

CONSUMER GENOMICS: Diversity and Potential for Disparities



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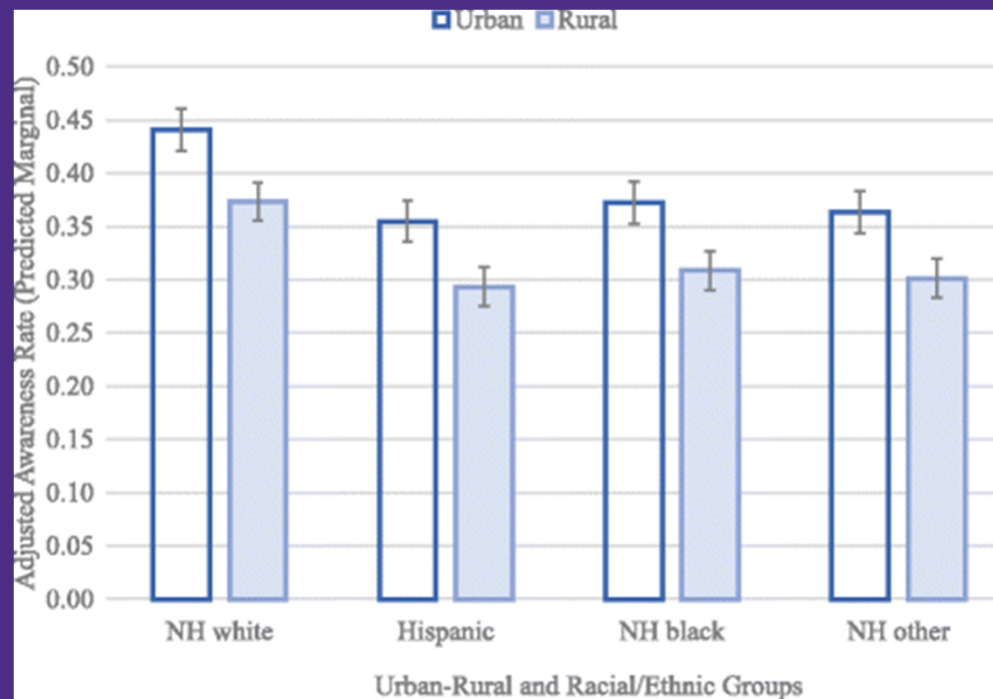


OVERVIEW

- > **Skewed evidence base matters for test performance**
- > **Underrepresented populations are more likely to be affected**
 - Lack of benefit due to uncertain results
 - Potential harms due to false positive or negative results
- > **Cannot wait for the evidence base to catch up**

W

Awareness of DTC Testing

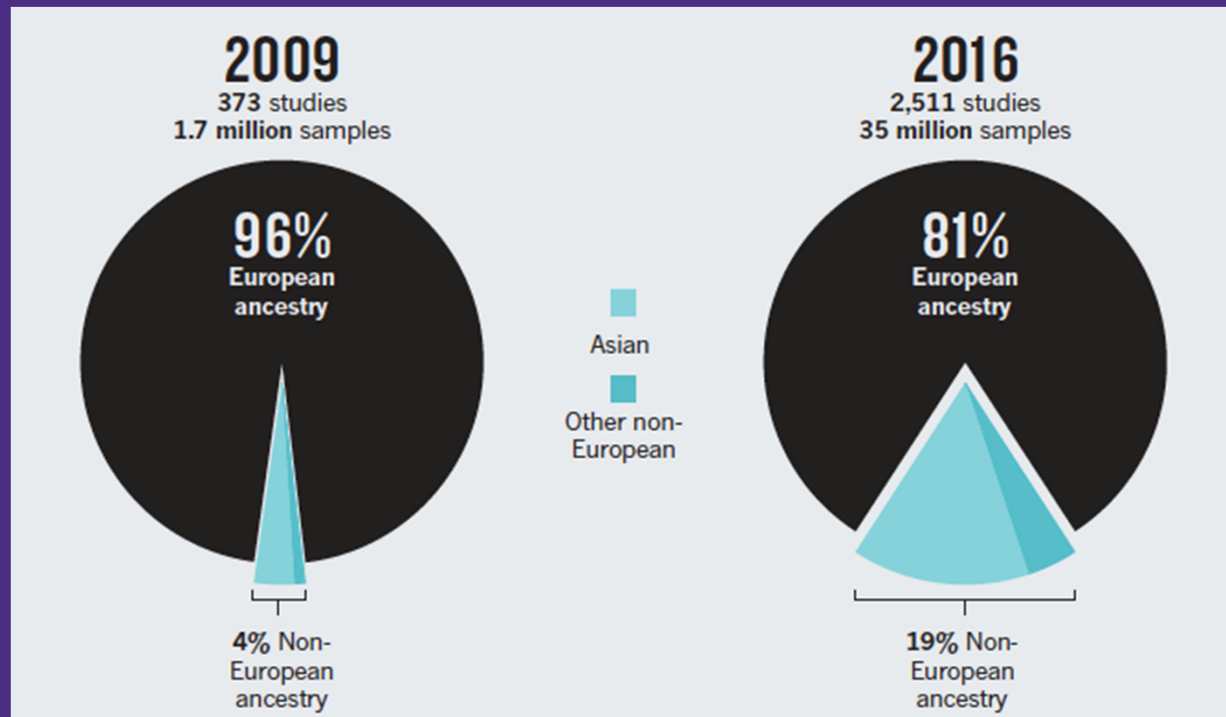


Salloum et al. (2019) *BMC Publ Health*

Even if ALL Awareness and Access
Differences Disappeared
Tomorrow...

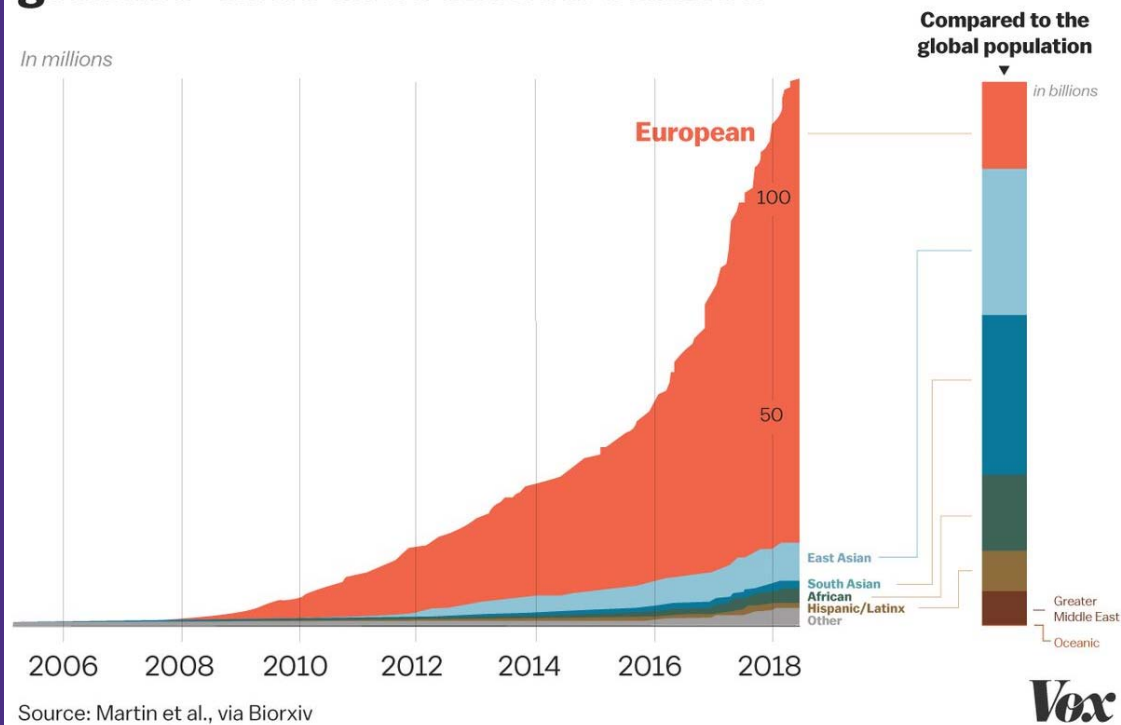
There Would STILL BE Disparities

Research Evidence Skews White



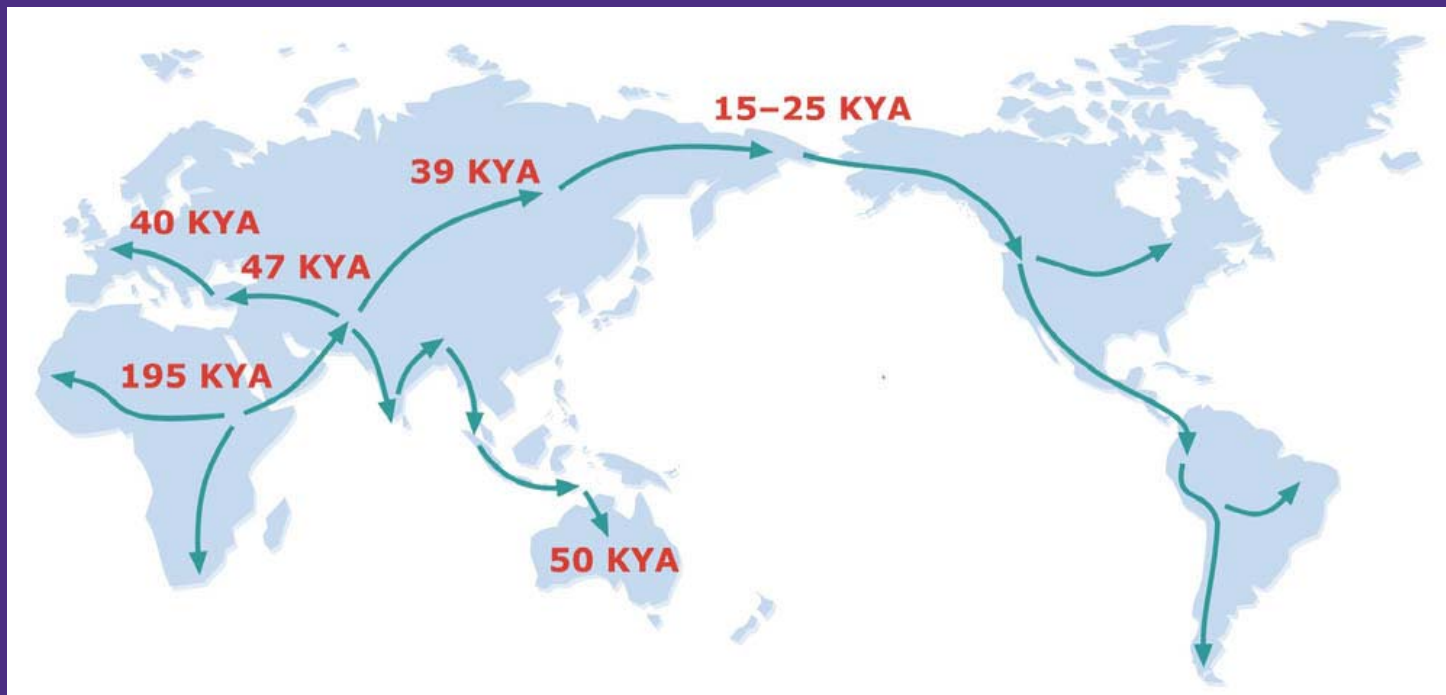
Popejoy & Fullerton (2016) *Nature*

Racial breakdown of participants in genome-wide association studies



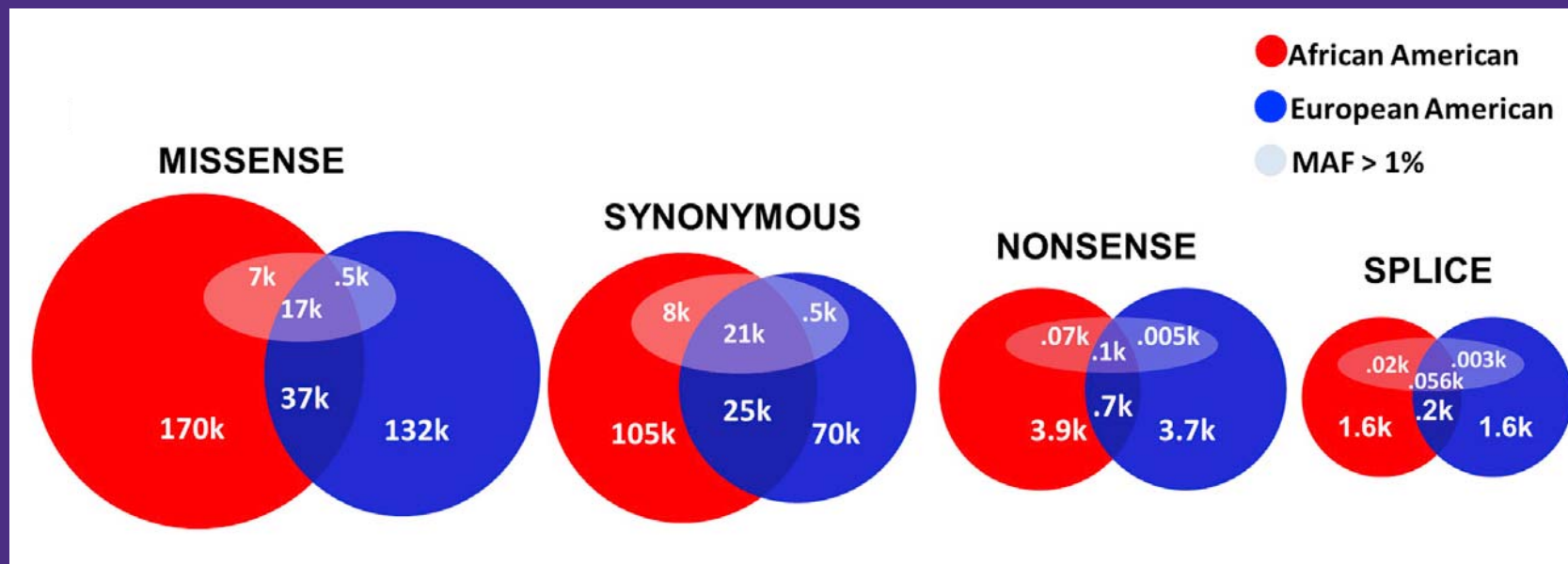
Resnik, Vox, 27 October 2018

This Matters Because Populations Vary



<https://www.sanger.ac.uk/research/projects/humanevolution/>

And Rare Variation is Less Often Shared

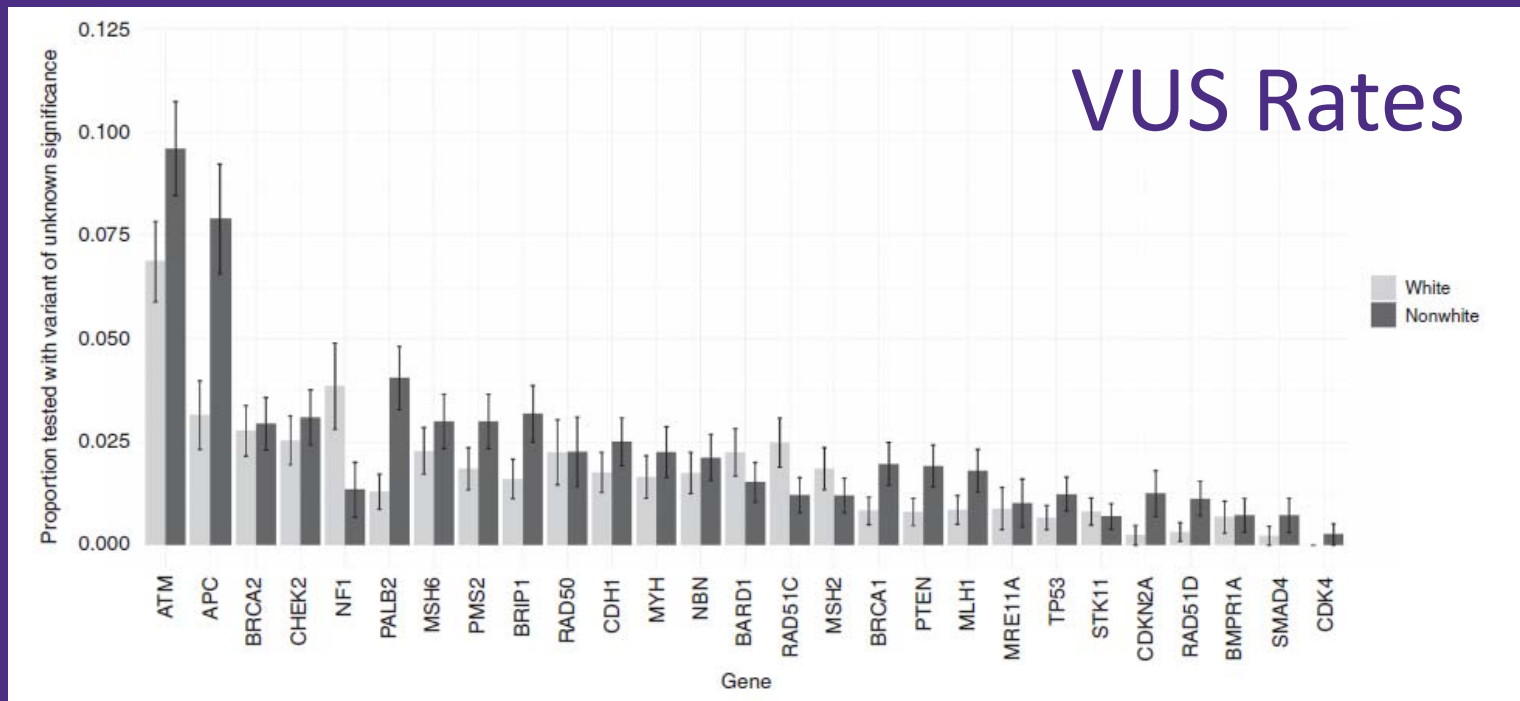


Auer et al. (2016) *Am J Hum Genet*

Same Test Different Answer

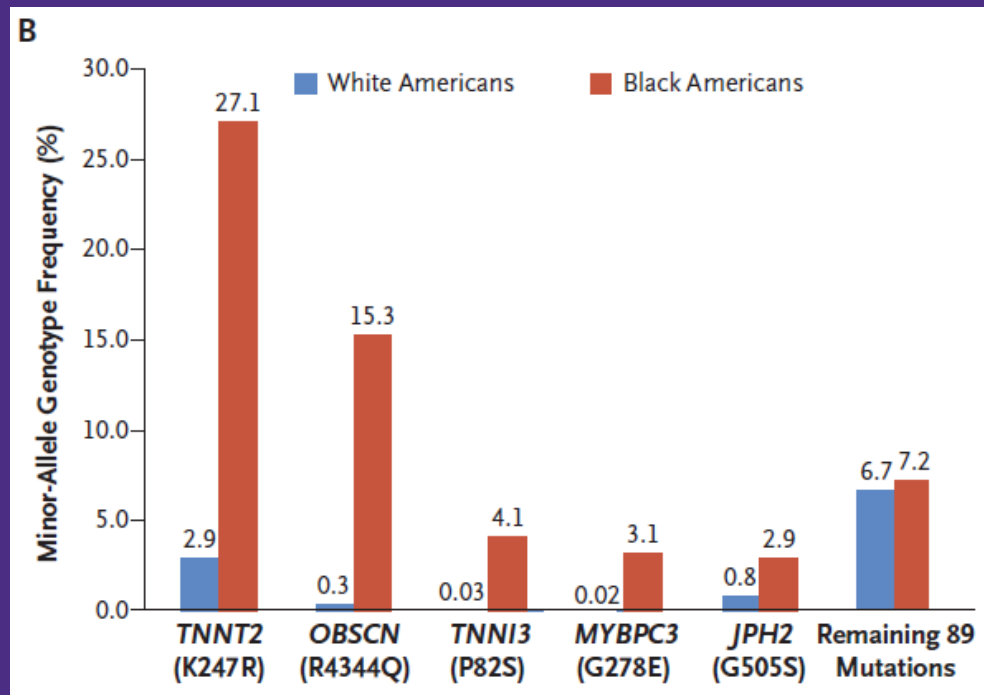
(not always, but more often)

Uncertain Results



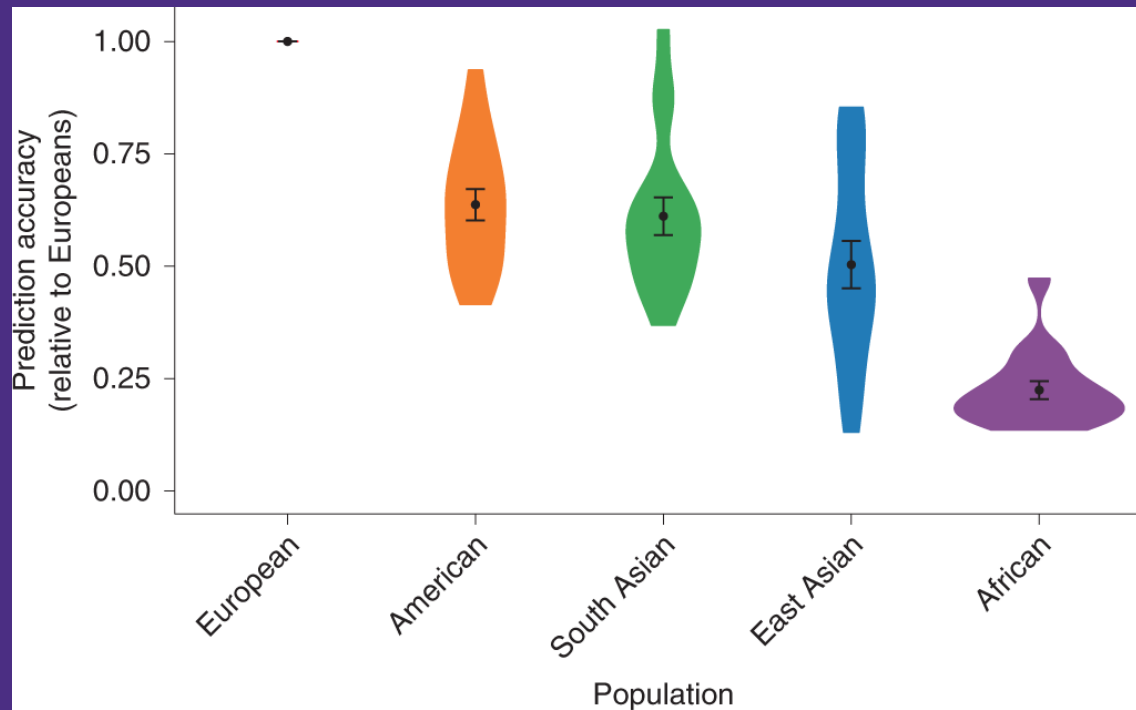
Caswell-Jin et al. (2017) *Genet Med*

False Positive Results



Manrai et al. (2016) *NEJM*

False Negative Results



Martin et al. (2019) *Nat Genet*

Third Party Interpretation?



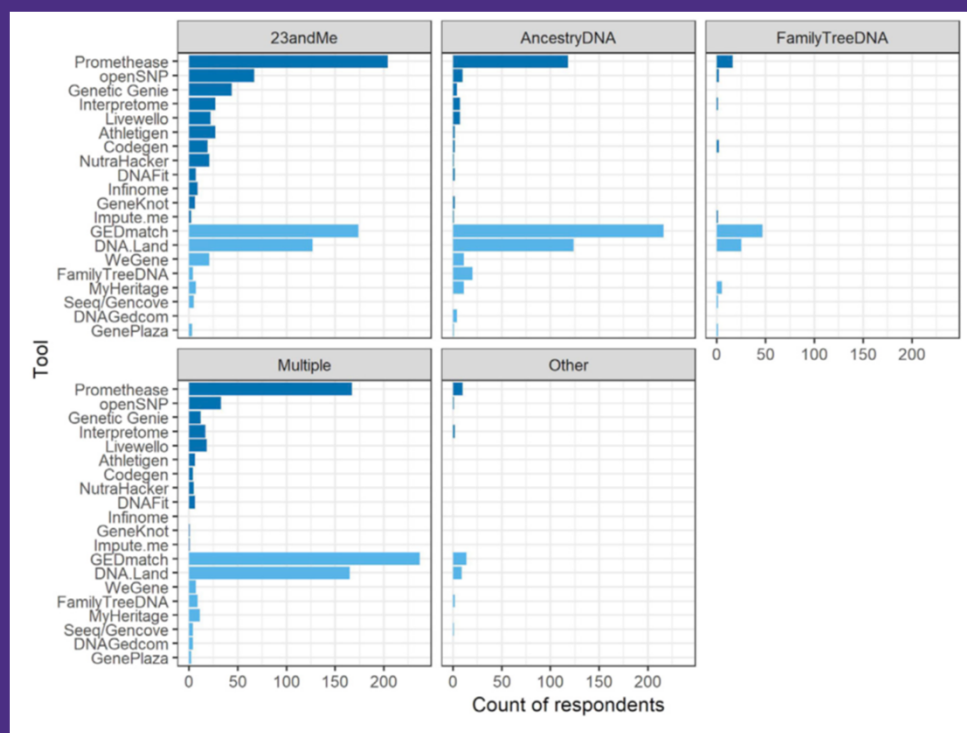
This data file generated by 23andMe at: Tue Nov 26 21:21:54 2013
Below is a text version of your data. Fields are TAB-separated
Each line corresponds to a single SNP...
rsid chromosome position genotype
rs4477212 1 82154 AA
rs3094315 1 752566 AA
rs3131972 1 752721 GG
rs12124819 1 776546 AA
rs11240777 1 798959 AG
rs6681049 1 800007 CC
rs4970383 1 838555 CC
rs4475691 1 846808 CC



Raw Data File

Courtesy of S. C. Nelson

Consumers Use MANY Different Tools

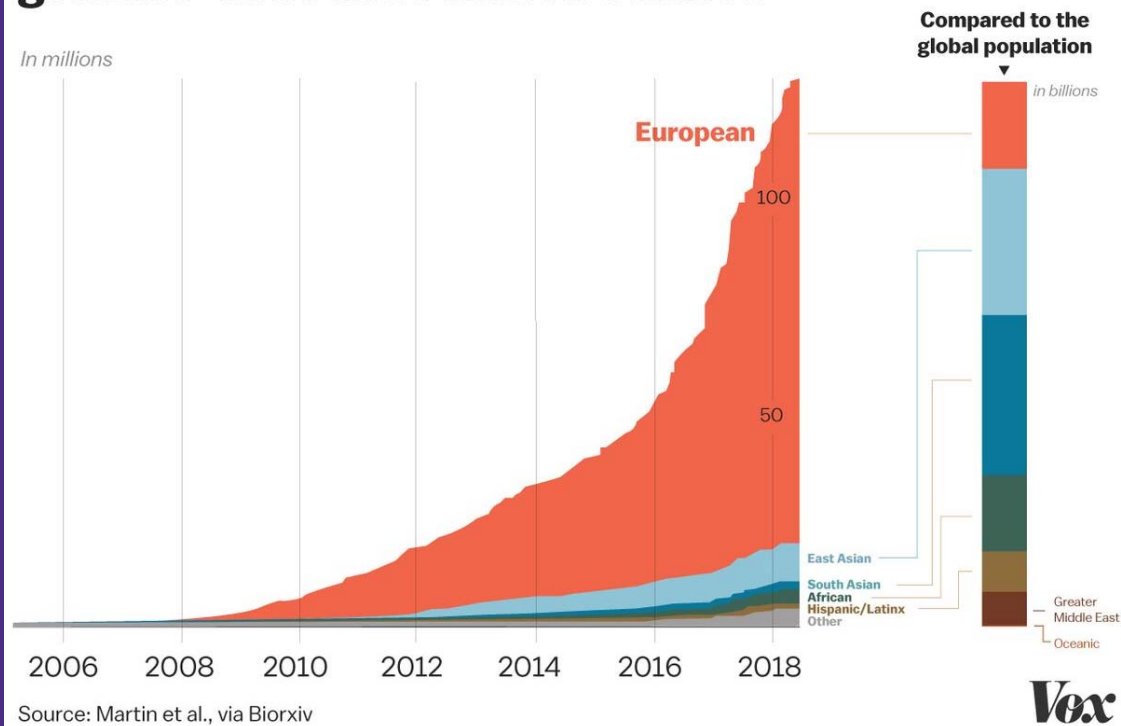


Health Related

Ancestry/Genealogy
Related

Nelson et al. (2019) *AJHG*

Racial breakdown of participants in genome-wide association studies



Solutions?

- Analytic (algorithmic) adjustment
- Minimum evidence standards for inclusion of genes on gene panels
- Greater transparency about limitations, especially for underrepresented groups

SUMMARY

- > **Skewed evidence undercuts the value of all genetic testing, including DTC genetic testing**
- > **Even with attention to access, URM populations will less often benefit and more often be harmed**
- > **Analytical approaches that can be implemented even as we rectify data biases are urgently needed**

W

Thanks To

Achieving Justice in Genomic Translation

Re-Thinking the Pathway to Benefit

The eMERGE Network

electronic Medical Records & Genomics

*A consortium of biorepositories linked to electronic medical records data
for conducting genomic studies*

Wyllie Burke
Kelly Edwards
Sara Goering
Suzanne Holland
Susan Brown Trinidad

OXFORD



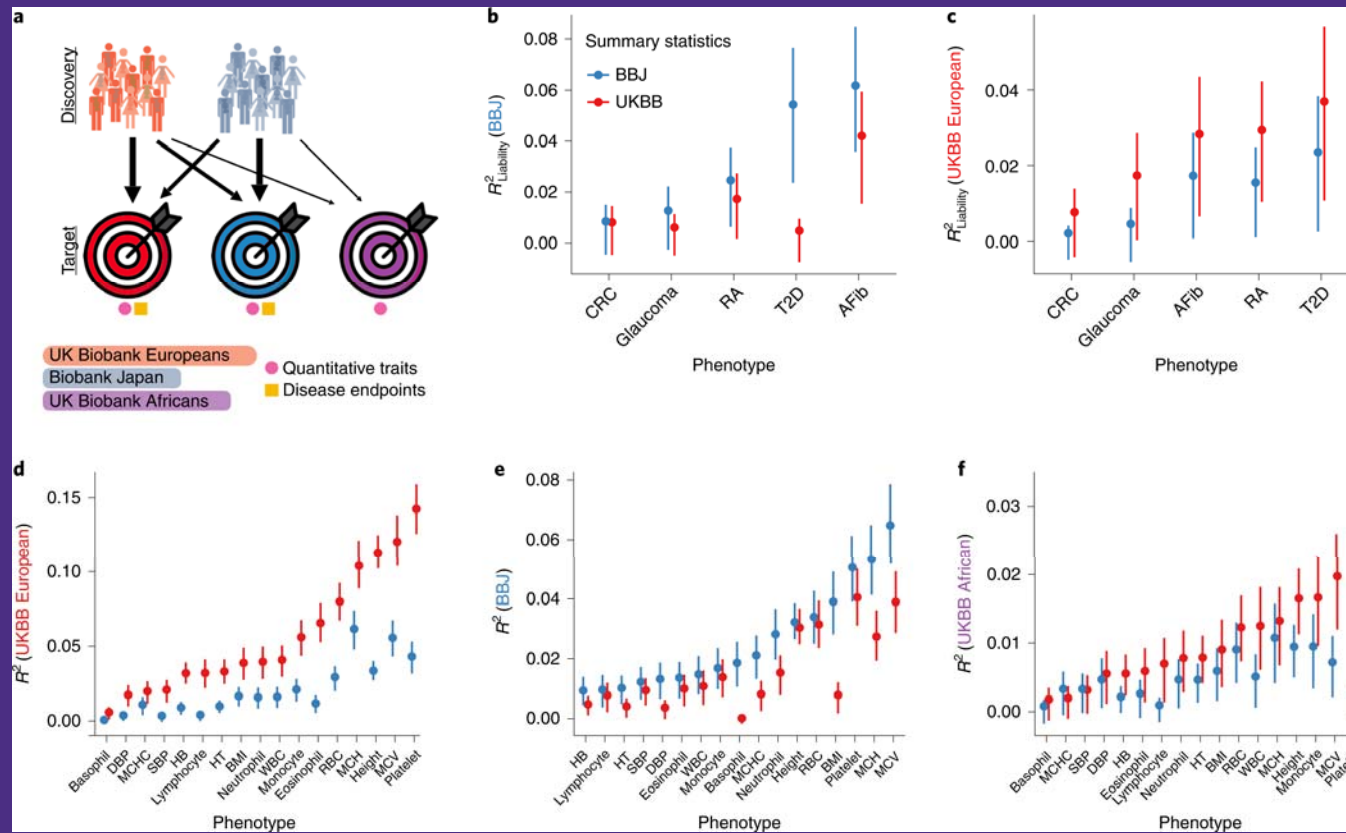
cser

Clinical Sequencing
Evidence-Generating
Research



University of Washington
Center for Genomics & Healthcare Equality





Martin et al. (2019) *Nat Genet*