Sex with Neandertals Introduced Helpful and Harmful DNA into Modern Human Genome

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Kate Wong January 30, 2014

Neandertal DNA survives in Asian and European people today. Image: 120, via Wikimedia Commons

Over the past few years a number of studies of ancient and contemporary genomes have reached the same stunning conclusion: early human species interbred, and people today carry DNA from archaic humans, including the Neandertals, as a result of those interspecies trysts. Now two new analyses of modern human genomes are providing insights into how the acquisition of Neandertal DNA affected anatomically modern *Homo sapiens* tens of thousands of years ago and how it continues to affect people today.

In the first study, Sriram Sankararaman and David Reich of Harvard

University and their colleagues compared a complete Neandertal genome sequence with 1,004 modern human sequences to see which regions of the modern genome contain Neandertal DNA. Like other researchers before them, they observed that Asians and Europeans have DNA from Neandertals, whereas Africans have little or no Neandertal DNA. The pattern is consistent with a scenario in which early modern humans mated with Neandertals they encountered when they migrated out of Africa and into Eurasia, where Neandertals had lived for hundreds of thousands of years.

Moreover, the team determined that Neandertal DNA is not distributed evenly across the genome. Some genes have a high proportion of Neandertal ancestry (which is to say, many people today carry the Neandertal versions of these genes). Those genes with the highest Neandertal ancestry are associated with keratin, a protein found in skin and hair. The Neandertal variants of these genes may well have helped early modern humans adapt to the new environments they found themselves in as they spread into Eurasia. But the researchers also found that people today carry Neandertal genes that are associated with diseases including Crohn's, type 2 diabetes and lupus.

Intriguingly, other regions of the modern human genome have no or very low Neandertal contribution, notably the X chromosome and genes related to the functioning of the testes. According to Sankararaman, Reich and their collaborators, the absence of Neandertal genetic material in these regions suggests that male hybrids who inherited a Neandertal X chromosome were infertile, and thus unable to pass their genes along to the next generation. The researchers detail their findings in a paper published in the December 30 *Nature*. (*Scientific American* is part of Nature Publishing Group.)

In the second study, published by *Science,* Benjamin Vernot and Joshua M. Akey of the University of Washington screened whole genome

sequences from 665 living Europeans and Asians for telltale signs of Neandertal contributions. Their results show that although non-Africans individually inherited between 1 and 3 percent of their genomes from Neandertals, different people carry different bits of Neandertal genetic material. Together these sequences represent around 20 percent of the Neandertal genome.

Like the other team, Vernot and Akey found evidence that Neandertals passed along beneficial skin genes to modern humans, including some linked to pigmentation. And they, too, observed genome regions devoid of Neandertal contributions. One such region contains the gene FOXP2, which plays an important role in speech.

Vernot and Akey's work is additionally interesting in that they were able to use statistical and computational methods to identify the Neandertal contributions in the genomes of modern-day people without using a Neandertal genome to guide their search. This work raises the possibility that simply by analyzing the genomes of people alive today, scientists will be able to discover and describe extinct human species that mated with early *H. sapiens* but that, unlike Neandertals, are unknown from the fossil record. Previous studies of genomes of living people have hinted at dalliances between early *H. sapiens* and unknown archaic humans in Africa. Perhaps this approach will shine a light on these mysterious skeletons in our closet.

The views expressed are those of the author(s) and are not necessarily those of Scientific American.