No Bones about It: Ancient DNA from Siberia Hints at Previously Unknown Human Relative

For the first time, researchers describe a new type of human ancestor on the basis of DNA rather than anatomy

Kate Wong March 24, 2010

Credit: Johannes Krause

For much of the past five million to seven million years over which humans have been evolving, multiple species of our forebears co-existed. But eventually the other lineages went extinct, leaving only our own, *Homo sapiens*, to rule Earth. Scientists long thought that by 40,000 years ago *H. sapiens* shared the planet with only one other human species, or hominin: the Neandertals. In recent years, however, evidence of a more happening hominin scene at that time has emerged. Indications that *H. erectus* might have persisted on the Indonesian island of Java until 25,000 years ago have surfaced. And then there's *H. floresiensis*—the mini human species commonly referred to as the hobbits—which lived on Flores, another island in the Indonesian archipelago, as recently as 17,000 years ago.

Now researchers writing in the journal *Nature* report that they have found a fifth kind of hominin that may have overlapped with these species. (*Scientific American* is part of Nature Publishing Group.) But unlike all the other known members of the human family, which investigators have described on the basis of the morphological characteristics of their bones, the new hominin has been identified solely on the basis of its DNA.

Johannes Krause and Svante Pääbo of the Max Planck Institute for Evolutionary Anthropology in Leipzig, Germany, and their colleagues obtained the DNA from a fossilized pinky finger bone found at Denisova Cave in the Altai Mountains of southern Siberia. The species was impossible to determine from the shape and size of the bone—it simply did not contain any diagnostic morphological traits. But there were good reasons to believe it came from a Neandertal or an early modern human. For one, the bone was recovered from a stratigraphic layer of the cave dated to between 50,000 and 30,000 years ago that contained artifacts belonging to the so-called Middle Paleolithic and Upper Paleolithic industries associated with these two groups. For another, Neandertals and modern humans were the only hominins known to have lived in this region during that time period. But the DNA the team extracted from the Denisova pinky bone turned out to be markedly different from DNA sequences previously obtained from early modern humans and Neandertals.

The researchers focused on a type of DNA known as mitochondrial DNA (mtDNA). Mitochondria are the power plants of the cell, and they have their own DNA that is separate from that housed in the cell nucleus and is passed down from mother to offspring. Because each cell has thousands of mitochondria, but only a single nucleus, mitochondrial DNA is much more abundant than nuclear DNA and is therefore more likely than the latter to be preserved in fossilized bone. To date, scientists have sequenced the mitochondrial genomes of both Neandertal and early modern human individuals, and the sequences for the two groups are quite distinctive.

Comparing the order of the genetic "letters"—or base-pairs, as they are termed—making up the Denisova mtDNA with the sequences of modern day humans and an early modern human, Krause and his collaborators found that the Denisova mtDNA differed from humans today in nearly twice as many letter positions as Neandertal mtDNAs do. Further analysis indicated that the most recent common mtDNA ancestor of the Denisova individual, Neandertals and modern humans dates to around a million years ago (making it twice as old as the most recent common mtDNA ancestor of Neandertals and moderns). This divergence date, the team says, indicates that the Denisova mtDNA is distinct from that of the H. erectus population that left Africa 1.9 million years ago, and also from that of the Neandertal ancestor *H. heidelbergensis*, which branched off from the lineage leading to modern humans around 466,000 years ago. As such, the researchers contend the Denisova mtDNA reveals a previously unrecognized migration out of Africa by a hitherto unknown group of hominins. (The team is holding off on giving the creature a formal name for now, but informally they refer to it as X-woman.)

"The data that they provide is certainly of the nature to arrive at the conclusions that they do," comments Stephan Schuster of The

Pennsylvania State University, who worked on the recent sequencing of Archbishop Desmond Tutu's nuclear genome as well as the nuclear genome sequencing of a woolly mammoth. "All the detected sequence differences clearly indicate that this is a novel variant of a [hominin]."

Paleoanthropologist Ian Tattersall of the American Museum of Natural History in New York City noted that the finding should not necessarily come as a surprise. "We know the fossil record is far from complete, but what we have already shows that the [hominin] evolutionary bush is quite luxuriantly branching," he remarks. "One more branch is not something that ought to give us indigestion."

The association of the mystery hominin with those Middle and Upper Paleolithic artifacts is peculiar though, because elsewhere in Eurasia they have only turned up with Neandertal and modern human remains. Krause notes that it is possible that the pinky bone originated in an older, deeper layer of the cave sediments and over time got mixed in with the overlying artifacts. Thus far, however, there is no evidence for extensive perturbation. Another possibility, he says, is that the finger bone is that of an early modern human who carried an ancient mtDNA as a result of interbreeding between his or her ancestors and this previously unknown hominin group.

But other experts are not so sure about the team's interpretation of their data. "I don't know—and nobody else does—how many base-pair changes make a new species," says Erik Trinkaus of Washington University in Saint Louis, an authority on Neandertals and early modern humans. "I would like to have more than the number of mtDNA base pair differences to go on."

"The result doesn't mean that they've found a new species, and I don't believe it requires a separate pre-Neandertal migration out of Africa," argues John Hawks of the University of Wisconsin–Madison, whose research focuses on human genetic evolution. "Those explanations are both compatible with the result, but I don't think the data require them yet." Hawks notes that the history of an mtDNA sequence—which is just a tiny fraction of a person's total DNA—does not necessarily reflect the history of a species.

A comparably distinctive nuclear genome sequence would significantly strengthen the claim that the Denisova mtDNA represents a previously unknown type of hominin. To that end, Krause and Pääbo are launching a Denisova genome project to obtain a full nuclear genome sequence from the bone that yielded the novel mtDNA. Comparisons of this genome with the full genome sequence they have obtained for the Neandertal as well as with the genomes of people living today could yield insights into the genetic changes that defined H. sapiens. "At the end we get more information about the big question [of] what makes humans humans," Krause reflects.

Meanwhile, paleoanthropologists are eager for more fossils to confirm the DNA-based claim. With luck, continued excavation at Denisova cave this summer will turn up additional remains—and put a face on this long-lost relative.