

Everyone alive may carry DNA from a ‘ghost lineage’ of human ancestors

Scientists analyzing modern genomes may have found a long-lost branch of the human family tree

In a new *Science* study, scientists read the family tree buried inside modern genomes and turned up “ghost ancestors” in every population on Earth that no fossil records can account for.

Anatomically modern humans are just one branch of a broader family tree. The other branches—the archaic hominins, as scientists call them—include the Neanderthals, who lived across Europe and western Asia, and the Denisovans, who are known mostly from a handful of fossils found in Siberia and Tibet. These archaic lineages interbred with ancient human groups who had migrated out of Africa, which scientists can confirm by comparing rare preserved samples of their DNA with ours.

But did our direct ancestors interbreed with other long-lost hominin lineages before the first major migrations out of Africa roughly 70,000 years ago? Because we have never found any archaic hominin genomes in sub-Saharan Africa, scientists couldn’t tell. To get around that limitation, a team of scientists co-led by Yulin Zhang, a computational biologist at the University of California, Berkeley, and geneticist Arjun Biddanda, now at Johns Hopkins University, developed a new method for discovering archaic ancestry using only the DNA of humans from our time.

Our genomes contain not just one but thousands of family trees, each embedded in a short stretch of genetic code that has been shuffled around (or “recombined”) with each generation. “It’s a collection of gene trees ... representative of the entire genealogical history,” Zhang explains. Reconstructed correctly, these trees make it possible to estimate when each contribution to our DNA got there. Most of the modern human genome coalesces to a common ancestor 100,000 to 200,000 years back, but DNA inherited from deeply divergent populations like Neanderthals or Denisovans does not—it produces a branch in a family tree that’s unusually long. “At the end you get this nice tracing along the chromosome: this section contains the signature of archaic ancestry, and the remaining sections don’t,” Biddanda says.

The researchers analyzed 503 contemporary genomes from across the world with a new type of mathematical model they developed called TRACE (Tracking Archaic Contributions via ARG Estimation). It found that 2 percent of the DNA in non-African genomes came from archaic lineages, consistent with prior research. Roughly half of that, or 1 percent of the genome, matched sequenced Neanderthals, and a sliver matched Denisovans. The rest, though, ranging from about 0.5 to 1 percent depending on the population, matched nothing anyone has ever sequenced. This “ghost” lineage showed in every population Zhang and her colleagues examined and likely split off from our branch more than 500,000 years ago, around the time that Neanderthals and Denisovans did.

On top of that, the unknown lineage showed up in stretches of DNA that are almost devoid of Neanderthal and Denisovan genes—and which scientists had interpreted as uniquely our own. “When we specifically looked at these regions, they also harbored this ghost ancestry,” says Priya Moorjani, a population geneticist at Berkeley and a co-author of the study. “We were really surprised.” One of these regions sits around *FOXP2*, a gene linked to speech.

The results will need to be confirmed with another line of evidence. “As with any method, there will always be false positives,” says Patrick F. Reilly, a computational biologist at Yale University, who was not involved in the study. Further evidence could come from other computational methods (each carrying their own limitations) or directly from as-yet-undiscovered fossils with preserved ancient DNA in sub-Saharan Africa.

“Unless we get extraordinarily lucky with DNA preservation, we’ll likely have to rely on methods like TRACE,” Reilly says. One way to be more confident in the new results, he suggests, would be checking how similar the supposed ghost lineage sequences in our genome are to Neanderthal and Denisovan genomes. If both have equally low similarity, Reilly argues, we could infer that these ghosts formed an equally distant sister group to Denisovans, Neanderthals and possibly modern humans.

“Perhaps these methods really have opened up a new frontier for looking at human evolution,” Moorjani says.

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