Genetic data added to archaeology and linguistics to get picture of African population history

May 26 2010

Genetic researchers from the University of Pennsylvania have combined data from existing archaeological and linguistic studies of Africa with human genetic data to shed light on the demographic history of the continent from which all human activity emerged.

The study reveals not just a clearer picture of the continent's history but also the importance of having independent lines of evidence in the interpretation of genetic and genomic data in the reconstruction of population histories.

The study is published in the current issue of the Proceedings of the National Academy of Sciences.

The results, according to Penn geneticist Sarah Tishkoff, a Penn Integrates Knowledge professor with joint appointments in the schools of Medicine and Arts and Science, is that genetic variation in Africa is structured geographically, and to a lesser extent, linguistically. The findings are consistent with the notion that populations in close geographic proximity that speak linguistically similar languages are more likely to exchange genes.

Furthermore, genetic variation in Africa appears consistent with the natural, geographic barriers that limit gene flow. In particular, there are geographic, and therefore genetic, distinctions between northern African and sub-Saharan African populations due to the vast desert that limited migration.

"Focusing on particular exceptions to these broad patterns will enable us to discern and fully appreciate the complex population histories that have contributed to extant patterns of genetic variation," said Tishkoff, the David and Lyn Silfen University Associate Professor. "Disentangling past population histories is a formidably complicated task that benefits from the synthesis of archaeological, linguistic and genetic data."

With this three-pronged approach, a clearer picture emerges.

Archaeology provides insights into ancient technology and culture, affording a timeline for the emergence of innovations. Historical linguistic data complement the archaeological record by contributing an independent phylogenetic analysis of language relationships and providing clues about ancient population migration and admixture events. Genetic data provide an independent data source to understand the biological relationships among modern peoples and likely points of origin and expansion of their ancestors.

"The details of modern human demography are complex and not well understood, so we have taken a cross-disciplinary approach to highlight broad patterns of population history in Africa," said Laura Scheinfeldt of the Department of Genetics at Penn's School of Medicine.

Patterns will continue to emerge as geneticists from Penn and elsewhere further analyze a mountain of genetic data acquired from these understudied populations.

The development of sequencing and genotyping technologies has advanced at an unprecedented rate and is allowing for the genotyping of millions of single-nucleotide polymorphisms and the sequencing of millions of nucleotides across populations.

This data, coupled with computational methods for inferring demographic parameters and testing demographic models - for example, maximum likelihood and approximate Bayesian computation -- can refine our understanding of African past population histories.

"The incorporation of archaeological and linguistic data will be important for establishing testable hypotheses and elucidating the evolutionary processes or forces that have shaped the genomic landscape in Africa," Tishkoff said.

Analyzing patterns of population structure and ancestry in Africans 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 and human origins and provides a greater understanding of evolutionary forces, it also allows researchers to examine disease susceptibility and pharmacogenic response and to develop personalized drugs and treatments, a frontier in public health.

Provided by University of Pennsylvania