

Researchers discover evidence of very recent human adaptation

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A Cornell study of genome sequences in African-Americans, European-Americans and Chinese suggests that natural selection has caused as much as 10 percent of the human genome to change in some populations in the last 15,000 to 100,000 years, when people began migrating from Africa.

The study, published in the June 1 issue of *PLoS Genetics*, looked for areas where most members of a population showed the same genetic changes. For example, the researchers found evidence of recent selection on skin pigmentation genes, providing the genetic data to support theories proposed by anthropologists for decades that as anatomically modern humans migrated out of Africa and experienced different climates and sunlight levels, their skin colors adapted to the new environments.

However, the study found no evidence of differences in genes that control brain development among the various geographical groups, as some researchers have proposed in the past.

"We undertook a very careful study of genetic differences within and among major human groups, and aimed to explain why certain parts of the genome differed," said Scott Williamson, the study's lead author and a Cornell assistant professor of biological statistics and computational biology. "We aimed to eliminate as many possible confounding variables as possible, and when all is said and done, we find that as much as 10 percent of the genome may have been affected by one of these bouts of recent selection."

Previous studies at Cornell and elsewhere have searched for signs of selection -- the divergence of genes from a common ancestor millions of years ago -- by comparing an individual human to a chimpanzee or mouse, for example, or by comparing genetic variation in protein coding

genes among humans to differences between humans and a chimpanzee. But this study scanned genome sequences that compared many humans to each other throughout the entire genome, with new strict statistical methods that correct for many potential biases that creep into this kind of analysis.

In the latest study, the researchers identified 101 regions of the human genome with strong evidence of very recent selection. These regions include genes that control proteins that help muscle cells attach to surrounding cells (mutations of this gene lead to muscular dystrophy), receptors that relate to hearing, genes involved in nervous system function and development, immune system genes and heat shock genes.

The gene scan method also detected selection in a gene involved in digestion of lactose, an enzyme found in milk. Prior to animal domestication, humans lost the ability to digest milk after infancy. But, as humans migrated and domesticated animals, Europeans and other populations developed a gene for tolerating lactose (and milk) throughout their lives. This finding has been well established in previous research, so arriving at similar results provided an internal validation for the accuracy of the new method.

Overall, close to 10 percent of the Chinese and European-American genomes and only 1 percent of the African-American genome were linked to areas with evidence of recent selection. Since Africans have the greatest genetic diversity and the statistical method searched for areas where the majority of members within a population group have the same genetic changes, signs of evolution were much easier to detect in the less diverse European-American and Chinese genomes.

"It is important to emphasize that the research does not state that one group is more evolved or better adapted than another," said co-author Carlos

Bustamante, a Cornell assistant professor of biological statistics and computational biology. "Rather as humans have populated the world, there has been strong selective pressure at the genetic level for fortuitous mutations that allow digestion of a new food source or tolerate infection by a pathogen that the population may not have faced in a previous environment."

Source: Cornell University News Service

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