

# Genetic Ancestry Testing: Implications for Diversity and Communication about Health Risk

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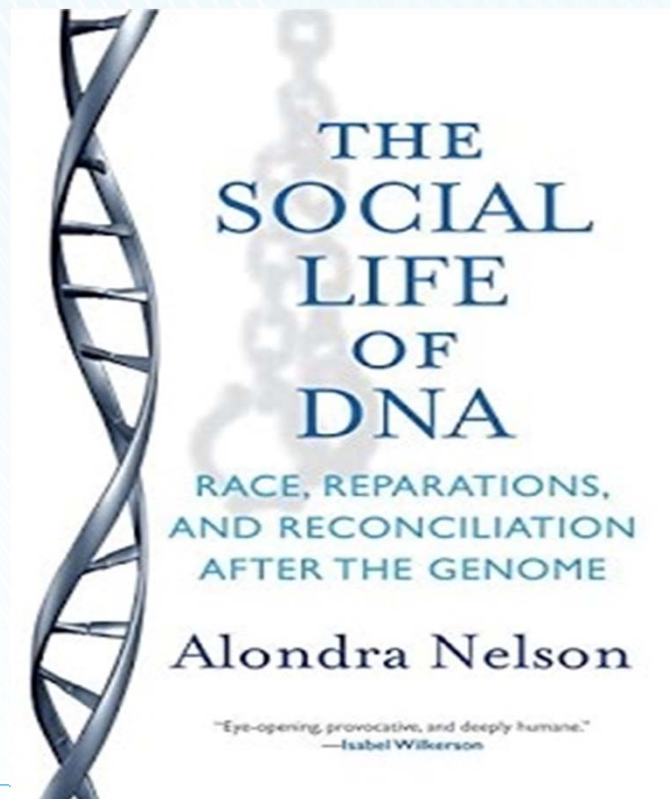
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Exploring the Current Landscape of Consumer Genomics  
National Academies of Sciences, Engineering, and Medicine  
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# Objectives

- Describe African American engagement in commercial genetic ancestry testing
- Discuss the selling of “identity” in the marketplace and potential implications for perceptions of ancestry and health

# Genetic Ancestry Testing: African Americans & “Root Seeking”





# How are genetic ancestry tests advertised?



# How are genetic ancestry tests advertised?

- “. . . population geneticists have observed an increased amount of African ancestry on the X chromosome in African Americans, suggesting involuntary mating between European-American men and African-American women.” Dr. Janina Jeff, *The Root*, April 2019
- “Only the white male character speaks. The enslaved Abigail is portrayed as passive and mute which unintentionally makes her the supporting actress in her own story.” Andre Kearns, *Medium*, April 2019

# How are genetic ancestry tests advertised?

## Ancestry Composition

Your DNA tells the story of who you are and how you're connected to populations around the world. Trace your heritage through the centuries and uncover clues about where your ancestors lived and when.



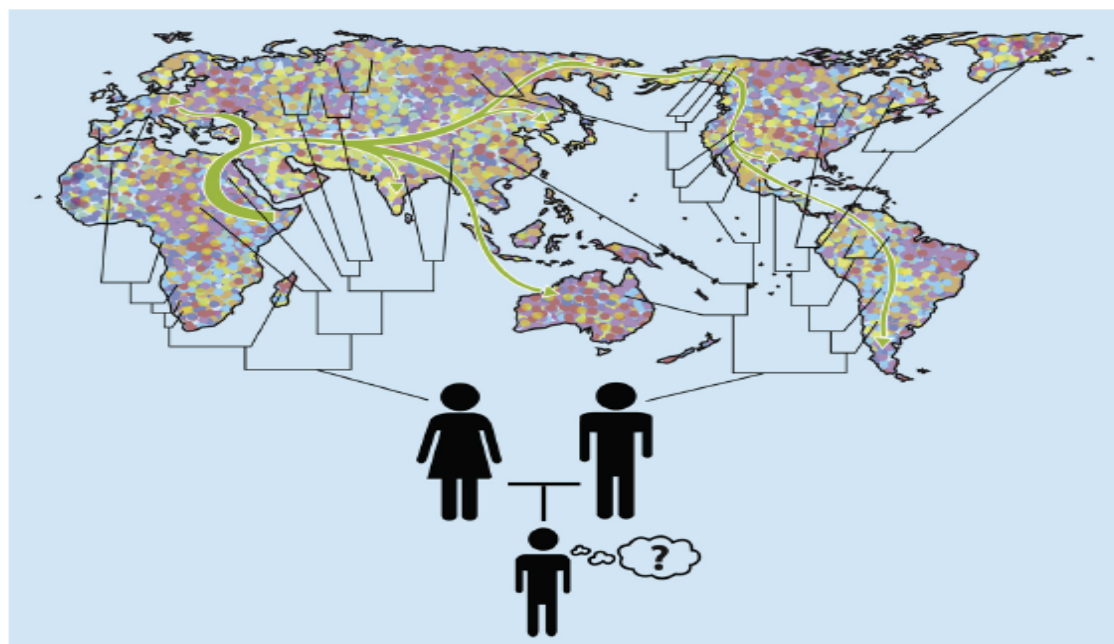
AncestryDNA® gives you much more than just the places you're from.

With precise geographic detail and clear-cut historical insights, we connect you to the places in the world where your story started—from unique regions to living relatives.

[Explore AncestryDNA](#)



# Do consumers understand genetic ancestry?



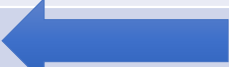



**Figure 1. Global Ancestry**

The arrows symbolize migration of early human ancestors out of Africa. The color mosaic denotes global population diversity resulting from various subsequent inter- and intra-continental and regional migrations. The pedigree represents the complex network of intermediate and recent ancestors that is the subject of individual genetic genealogy testing.

## Inferring Genetic Ancestry: Opportunities, Challenges, and Implications

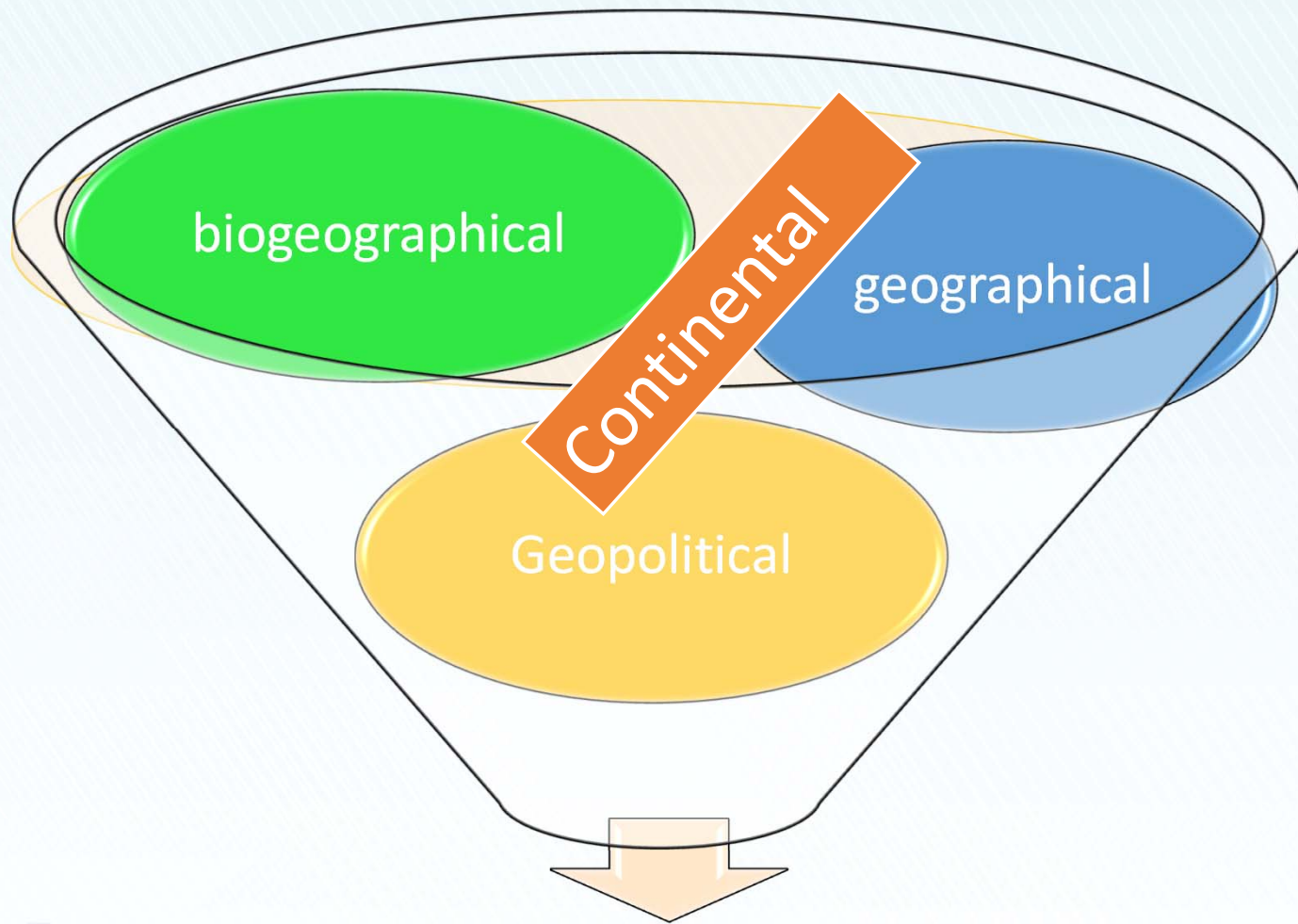
Charmaine D. Royal,<sup>1,\*</sup> John Novembre,<sup>2</sup> Stephanie M. Fullerton,<sup>3</sup> David B. Goldstein,<sup>1</sup>  
Jeffrey C. Long,<sup>4</sup> Michael J. Bamshad,<sup>5</sup> and Andrew G. Clark<sup>6</sup>

# What is the value for African Americans?

Region	Ancestry	FamilyTreeDNA	23andMe
<b>Africa</b>	96%	95%	95%
Nigeria, Benin, Togo, Ghana	43%		45%
West Africa	18% 	87%	39% 
Central Africa	33% 	3%	6% 
East Africa or Madagascar		5%	6%
Other regions	2%		4%
<b>Europe</b>	4%	4%	5%
Great Britain, Ireland		4%	
Other regions	4%		5%

**Results for Nicole: Her parents and grandparents are African American. She hoped to learn more about distant relatives and her connections to specific tribes or regions in Africa.** See Kevin Brasler, "DNA Ancestry Testing: Are Their Reports Rooted in Reality?" Available at Checkbook.org (May 2019)





**Are the tests changing the way we describe and/or think about population differences?**

# Are genetic ancestry tests improving the way consumers think about Race?

- Wendy D. Roth, Biorn Ivermark. Genetic Options: The Impact of Genetic Ancestry Testing on Consumers' Racial and Ethnic Identities. *American Journal of Sociology*, 124, no. 1(2018):150.
- **Genetic options theory:** “. . .consumers do not accept the tests' results as given but choose selectively from the estimates, embracing or ignoring particular genetic ancestries according to two mechanisms: their identity aspirations, or preferences for the ethnic or racial identities they seek to claim, and their social appraisals, their assessment of how others will accept their identity claims.”
- “. . . selective geneticization, with consumers picking and choosing the genetic ancestries they want to embrace.”

# Are genetic ancestry tests improving the way consumers think about Race?

- Janet K. Shim, Sonia Rab Alam, and Bradley E. Aouizerat. "Knowing something versus feeling different: the effects and non-effects of genetic ancestry on racial identity." *New Genetics and Society* 37, no. 1 (2018): 44-66.
  - "Our participants drew a fairly firm distinction between their genetic ancestry information – which they saw as occasionally meaningful but yet still 'just information' – and their self-identity."
  - "... results had no effect on how they conceived of who they were, or the communities and peoples with whom they affiliated."
  - "... they did 'feel different,' but not in ways that displaced their existing ideas about their racial identities."



# Are genetic ancestry tests improving the way consumers think about Race?

- Mwenza Blell and M. A. Hunter. "Direct-to-Consumer Genetic Testing's Red Herring: "Genetic Ancestry" and Personalized Medicine." *Frontiers in Medicine* 6 (2019).
  - Ancestry tests misrepresent human genetic diversity
- Charmaine D. Royal, John Novembre, Stephanie M. Fullerton, David B. Goldstein, Jeffrey C. Long, Michael J. Bamshad, and Andrew G. Clark. "Inferring genetic ancestry: opportunities, challenges, and implications." *The American Journal of Human Genetics* 86, no. 5 (2010): 661-673.
  - "continental ancestry"; "biogeographic ancestry"; "lineage or family history"
  - "... Consumers might share ancestry test results or ancestry-related estimates of disease risk with their healthcare providers and expect that the information be factored into their care."

# Ancestry & Health

- Sarah E. Ali-Khan, Tomasz Krakowski, Rabia Tahir, and Abdallah S. Daar. "The use of race, ethnicity and ancestry in human genetic research." The HUGO journal 5, no. 1-4 (2011): 47-63.
  - "... no article using 'race', 'ethnicity' or 'ancestry' defined or discussed the meaning of these concepts in context; a third of articles still do not provide a rationale for their use, with those using 'ancestry' being the least likely to do so."
  - "... there remains a clear imperative for highlighting the importance of consistent and comprehensive reporting on human populations to the genetics/genomics community globally, to generate explicit guidelines for the uses of ancestry and genetic ancestry, and importantly, to ensure that guidelines are followed."

# Ancestry & Health

- Alice B. Popejoy, Deborah I. Ritter, Kristy Crooks, Erin Currey, Stephanie M. Fullerton, Lucia A. Hindorff, Barbara Koenig et al. "The clinical imperative for inclusivity: Race, ethnicity, and ancestry (REA) in genomics." Human mutation 39, no. 11 (2018): 1713-1720.
- Overall, no two clinical laboratories provided the same descriptive categories to designate a group or population on their RFs. Nor was there consistency in the way laboratories described the category of information being sought about REA. Metadata headers (for the REA section of lab RFs) included terms such as "ancestry," "ethnicity," "race and ethnicity," or blank (preselected race and ethnicity options, field was not named).



# Ancestry & Health

A content analysis of the views of genetics professionals on race, ancestry, and genetics

Sarah C. Nelson, Joon-Ho Yu, Jennifer K. Wagner, Tanya M. Harrell, Charmaine D. Royal & Michael J. Bamshad



## Race, Ancestry, and Reporting in Medical Journals

Richard S. Cooper, MD; Girish N. Nadkarni, MD, MPH; Gbenga Ogedegbe, MD, MS, MPH

[Abstract](#) | [Full Text](#)

*JAMA*. 2018;320(15):1531-1532. doi:10.1001/jama.2018.10960

## Examining How Race, Ethnicity, and Ancestry Data Are Used in Biomedical Research

Vence L. Bonham, JD; Eric D. Green, MD, PhD; Eliseo J. Pérez-Stable, MD

[Abstract](#) | [Full Text](#)

*JAMA*. 2018;320(15):1533-1534. doi:10.1001/jama.2018.13609

# Are we missing important opportunities?

- Clear and concise language related to race, ethnicity, and ancestry
- Trust related to genetic/genomic testing services
- Tools for patient and provider communication about ancestry and risk