- What are researchers' responsibilities to mitigate risks in this case?

Session 5: Behavior and Genetic Determinism

Recognize how genetics research can be mis-used to justify harmful, genetically determinist viewpoints and the associated harms

Describe current efforts by the research community to combat the weaponization of research, and articulate strategies that individual scientists can employ

Taught by
Alvina Adimoelja (she/her)

Taught by
Naiomi Hunter (she/her)