

Exceptions to the “One Drop Rule”?

DNA evidence of African Ancestry in European Americans



J.L. Mountain¹, J.M. Macpherson¹, C.B. Do¹, B.T. Naughton¹, R.A. Kittles², N. Eriksson¹.

¹23andMe, Inc, Mountain View, CA; ²Institute of Human Genetics, University of Illinois at Chicago, Chicago, IL.

Introduction

Genetic studies have revealed that most African Americans trace the majority (80%, on average) of their ancestry to western Africa.¹ Most of the remaining ancestry traces to Europe, and paternal lines trace to Europe more often than maternal lines. This genetic pattern is consistent with the “One Drop Rule,” a social history wherein children born with one or more ancestors of African descent were considered “black” in the United States. The question of how many European Americans have DNA evidence of African ancestry has been studied far less, although recent estimates for several southern European populations indicate that between 1% and 3% of ancestry traces to Africa.² Here we describe the analysis of genome-wide data for over 98,000 individuals. The aim of this study is to characterize the distribution of African ancestry among those likely to self-identify as European American or of European descent. The findings have implications for the type of personal discoveries individuals can make through exploring their DNA.

Methods

We examined genetic ancestry for 98,240 customers of 23andMe who had consented to participate in research. The vast majority reside in the United States. We identified a subset of individuals with genetic evidence of fewer than one in 16 great-great-grandparents tracing ancestry to a continental region other than Europe (estimated African ancestry less than 5%). Principal component analysis and self-report indicate that most of these individuals trace ancestry primarily to northern Europe. These individuals are likely to consider themselves to be entirely of European descent, at least within the last 500 years.

We conducted two analyses to understand what fraction of this group has genetic evidence of some ancestry tracing recently to Africa. We first identified individuals whose autosomal DNA indicates that they are predominantly of European ancestry, but who carry either a mitochondrial (mt) DNA or Y chromosome haplogroup that is highly likely to have originated in sub-Saharan Africa (Table 1), since that region was the primary source of enslaved individuals brought to North America.

Y chromosome

A, B2a1a, E1a, E1b1, E2

Mitochondrial DNA

L0, L1, L2, L3a, L3b, L3c, L3d, L3e, L3f, L3h, L3k, L3x, L4b2a, L6, M1

Table 1. Y chromosome and mtDNA haplogroups detected among research participants that are found primarily in sub-Saharan Africa, the source of most enslaved individuals brought to North America. These haplogroups plus all sub-haplogroups were considered as evidence of African ancestry.³⁻⁶

We then identified the subset of individuals of European descent with estimates of greater than 0% and up to 5.0% of their ancestry tracing to Africa. These estimates were obtained from genotype data for between 580,000 and 1 million single nucleotide polymorphisms, via an algorithm similar to STRUCTURE⁷ that underlies 23andMe’s Ancestry Painting feature.

We also explored the self-reported ancestry of these individuals in order to better understand their perceptions of their ancestry and to assess whether DNA results indicating African ancestry were consistent with their previous understanding, or were likely to be seen as novel information.

Results

Of the 78,552 23andMe customers likely to consider themselves of European descent, 501, about 2/3 of 1%, carry an mtDNA haplogroup indicating African ancestry. Of approximately 33,000 males, 135, or about one in 300, trace their paternal line to Africa. Overall about 1% of individuals with predominantly European ancestry trace a maternal or paternal line to Africa. It is possible that for some individuals these haplogroups reflect very deep African ancestry tracing back 1000s of years. The estimate of 1%, however, provides an upper bound on the African ancestry tracing back to interactions between individuals of African and European descent within the United States.

We then identified the set of Europeans and European Americans who have estimates of between 0.5% and 5.0% of ancestry tracing to Africa, leaving out the subset with estimates of less than 0.5% since such estimates may be an artifact of the ancestry estimation algorithm. Figure 1 shows the distribution of % estimated African ancestry for the 2736 individuals meeting these criteria.

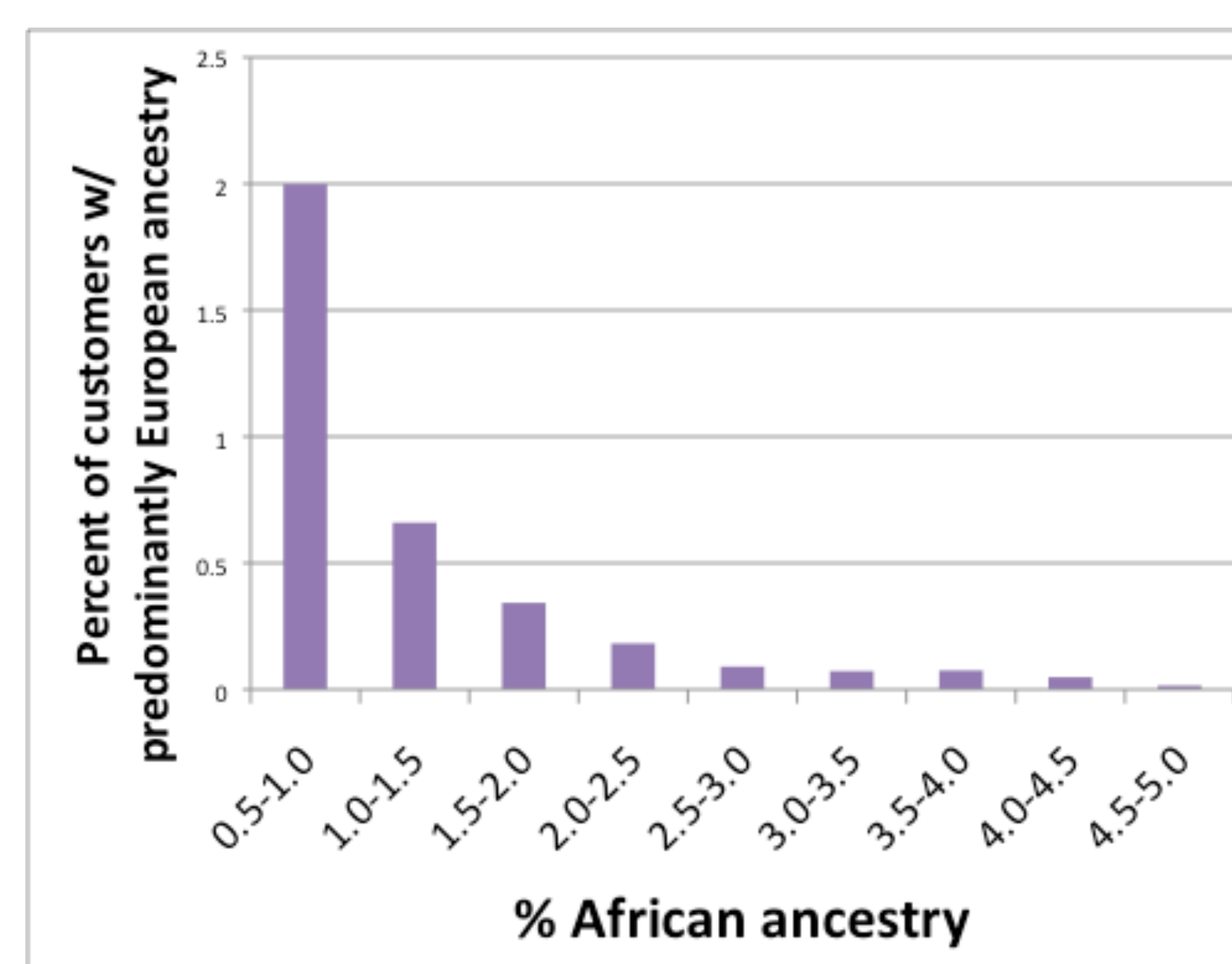


Figure 1. Percentage of individuals likely to self-identify as of European descent who have genetic evidence of African ancestry. Limited to those with at least 0.5% estimated African ancestry.

The data underlying the distribution of Figure 1 can be interpreted in terms of number of generations (Fig. 2).

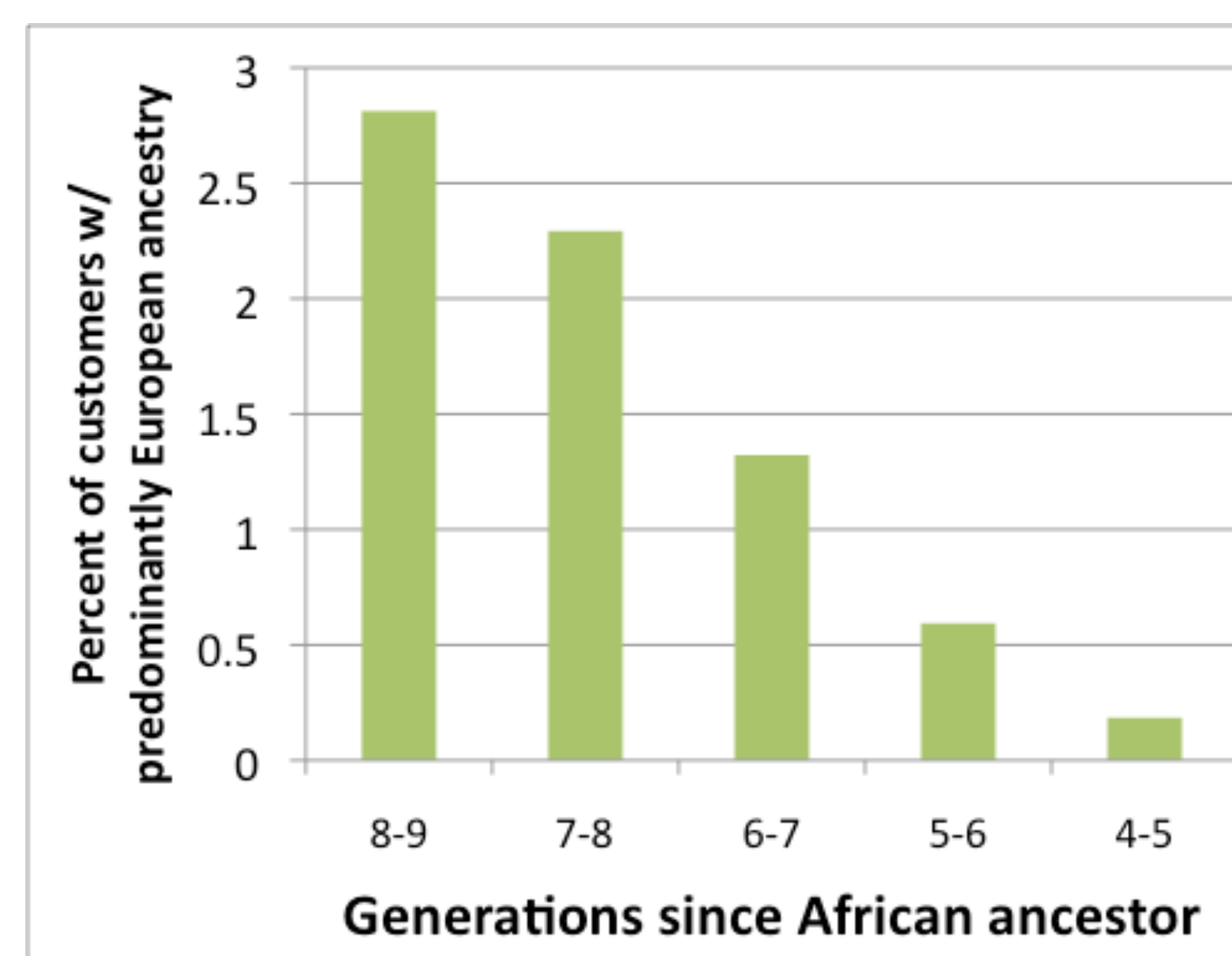


Figure 2. Percentage of individuals likely to self-identify as of European descent who have genetic evidence of African ancestry a given number of generations in the past.

The set of individuals within the distributions of figures 1 and 2 subset constitutes about 3.5% of the individuals likely to be aware only of their European ancestry. The majority (75%) of that group has a very small estimated fraction of African ancestry (about 0.5%), likely to reflect African ancestry over seven generations (roughly 200 years) ago.

Our exploration of the ancestry reported by a subset of the individuals indicated that only about 0.2% of the individuals with estimates of African ancestry between 0.5% and 5% identify as having ancestry other than European. That fraction increases as the % of estimated proportion of African ancestry increases.

Discussion

About 1% of individuals likely to identify as only of European descent have a Y chromosome or mtDNA haplogroup associated primarily with sub-Saharan Africa. We estimate that, overall, 3-4% of 23andMe customers with predominantly European ancestry have genetic patterns suggesting relatively recent (within last 10 generations) ancestry tracing to Africa. This fraction is far lower than the genetic estimates of European ancestry of African Americans, which is closer to 20%. This finding is consistent with the social history of the United States, wherein individuals with at least one ancestor of African descent were considered to be black. However, the analysis indicates that a small percentage of “mixed race” individuals were integrating into the European American community 200 years ago and earlier, prior to and during the era of slavery in the United States. Anecdotal evidence indicates that individuals discovering DNA evidence of African ancestry have been prompted to conduct genealogical research to search for the identity of their African ancestors.⁸⁻¹⁰

Acknowledgments

We thank 23andMe’s customers who consented to participate in research for enabling this study. We also thank the employees of 23andMe who contributed to the development of the infrastructure that made this research possible.

References and Resources

1. Zakharia F et al (2009) Characterizing the admixed African ancestry of African Americans *Genome Biology* 10:R141
2. Moorjani P et al (2011) The history of African gene flow into southern Europeans, Levantines, and Jews *PLoS Genet* 7:e1001373
3. Salas et al (2002) The making of the African mtDNA landscape *Am J Hum Genet* 71:1082-1111
4. Y-DNA Haplogroup Tree 2011. International Society of Genetic Genealogy. www.isogg.org Retrieved March 2010
5. Tishkoff SA, Gonder MC, Henn BM, Mortenson H, Knight A, Gignoux G, Fernandez-Pellegrini N, Lema G, Nyambo TB, Ramakrishnan U, Reed F, Mountain JL (2007) History of click-speaking populations of Africa inferred from mtDNA and Y chromosome genetic variation *Mol Biol Evol* 24: 2180-2195
6. van Oven M, Kayser M (2009) Updated comprehensive phylogenetic tree of global human mitochondrial DNA variation. *Hum Mutat* 30(2):E386-E394. <http://www.phylotree.org>.
7. Pritchard, JP, Stephens M, Donnelly P (2000) Inference of population structure using multi-locus genotype data *Genetics* 155: 945-959
8. <http://spittoon.23andme.com/2011/08/19/stories-from-23andme-james-larry-vick/>
9. <http://spittoon.23andme.com/2011/09/30/our-hidden-african-ancestry/>
10. <http://www.yourgeneticgenealogist.com/2011/09/dna-test-spurs-surprising-discovery-of.html>