

Adaptation Plays Significant Role In Human Evolution

ScienceDaily (Jan. 20, 2009) — For years researchers have puzzled over whether adaptation plays a major role in human evolution or whether most changes are due to neutral, random selection of genes and traits.



Geneticists at Stanford now have laid this question to rest. Their results, scheduled to be published Jan. 16 online in Public Library of Science Genetics, show adaptation-the process by which organisms change to better fit their environment-is indeed a large part of human genomic evolution.

"Others have looked for the signal of widespread adaptation and couldn't find it. Now we've used a lot more data and did a lot of work cleaning it up," said Dmitri Petrov, associate professor of biology at Stanford University and one of two senior authors of the paper. "We were able to detect the adaptation signatures quite clearly, and they have the characteristic shape we anticipated."

All genetic mutations start out random, but those that are beneficial to an organism's success in their environment are directly selected for and quickly perpetuate throughout the population, providing a uniform, traceable signature.

With the help of post-doctoral researcher James Cai and recent graduate student Michael Macpherson, Petrov and co-senior author Guy Sella, a biologist at the Hebrew University of Jerusalem, used different methodology from what's been used before to look for signatures of adaptation left in the human genome.

"We detected a number of signatures that suggest adaptation is quite pervasive and common," Petrov said.

Humans have a very complex history from traveling around the globe, and the human genome is also highly structured, making it complicated and difficult to work with, he said.

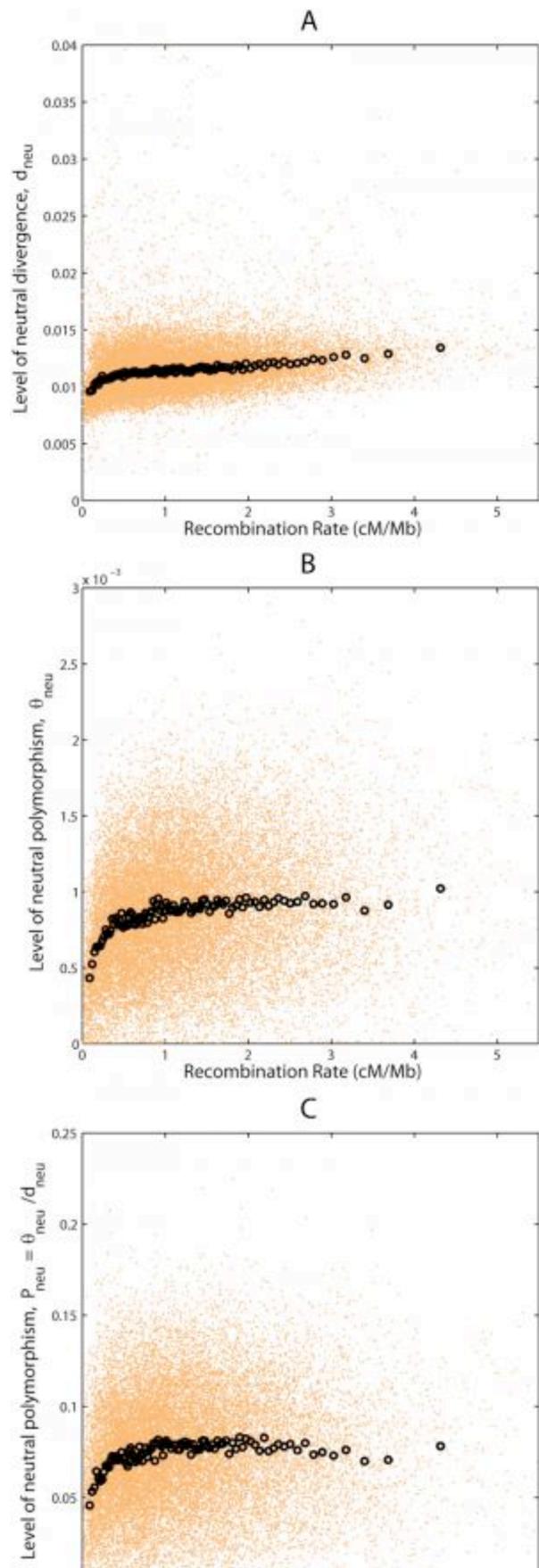
To find the adaptation signal, Petrov and his colleagues looked for regions of the genome that "hitchhiked" along with an adaptation. When a genetic adaptation occurs and is passed on to offspring, other genes on both sides of the adaptation typically accompany it. The result is a whole region of the genome where all humans are unusually similar to each other, referred to as a "selective sweep," that researchers can identify and trace through human genetic history.

"Adaptation becomes widespread in the population very quickly," Petrov said. "Whereas neutral random mutation doesn't and would not have the selective sweep signature."

"We tried to see if these regions of unusual similarity among all humans tended to be in particular places in the genome as the theory predicts they should be, and indeed we find them there," Petrov said. "The work suggests human beings have undergone rampant adaptation to their environment in the last 200,000 years of history."

In the past, these sweeps were difficult to discern because the data were not sufficiently abundant and were filled with noise. Depending on the methodology, estimates of the degree of adaptation in humans ranged from as high as 30 percent down to zero. Signatures were impossible to interpret with confidence.

"People would find changes in specific genes suggesting that recent adaptations in humans might be common but could not find genome-wide signatures of pervasive adaptation. That was unsettling," Petrov said. "I'm hoping that people



will react with relief that things are starting to make sense."

Petrov hopes that researchers can now do a much better job of finding the regions within the genome responsible for specific human adaptations and relate them to changes in human history or past environments. For example, one could trace the arrival of lactose tolerance to the domestication of cattle and the introduction of milk into our adult diet.

"As the data are going to grow, we should be able to locate specific adaptive events quite well," Petrov said. "By identifying specific genes, we can unravel this evolutionary history of adaptive change."

Another possibility is tracing the origin of skin pigmentation genes, which give people their different skin-color types. Many of these genes are linked to skin cancer. Researchers may be able to recreate past environments while better understanding how adaptation comes into play.

"We see signatures of possibly hundreds of recent adaptive events, and now we can ask what are they doing there," he said. "It's both exiting and puzzling."

This paper follows similar work in bacteria and fruit flies indicating adaptation is a significant contribution to evolution as a whole.

"We are on a crest of a wave showing that adaptation is a lot more prevalent than we thought," Petrov said.

Correlations between recombination rate and neutral divergence rate and neutral polymorphism: Scatter plots display values of two variables in orange dots for (A) recombination rate and the level of neutral divergence rate (d_{neu}), (B) recombination rate and the level of neutral polymorphism (θ_{neu}), and (C) recombination rate and the level of normalized neutral polymorphism ($P_{neu} = \theta_{neu}/d_{neu}$). Black circles are average values for orange dots pooled in 100 bins each containing 1% of the data points. (Credit: DOI: 10.1371/journal.pgen.1000336.g001)

Journal reference:

1. James J. Cai, J. Michael Macpherson, Guy Sella, Dmitri A. Petrov. **Pervasive Hitchhiking at Coding and Regulatory Sites in Humans.** *PLoS Genetics*, 2009; 5 (1): e1000336 DOI: [10.1371/journal.pgen.1000336](http://dx.doi.org/10.1371/journal.pgen.1000336)

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