

## GENES &amp; CELLS

# Neandertal hot spots highlighted in modern humans' DNA

Stone Age people's mating with now-extinct species had both genetic pros and cons

BY BRUCE BOWER

Humans appear to have inherited several traits related to skin, hair and some autoimmune diseases from Neandertal ancestors.

Two independent investigations identify for the first time the specific parts of the human genome that seem to have been most affected by Stone Age interbreeding with Neandertals. The research locates part of Neandertals' legacy in sections of present-day Europeans' and East Asians' DNA that are stocked with genes influencing the production of keratin, a key substance in skin, hair and nails.

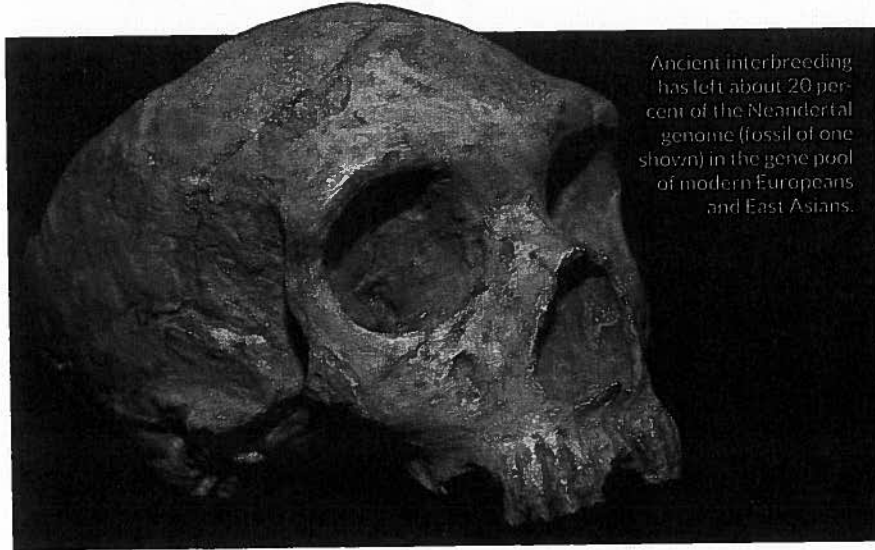
By occasionally interbreeding with Neandertals after leaving Africa around 70,000 years ago, Stone Age humans inherited and retained keratin-related genes that must have aided survival outside of Africa, Sriram Sankararaman, a computational geneticist at Harvard Medical School, and his colleagues propose January 29 in *Nature*.

Neandertals' DNA contributions to modern humans also encompass genes related to several medical conditions, including lupus and Crohn's disease, Sankararaman's group says.

Neandertals lived in Europe and Asia between around 200,000 and 30,000 years ago. Previous studies estimated that 1 to 3 percent of non-Africans' DNA today comes from Neandertals, while present-day Africans have little or no Neandertal ancestry.

About 20 percent of the Neandertal genome shows up in various places in the DNA of living Europeans and East Asians, geneticists Benjamin Vernot and Joshua Akey of the University of Washington in Seattle conclude January 29 in *Science*.

These new reports represent "the first steps toward getting a genomic picture of the actual Neandertal individuals that mixed with modern humans," remarks paleogeneticist Mattias Jakobsson of



Ancient interbreeding has left about 20 percent of the Neandertal genome (fossil of one shown) in the gene pool of modern Europeans and East Asians.

Uppsala University in Sweden.

The genome of a Neandertal woman who lived about 50,000 years ago has already been sequenced (*SN*: 1/25/14, p. 17). The two research teams compared the ancient woman's genome with modern-day humans' DNA, using different statistical techniques. Future work will be able to use genomes from additional Neandertal fossils to home in on genes that modern humans inherited through particular instances of Stone Age interbreeding.

Sankararaman's team developed a method for calculating the probability that modern human gene variants and DNA segments containing multiple genes came from Neandertals. The researchers compared the Neandertal woman's genome with the DNA of 1,004 modern-day humans living in different parts of the world.

If, for instance, a European carried a gene variant found in the Neandertal but not in present-day West Africans, whose ancestors do not appear to have interbred with Neandertals, the researchers concluded that the gene variant probably originated in Neandertals.

In a genetic analysis of 379 Europeans and 286 East Asians, Vernot and Akey identified unusually long chains of gene

variants that people probably inherited via Stone Age interbreeding, presumably from Neandertals. These Neandertal hot spots did not appear in the DNA of 13 West Africans.

Sankararaman's group found that far fewer signs of Neandertal ancestry appear on the X chromosome and along a stretch of DNA containing genes that affect the testicles than in other parts of modern humans' genomes. Genes that reduce male fertility tend to accumulate on the X chromosome when closely related species of modern animals interbreed, suggesting that such genes initially might have passed from Neandertals to humans before disappearing due to natural selection. Neandertal genes that compromised how the testicles work may have met the same fate.

The patterns of Neandertal DNA found in the human genome suggest that ancient populations interbred at least twice across Europe and Asia, Vernot and Akey say.

Still, large swaths of Europeans' and East Asians' genomes contain unexpectedly little Neandertal DNA, Akey says. "These regions potentially are a road map to finding genes that make us human." ■

## New demise date for Neandertals

Study estimates extinction in Europe at 40,000 years ago

BY BRUCE BOWER

Neandertals died out in Western Europe earlier than many scientists thought, between about 41,000 and 39,000 years ago, after interbreeding with modern humans for a few thousand years, a new study suggests.

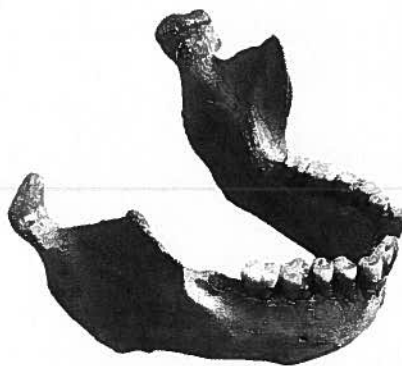
These new findings join a long-standing debate about the fate of the Neandertals that shows no signs of diminishing.

Previous reports that some Neandertals survived in southwestern Europe until about 30,000 years ago hinged on underestimates of the age of carbon in ancient bones and other material, says archaeologist Tom Higham of the University of Oxford and colleagues. Improved methods now indicate that Neandertals disappeared at different times in different regions of Europe before going extinct about 40,000 years ago, the scientists report in the Aug. 21 *Nature*.

The new dates suggest that Neandertals and modern humans (*Homo sapiens*) simultaneously inhabited Western Europe for 2,600 to 5,400 years. While populations of the two hominids overlapped, they could have interbred and exchanged cultural knowledge.

Stone Age sites in Central and Eastern Europe have yet to be dated with the new techniques. That leaves a gap in what's known about how long Neandertals survived and the extent to which they mingled with modern humans, archaeologist William Davies of the University of Southampton in England writes in the same issue of *Nature*. Still, he says, "future researchers will need to try hard to demonstrate Neandertal survival in Europe after 40,000 years ago."

Higham's team dated finds from 40 sites, all in Western Europe except for one in Russia and another in Lebanon. The researchers mainly dated animal bones displaying butchery marks.



European Neandertals, including one represented by this lower jaw excavated in southern Spain, survived no later than about 40,000 years ago, new radiocarbon dates suggest.

Stone tools at most sites had previously been attributed to the Mousterian and Châtelperronian cultures, usually regarded as Neandertals' handiwork.

Neandertals had Europe largely to themselves 45,000 years ago, Higham's team says. They then vanished from different regions at different times. Mousterian tools found in Italy, for instance, were replaced between 44,800 and 43,950 years ago by cutting tools attributed to modern humans.

Châtelperronian finds in France, thought by some experts to represent a final phase of Neandertal culture and by others to be modern human creations (*SN*: 5/13/06, p. 302), were made no later than 40,000 years ago, the scientists say.

Higham adds that sites in southern Spain, reported to have hosted Neandertals until roughly 30,000 years ago (*SN*: 9/23/06, p. 205), also don't break the 40,000-year-old barrier in his analysis.

Higham's results confirm suspicions that Europe's last Neandertals and first modern humans inhabited parts of the continent at the same time, comments paleoanthropologist Jean-Jacques Hublin of the Max Planck Institute for Evolutionary Anthropology in Leipzig, Germany. In their last millennia, Neandertals made tools and personal ornaments based on techniques picked up from modern humans, he proposes.

João Zilhão, an archaeologist at the University of Barcelona, disagrees. It's more likely that Neandertals were variants of *H. sapiens* that achieved cultural advances long before being genetically swamped by large numbers of incoming modern humans from Africa, he asserts. ■