



Whose Genomes Matter?

Alice B. Popejoy

PhD Candidate

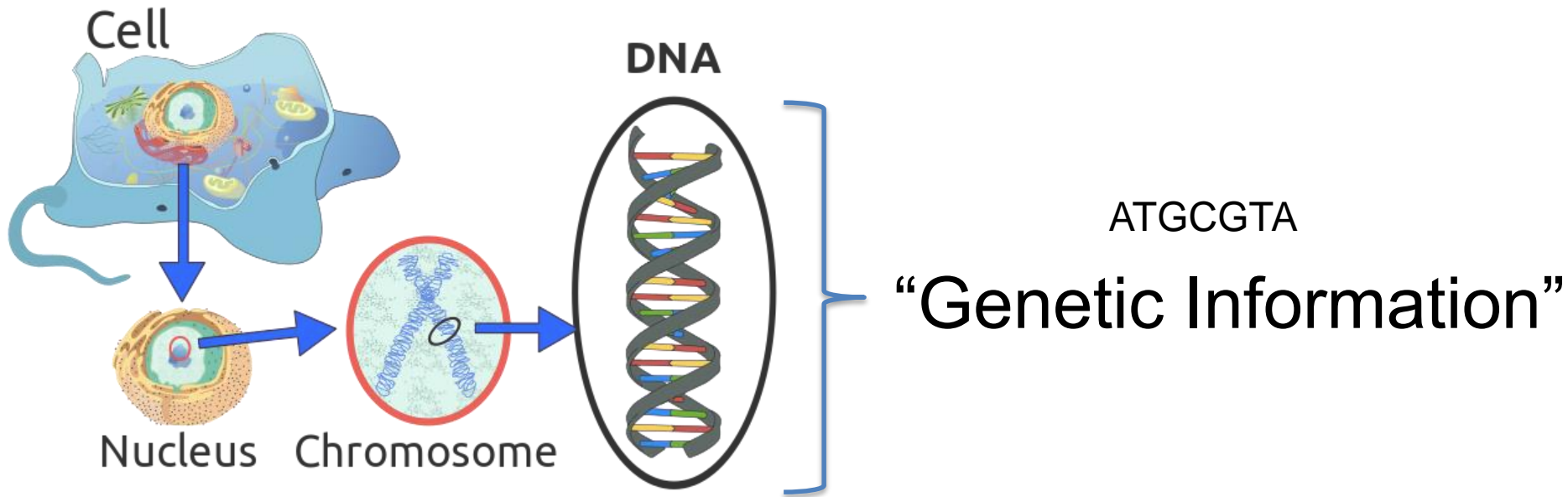
Institute for Public Health Genetics

“Precision Medicine”

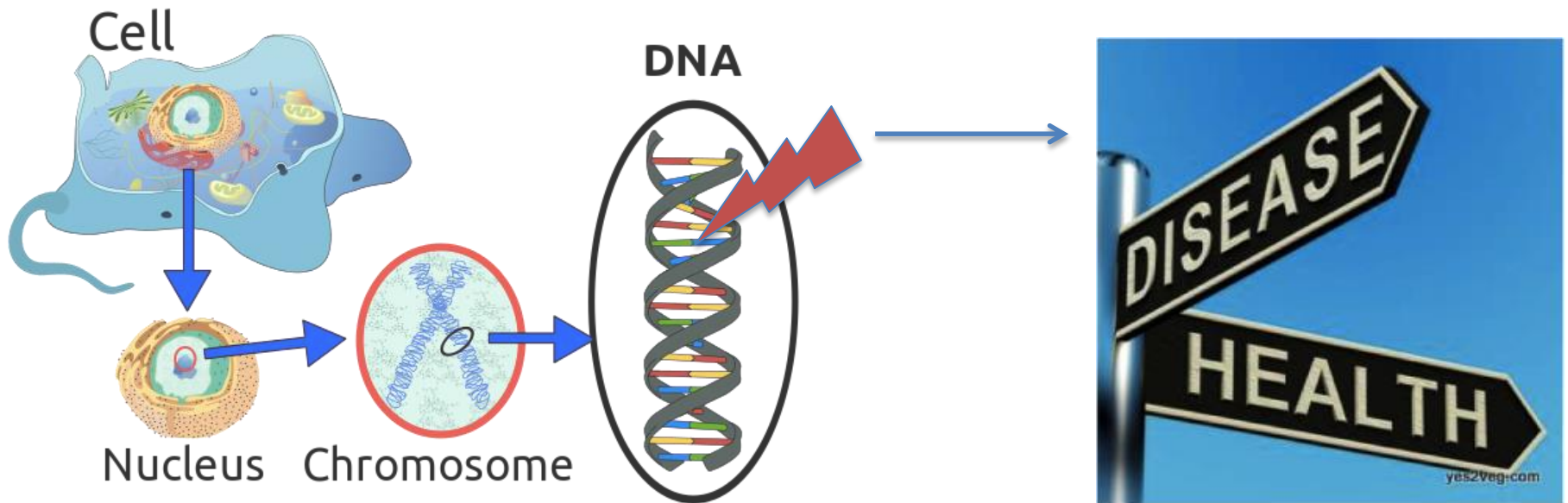


- Who will benefit?
- Who might be harmed?
- Will there be whole populations left out?

Genome-Wide Association Studies (GWAS)

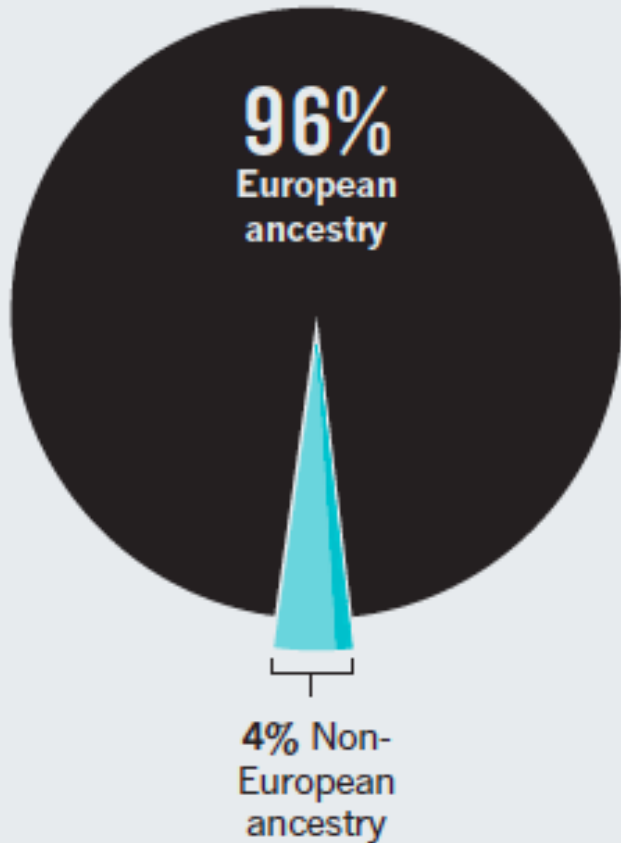


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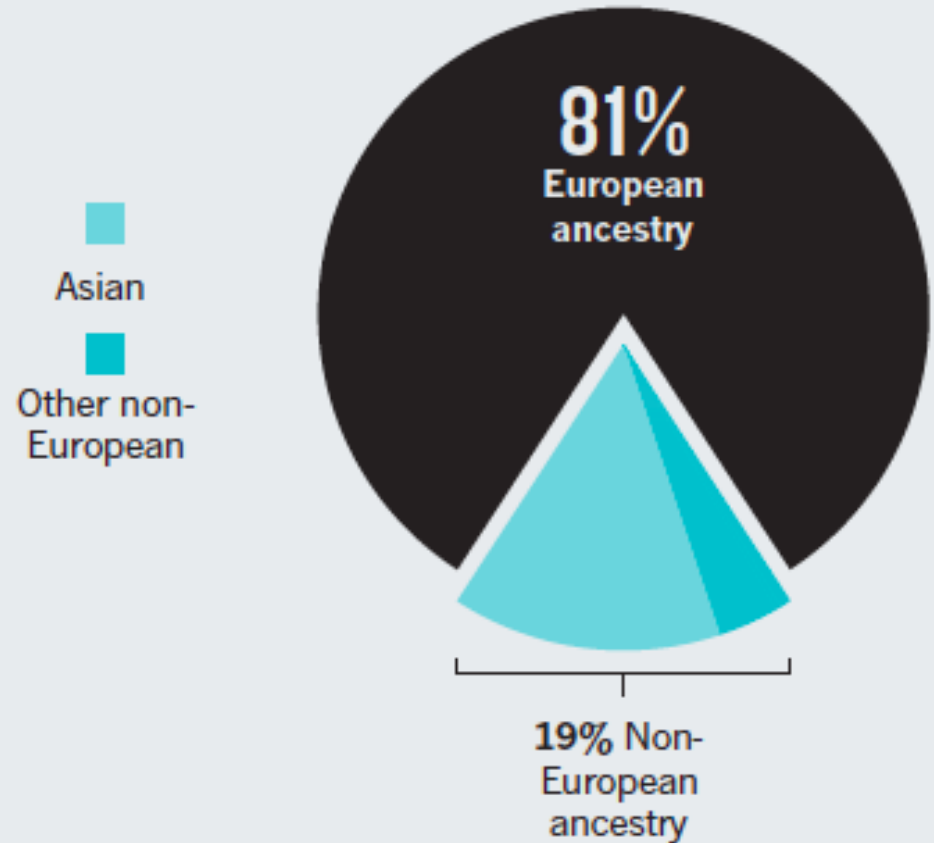


Tool for basic discovery in disease biology

2009
373 studies
1.7 million samples



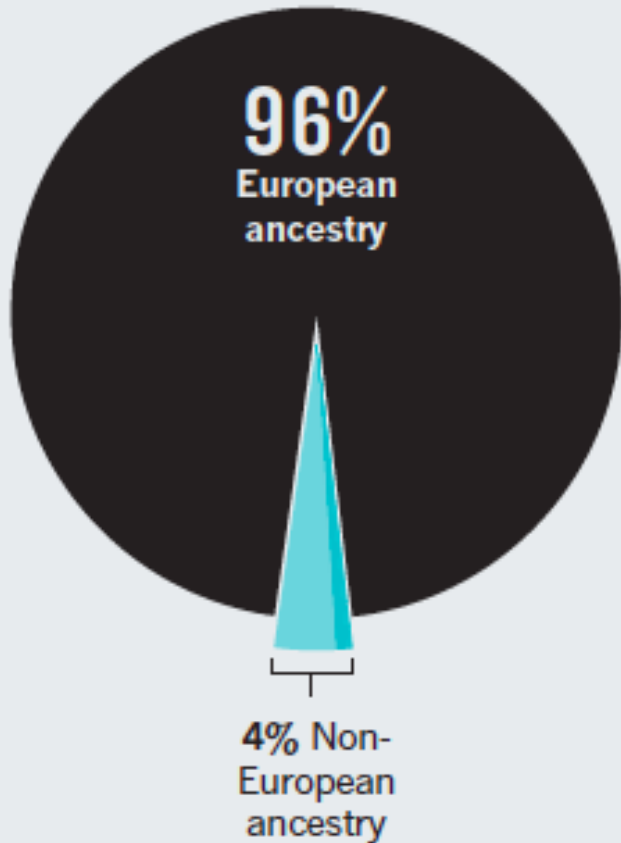
2016
2,511 studies
35 million samples



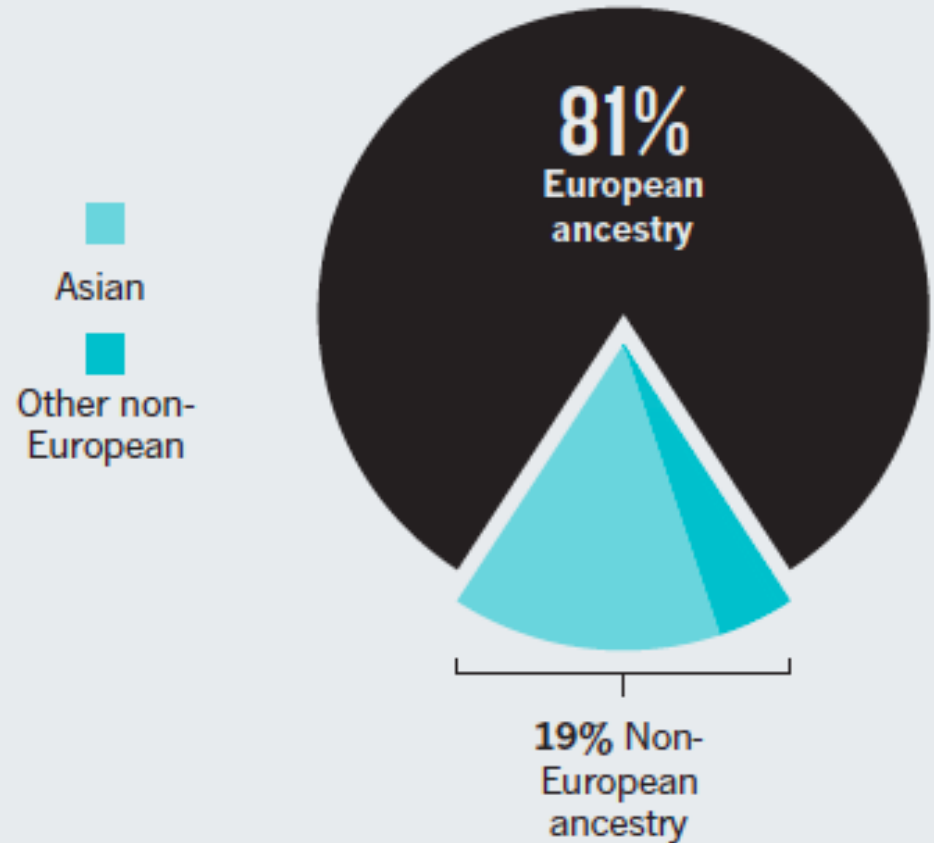
■ Asian
■ Other non-European

“Genomics is failing on diversity” (Popejoy & Fullerton, *Nature* 538: 2016)

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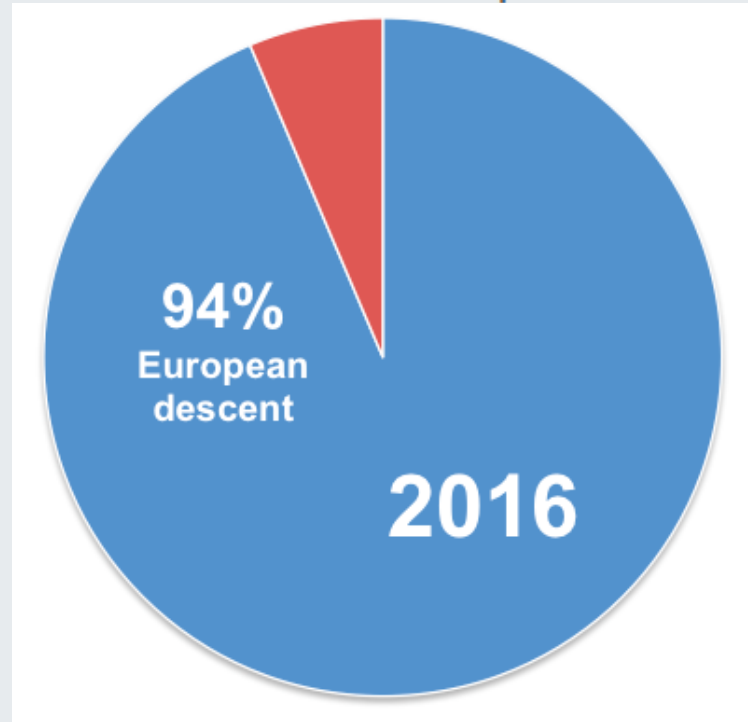
96%
European
ancestry

4% Non-
European
ancestry

Asian
Other non-
European

2016

2,511 studies
35 million samples



94%
European
descent

2016

< 4% African American,
Hispanic/Latino or Native American

(Popejoy & Fullerton, *Nature* 2016)

Why is this a problem?



**ALL
HUMANS
ARE
99.9%
IDENTICAL**

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- Different ancestry means different genetic “background variation”

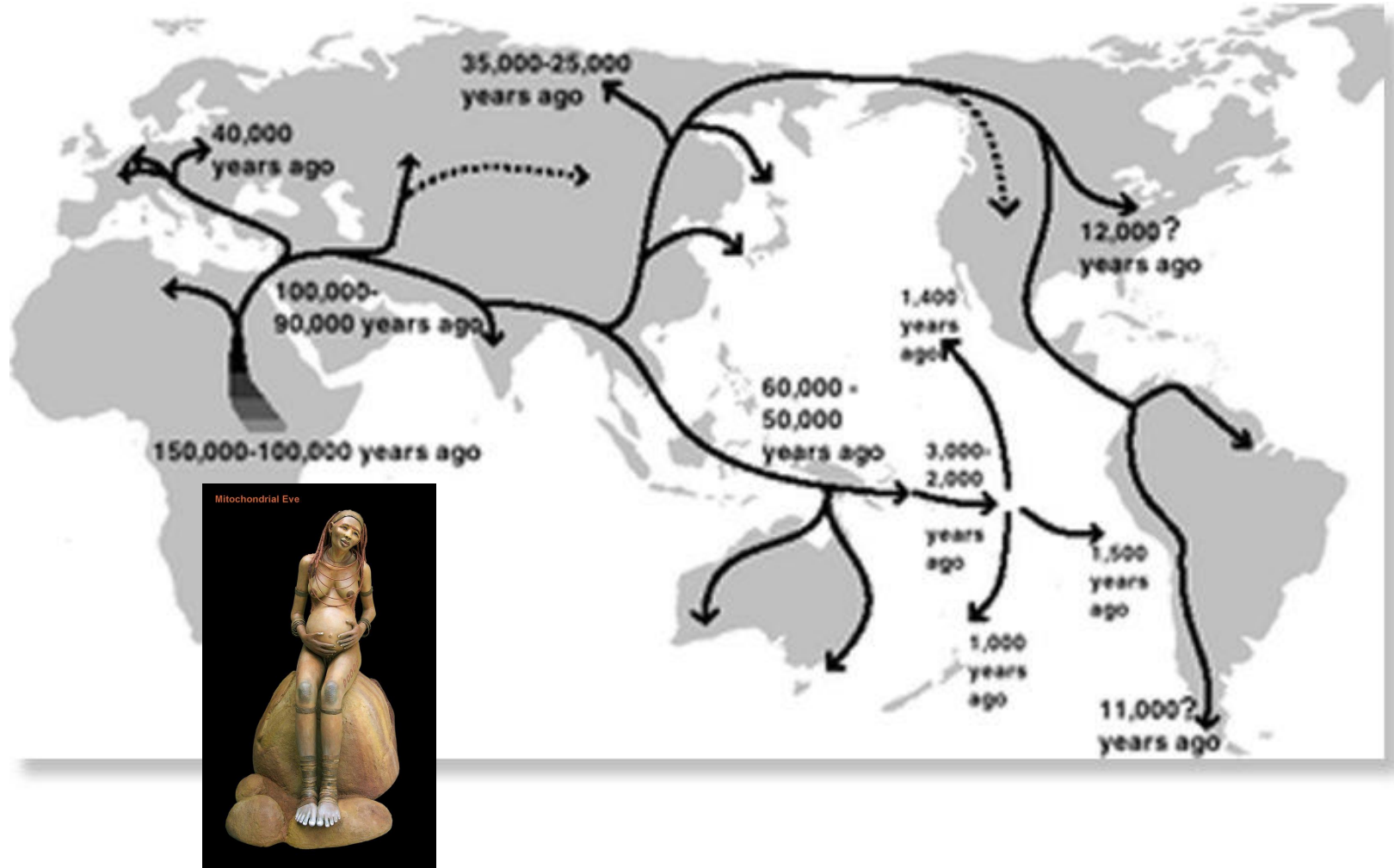
Ancestry \neq Race

- Race: social, cultural, self-identity
 - NOT biological
 - Only relevant to genomics as *racism* can cause bias in disease studies in populations
- Ancestry: biological inheritance of DNA
 - Can be traced through parts of the genome
 - Impacts background genetic variation

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All Genomes Out of Africa



Why So Biased?

- Scientific justifications
 - Simplicity, homogeneity
- Historical recruitment practices
 - Biased toward white communities
- Logistical convenience
 - Who has access to medical care

Why So White?

- Systemic racism and inequality.
- Science
 - Biological theory of race
- History
 - Abuse of minority populations
- Logistics
 - Disproportionate lack of access

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Health Disparities & Racism

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- Most vulnerable and underserved populations:
 - African American
 - Hispanic/Latino
 - Native American

Genomics & Society

- Research doesn't occur in a vacuum!

< 4%

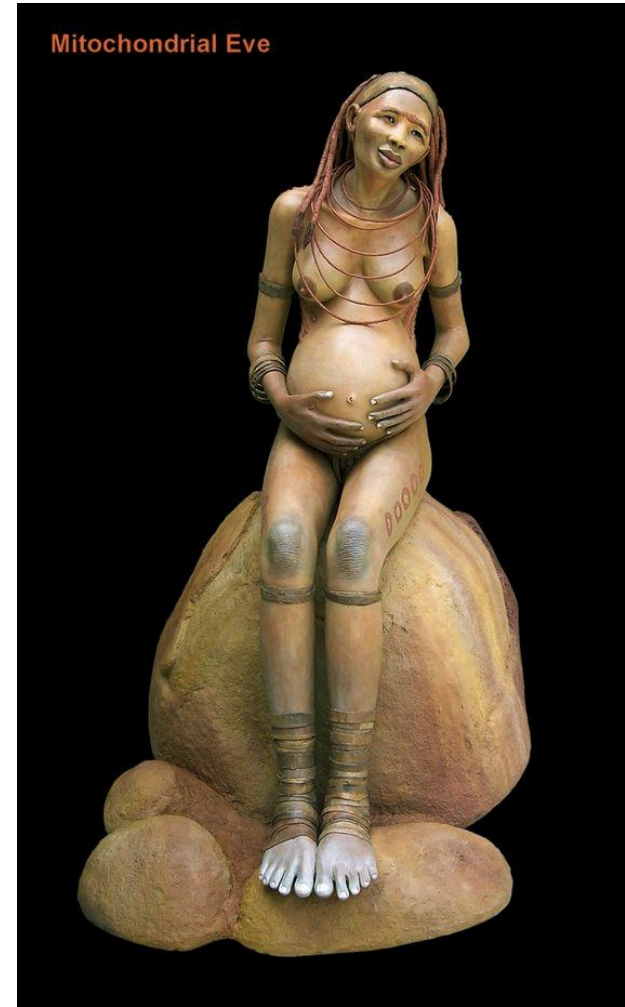
Genomics research
participants of color

< 4%

Genomics research
faculty of color

(Leboy & Madden, 2012)

Whose Genomes Matter?



Time's Up!



- **About you:**

- **Name: Alice B. Popejoy**
- **Department: Public Health Genetics**
- **Contact Information: popejoy@uw.edu**
- **Twitter/URL: @AlicePopejoy**
- **Quick bio: Public Health Geneticist, statistical & population genetics enthusiast, scholar of light receptors, social justice warrior, citizen of the world, parkour coach.**