

Migration from Africa left mark on European genetic diversity

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Human migration from Africa to Europe more than 30,000 years ago appears to have left a mark on the genes of Europeans today.

A Cornell-led study, reported in the Feb. 21 issue of the journal *Nature*, compared more than 10,000 sequenced genes from 15 African-Americans and 20 European-Americans. The results suggest that European populations have proportionately more harmful variations, though it is unclear what effects these variations actually may have on the overall health of Europeans.

Computer simulations suggest that the first Europeans comprised small and less diverse populations. That would have allowed mildly harmful genetic variations within those populations to become more frequent over time, the researchers report.

"What we may be seeing is a 'population genetic echo' of the founding of Europe," said Carlos Bustamante, assistant professor of biological statistics and computational biology at Cornell and senior co-author with Andrew Clark, a professor of molecular biology and genetics.

"Since we tend to think of European populations as quite large, we did not expect to see a significant difference in the distribution of neutral and deleterious variation between the two populations," said Bustamante. "It was quite surprising, but when we cross-checked our results to data sets gathered by other groups, we found the same trend."

The researchers focused on single nucleotide polymorphisms (SNPs), where a single DNA base pair (the smallest structural unit) in a gene's sequence had been altered. Genetic variations were classified as to whether a SNP was found in one or both populations. Some of these genetic changes led to amino acid changes in the proteins that the genes express, while others had no effect.

Collaborators at Max Planck Institute in Tübingen, Germany, and Harvard Medical School analyzed the amino acid changes and used a computer algorithm to predict whether the changes alter a protein's structure or function, and classified the changes into three categories: benign, possibly damaging or probably damaging.

Using that information, the Cornell group found that the European sample, while showing overall less genetic variation, had proportionately more amino acid changes and proportionately more harmful amino acid single nucleotide polymorphisms than the African sample.

"It's difficult to tell what the precise impact that a higher proportion of deleterious single nucleotide polymorphisms in the population will have on the average person's health," said Kirk Lohmueller, a graduate student in both Bustamante's and Clark's labs and the paper's lead author. "More detailed studies that involve sequencing many individuals both with and without certain diseases would better enable us to get at this question."

Future research may also reveal similar signatures as other populations left Africa for other geographic destinations.

Source: Cornell University

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