

Genetic study clarifies African and African-American ancestry

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This is a photo taken during data collection in Africa. Credit: Sarah Tishkoff and the University of Pennsylvania

People who identify as African-American may be as little as 1 percent West African or as much as 99 percent, just one finding of a large-scale, genome-wide study of African and African-American ancestry released today.

An international research team led by scientists from the University of Pennsylvania and Cornell University has collected and analyzed genotype data from 365 African-Americans, 203 people from 12 West African populations and 400 Europeans from 42 countries to provide a genome-wide perspective of African and African-American ancestry.

The data reveal genomic diversity among African and African-American populations far more complex than originally thought and reflect deep historical, cultural and linguistic impacts on gene flow among populations. The data also point to the ability of [geneticists](#) to reliably discern ancestry using such data. Scientists found, for example, that they could distinguish African and European ancestry at each region of the genome of self-

identified-African Americans.

Sarah Tishkoff, a geneticist at Penn, and Carlos Bustamante, a computational biologist at Cornell, led the study to analyze 300,000 [genetic markers](#) from across the genome from West African, African-American and European-American populations to see whether they could reliably distinguish ancestry.

The team found that, while some West African populations are nearly indistinguishable, there are clear and discernible genetic differences among some groups, divided along linguistic and geographic lines.

This newly acquired [genetic data](#) revealed a number of important advances, including:

- The rich mosaic of African-American ancestry. Among the 365 African-Americans in the study, individuals had as little as 1 percent West [African ancestry](#) and as much as 99 percent. There are significant implications for pharmacogenomic studies and assessment of disease risk. It appears that the range of genetic ancestry captured under the term African-American is extremely diverse, suggesting that caution should be used in prescribing treatment based on differential guidelines for African-Americans.
- A median proportion of European ancestry in African-Americans of 18.5 percent, with large variation among individuals.
- The predominately African origin of X chromosomes of African-Americans. This is consistent with the pattern of gene flow where mothers were mostly of African ancestry while fathers were either of African or European ancestry.

- A technique which can reliably distinguish African and European ancestry for any particular region of the genome in African-Americans. This could have implications for personalized ancestry reconstructions, personalized medicine and more effective drug treatments and could aid in developing more effective methods for mapping genetic risk factors for diseases common in African-Americans, such as hypertension, diabetes and prostate cancer.
- The similarity of the West African component of African-American ancestry to the profile from non-Bantu Niger-Kordofanian speaking populations, which include the Igbo and Yoruba from Nigeria and the Brong from Ghana
- A comparison of the West African segments of African-American genomes. This is wholly in line with historical documents showing that the Igbo and Yoruba are two of the 10 most frequent ethnicities in slave trade records; however, most African-Americans also have ancestry from Bantu-speaking populations in western Africa.
- Population structure within the West African samples reflecting primarily language and secondarily geographical distance, echoing the Bantu expansion from a homeland in West Africa across much of sub-Saharan Africa around 4,000 years ago.

"Africa, which is the homeland of all modern humans, contains more than 2,000 ethnolinguistic groups and harbors great genetic and phenotypic diversity; however, little is known about fine-scale population structure at a genome-wide level," said Tishkoff, professor in the departments of genetics and biology at Penn. "We were able to distinguish among closely related West African populations and showed that genetically inferred ancestry correlates strongly with geography and language, reflecting historic migration events in Africa.

"We were also able to show that there is little genetic differentiation among African-Americans in

the African portion of their ancestry, reflecting the fact that most African-Americans have ancestry from several regions of western Africa. The greatest variation among African-Americans is in their proportion of [European ancestry](#), which has important implications for the design of personalized medical treatments."

The study focused primarily on the genetic structure of West African populations, as previous genetic and historical studies suggested that the region was the source for most of the ancestry of present-day African-Americans. The results suggest that there are clear and discernible [genetic differences](#) among some of the West African populations, whereas others appear to be nearly indistinguishable, even when comparing more than 300,000 genetic markers. The researchers note that a larger sample size would likely reveal further substructure and diversity between these populations.

Analyzing patterns of population structure and individual ancestry in Africans and African-Americans illuminates the history of human populations and is critical for undertaking medical genomic studies on a global scale. Understanding ancestry not only provides insight into historical migration patterns, human origins and greater understanding of evolutionary forces, but also allows researchers to examine disease susceptibility and pharmacogenic response, and to develop personalized drugs and treatments, a frontier in public health.

There is also strong reason to believe that high-density genotype data from African and African-American populations may pinpoint more precisely the geographic origin of African [ancestry](#) in African-Americans, the researchers said. The study appears online in the *Proceedings of the National Academy of Sciences*.

Provided by University of Pennsylvania

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