

Geneticists publish largest-ever study on African genetics revealing origins, migration

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African, American, and European researchers working in collaboration over a 10-year period have released the largest-ever study of African genetic data--more than four million genotypes--providing a library of new information on the continent which is thought to be the source of the oldest settlements of modern humans.

The study demonstrates startling diversity on the continent, shared ancestry among geographically diverse groups and traces the origins of Africans and African Americans. It is published in the April 30 issue of the journal <u>Science Express</u>.

Researchers studied 121 African populations, four African American populations and 60 non-African populations for patterns of variation at 1327 DNA markers. The study traced the <u>genetic structure</u> of Africans to 14 ancestral population clusters that correlated with ethnicity and shared cultural and/or linguistic properties. The research team demonstrated that there is more <u>genetic diversity</u> in Africa than anywhere else on earth.

They also determined that the ancestral origin of humans was probably located in southern Africa, near the South Africa-Namibian border. Extrapolating the data, scientists were able to map ancient migrations of populations and determined that the exit point of modern humans out of Africa was near the middle of the Red Sea in East Africa. They also provide evidence for ancient common ancestry of geographically diverse hunter-gatherer populations in Africa, including Pygmies from central



Africa and click-speaking populations from southern and eastern Africa, suggesting the possibility that the original pygmy language may have contained clicks. Overall, they demonstrate remarkable correspondence between cultural, linguistic, and genetic diversity in Africa.

"This is the largest study to date of African genetic diversity in the nuclear genome," said Sarah Tishkoff, a geneticist with joint appointments in the School of Arts and Sciences and the School of Medicine at the University of Pennsylvania. "This long term collaboration, involving an international team of researchers and years of research expeditions to collect samples from populations living in remote regions of Africa, has resulted in novel insights about levels and patterns of genetic diversity in Africa, a region that has been underrepresented in human genetic studies. Our goal has been to do research that will benefit Africans, both by learning more about their population history and by setting the stage for future genetic studies, including studies of genetic and environmental risk factors for disease and drug response."

Tishkoff says that there is no single African population that is representative of the diversity present on the continent. Therefore, many ethnically diverse African populations should be included in studies of human genetic variation, disease susceptibility, and drug response.

Anthropologists, historians and linguists now have at their disposal a completely new volume of research with which to test theories of human migration, cultural evolution and <u>population</u> history in Africa. Basic scientists, physicians and public health officials now have a foundation for illuminating the complex history of Africans and African-Americans, with implications for studies aimed at finding disease genes in these populations and learning which genetic differences make some individuals more susceptible to diseases like HIV, cancer or malaria.

This study also sheds light on African American ancestry, which they



find originates predominantly from western African Niger-Kordofanian (~71 percent), European (~13 percent), and other African (~8 percent) populations, although admixture levels varied considerably among individuals. These results could have important implications for the design and interpretation of studies which aim to identify genetic and environmental risk factors for diseases common in the African American community, including prostate cancer, hypertension and diabetes.

Source: University of Pennsylvania (<u>news</u> : <u>web</u>)

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