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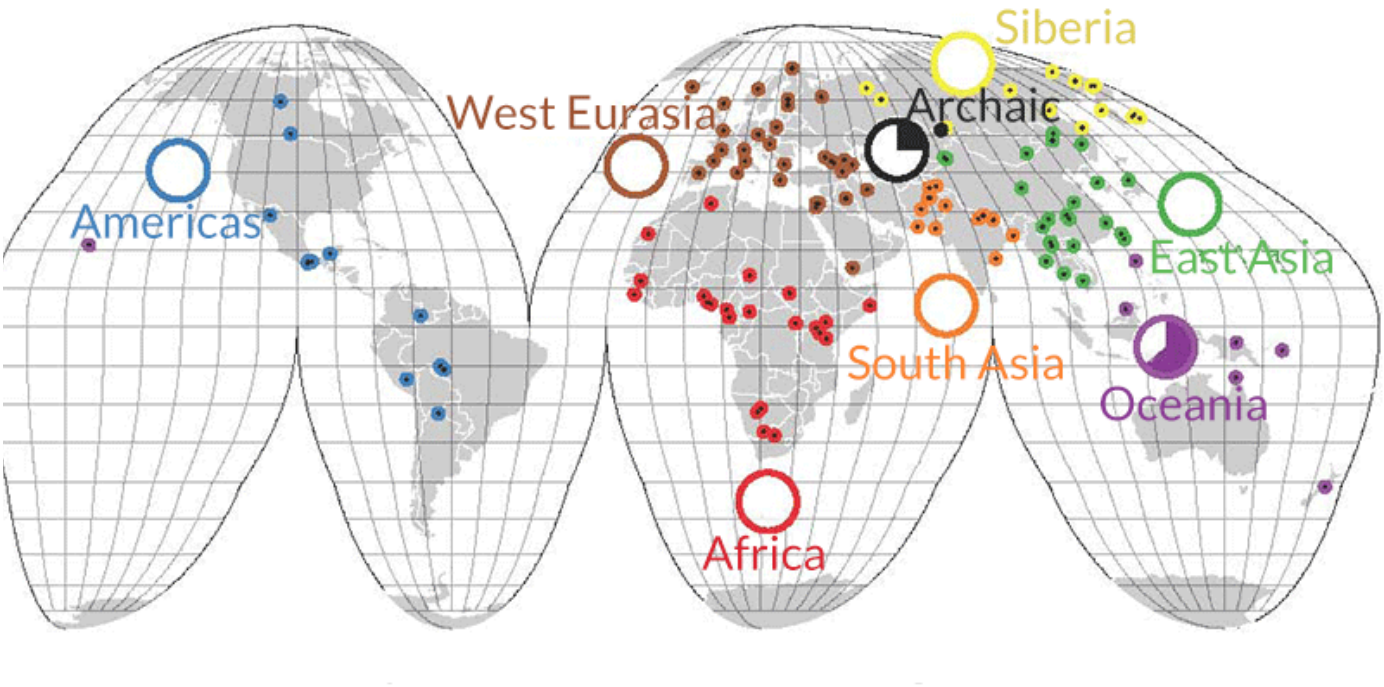
News: Genetics,Human Evolution,Ancestry

Ancestral humans had more DNA

Atlas mapping genetic deletions and duplications reveals evolutionary relationships

By Tina Hesman Saey 2:21pm, August 6, 2015



DNA DIVERSITY A global atlas of human genetic diversity reveals that some Oceanians (purple) carry duplicated DNA inherited from Denisovans (black). No Neandertals nor any other human group carry the duplication (indicated by open circles). The black "Archaic" circle represents a Denisovan and a Neandertal.

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A new atlas of human genetic diversity reveals what human ancestors' DNA may have looked like before people migrated out of Africa.

Ancestral humans carried 40.7 million more DNA base pairs than people do today, researchers repor online August 6 in *Science*. That's enough DNA to build a small chromosome, says study coauthor E Eichler, an evolutionary geneticist at the University of Washington in Seattle.

Human ancestors in Africa jettisoned 15.8 million of those DNA base pairs — information-carrying building blocks of DNA often referred to by the letters A, T, G and C — before dispersing around the

globe, the researchers discovered. As people left Africa and spread to other continents, they dropped more chunks of DNA. Eichler and colleagues have followed these genetic bread crumbs to map relationships among 125 human groups worldwide.

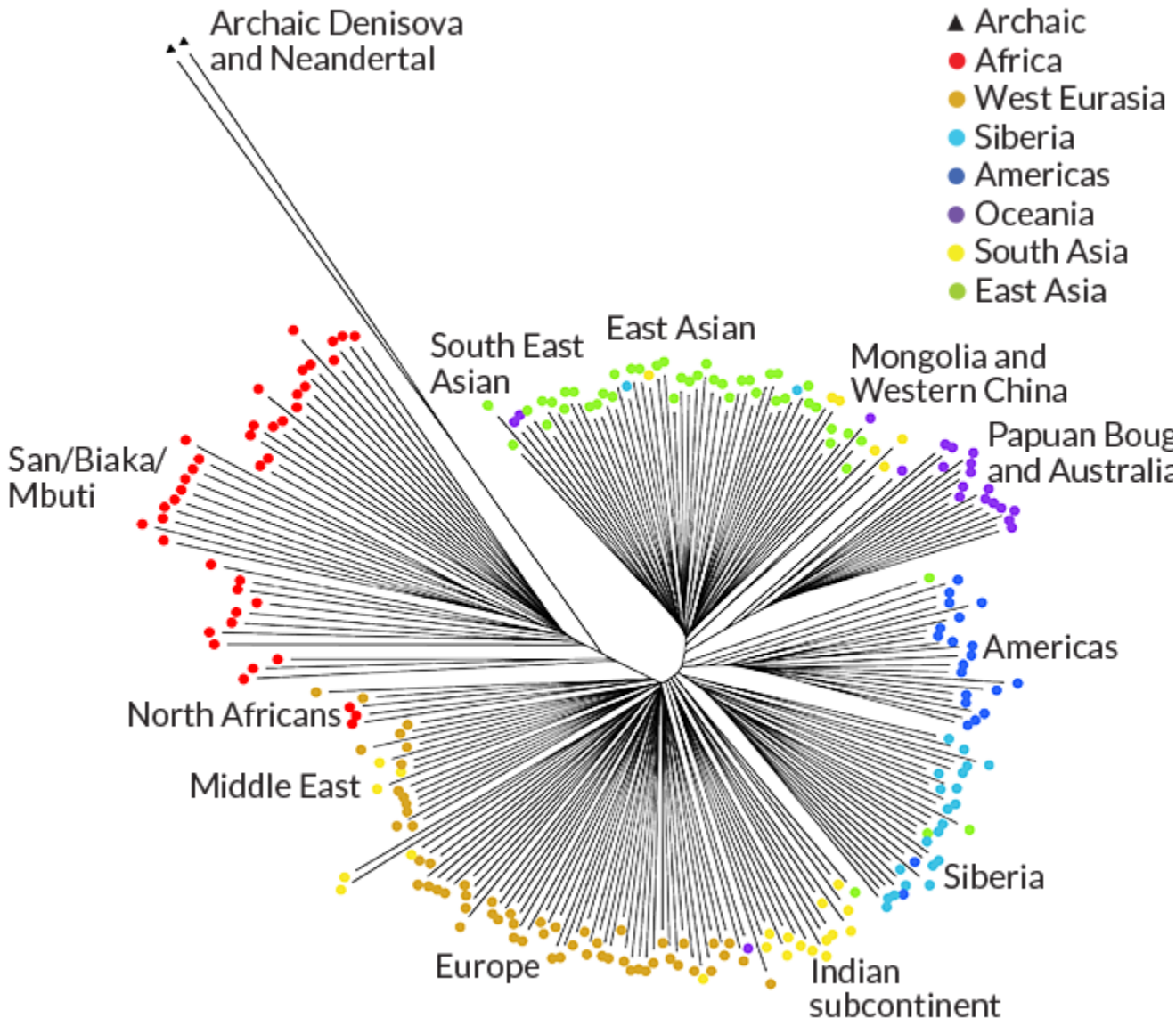
People didn’t just lose DNA. They also gained some. Compared with chimpanzees and orangutans, people have 728 extra pieces of DNA created when portions of the human genetic instruction book, tl genome, were copied. Everyone has at least three copies of those duplicated bits, although the exact number varies from person to person.

Previous maps of human genetic diversity have usually not marked the yawning chasms left by deleti or the new territory created by duplications. Most diversity maps have focused on single DNA base pa changes, often called single nucleotide polymorphisms, or SNPs. But all the SNPs together comprise only 1.1 percent of the genome. Duplications and deletions, collectively known as copy number varia have shaped more than 7 percent of the human genome.

Story continues below infographic

Common loss

Researchers used DNA deletions to trace relationships among human groups. Longer lines ind the group has more deletions unique to it.



P. Sudmant et al/Science 2015

Because duplications and deletions involve larger swaths of DNA than SNPs do, their influence on human evolution may also be bigger. Both duplications and deletions have been implicated in shapin human characteristics, such as bigger brains (*SN: 3/21/15, p. 16; SN: 4/9/11, p. 15*).

But researchers “can’t answer the question yet of whether what makes us human is in what was lost what was duplicated,” says David Liberles, a computational evolutionary biologist at Temple Universit Philadelphia.

Eichler's choice is clear. "Duplications rock," he says. "They affect more base pairs in the human genome than any other type of variation." Duplications span 4.4 percent of the genome, while deletion represent 2.77 percent. And duplications tend to involve genes, while deletions often fall in spaces between genes, the researchers found.

His team flagged many duplications as possible medical and evolutionary points of interest. For instance, some groups of people have up to six copies of *CLPS* genes, which encode pancreatic enzymes that may help reduce blood sugar levels. Some African groups carry duplications of genes t may protect against sleeping sickness caused by trypanosome parasites.

Another attraction is a very large duplication of about 225,000 base pairs that Papua New Guineans inherited from Denisovans, an extinct group of hominids related to Neandertals. The colossal hunk of DNA contains two microRNA genes. MicroRNAs are small molecules that help regulate protein production. Eichler and colleagues calculate that the original duplication happened about 440,000 ye ago in Denisovans. It was passed to Papuans and some other Melanesians about 40,000 years ago when their ancestors interbred with Denisovans. Now, about 80 percent of Papuans carry the duplication. Eichler speculates that the duplication may have given Papuan ancestors some evolution advantage, although what that advantage might be isn't known.

While the researchers make a compelling case that duplications and deletions may play an important role in evolution, the team has provided little evidence that copy number variants really determine trai differences between groups, says Edward Hollox, a human geneticist at the University of Leicester in England. "It's almost a paper saying, 'Look, isn't this interesting?' But why it's interesting they haven't quite gotten to the bottom of." Still, Hollox says the map will point other researchers to parts of the genome where evolution may have left its mark.

Citations

P.H. Sudmant et al. Global diversity, population stratification, and selection of human copy number variation. *Science.* Published online August 6, 2015. doi: 10.1126/science.aab3761

Further Reading

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