More mystery about Neanderthal and modern humans: How reliable is ancient DNA analysis?

A. J. Smuskiewicz | May 12, 2015 | Genetic Literacy Project

Twenty-two years ago, calcite-encrusted human remains were discovered in a limestone cave in Altamura, Italy, but the dating of the skeleton and its place in the history of human evolution remained murky. Now a new study evaluating the DNA from a piece of its right shoulder blade suggests that the fossil was a Neanderthal, the closest extinct relative of modern humans.

"It is a dream," said Fabio Di Vincenzo, the study's co-author. "His morphology offers a rare glimpse on the earliest phase of the evolutionary history of Neanderthals and on one of the most crucial events in human evolution. He can help us better understand when—and, in particular, how—Neanderthals evolved."

The DNA, dating as far back as 170,000 years ago, could pave the way for a broader picture of Neanderthal life.

> *The bone is so old that its DNA is too degraded for the researchers to sequence the fossil's genome—at least with current technology. However, they noted that next-generation DNA-sequencing technologies might be capable of such a task, which "could provide important results on the Neanderthal genome," [said] study co-author David Caramelli, a molecular anthropologist at the University of Florence in Italy…*

This new DNA analysis brought to mind recent news stories of DNA sequencing performed on molecules extracted from younger Neanderthal bones—the most prominent reports coming from research led by evolutionary biologist Svante Paabo of the Max Planck Institute in Germany. All these reports lead to questions about the decay rate of DNA and the technology used to reach conclusions about prehistoric genetics. How accurate are our DNA dating processes? How accurate is our picture of the evolution of modern humans?

Many questions

The analysis of prehistoric DNA is a fascinating and complicated

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subject, especially when it comes to determining genetic relationships between extinct forms of human beings and modern humans. DNA from Neanderthal (or Neandertal) bones is an especially intriguing topic. These long-gone (since from somewhere between 28,000 and 39,000 years ago) inhabitants of parts of Europe and central Asia were so similar to us, yet also so different. And for some period of time, modern-type humans (*Homo sapiens sapiens*) probably lived in the same geographical region as Neanderthals (*H. sapiens neanderthalensis*, or *H. neanderthalensis*, depending on your preferred classification scheme).

We can't help but wonder: What happened to the Neanderthals? Did we kill them off? Did we mate with them? Do their genes still survive in some people today? If so, which people?

These questions were long a matter a mere speculation. However, advances in DNA technology during the past decade have opened the questions up to scientific investigation. In May 2010, it was reported that a research team led by Paabo had sequenced "the whole Neandertal genome from powdered bone fragments taken from three females who lived in Europe 40,000 years ago." The sequencing revealed that "[b]etween 1% and 4% of the DNA in modern Europeans, Asians and those as far afield as Papua New Guinea, was inherited from Neanderthals."

Five years after this exciting report, which was formally published in *Science*, Paabo and other researchers continue to probe this matter. Although this research has much to teach us about our human heritage and evolution, it is important that we keep in mind the limitations of analyzing DNA molecules that are very, very old.

The "whole genome"?

To be perfectly clear, Paabo and his colleagues did not really sequence "the whole Neandertal genome" in precisely the same sense that the whole genome of a person living today can be sequenced and analyzed. As soon as any organism dies, its DNA begins to break down and decay as the nucleotide bonds fall apart. Chemical reactions with water in the ground accelerate the decay. After examining such factors, Danish and Australian researchers reported in 2012 that DNA has a half-life of only 521 years.

Thus, the analysis of ancient, partially decayed, and fragmentary DNA inevitably includes making certain assumptions and decisions about missing and degraded pieces, in order to fill in the gaps as best as possible. Some decisions about assembling Neanderthal sequences are based on sophisticated algorithms involving the use of modern human and chimpanzee genomes as guides. Further challenges are presented by the need to filter out contamination by modern DNA, such as from the researchers who handled the bones or from microorganisms that lived within or among the buried bones.

These challenges and complexities raise the possibility of the inadvertent introduction of errors in the sequencing process—even when all precautions are taken by highly professional researchers. These issues also open up any obtained data to different interpretations and to possible limitations in the application of the data.

Nevertheless, these analytical limitations have not prevented some media outlets from reporting each latest find as if it is clearly definitive

and absolutely conclusive. This can give the public faulty impressions of the findings. There is also a disturbing current trend among academic and scientific institutions and agencies (all types of science, not only anthropology) to inflate the significance of certain finds—perhaps to raise the public prominence of their institutions or to increase their chances of obtaining more government funds. The press and public relations departments of science-related institutions and university departments are light-years ahead of where they were a couple decades ago, in terms of their media savvy.

The scientific research may be genuinely valuable to society and/or to collective human knowledge, but caution and care must be used in the presentation of this data—both by the media and by the research institutions themselves. The public is apt to be left with a uncomfortable feeling as one reportedly firm and revolutionary scientific conclusion is soon contradicted by another reportedly firm and revolutionary scientific conclusion.

Yes, there was inbreeding between Neanderthals and modern humans

The current "consensus" among anthropologists is that there was some degree of inbreeding between Neanderthals and modern humans. This consensus is largely based on Paabo's research findings about DNA sequencing, which always receive a great deal of media coverage. However, even with this general agreement, much remains uncertain regarding the timing and other details of possible inbreeding.

But that's not how such information is always presented by scientists and the media. One of Paabo's most recent studies, published in *Nature* in October 2014, based on "a good genome of a 45,000 year old person who was close to the ancestor of all present-day humans outside Africa." According to Tech Times:

> *Scientists have long suspected that modern humans and our ancient Neanderthal cousins interbred, and now DNA evidence shows that it did occur, and dates when it occurred, researchers say.*
>
> *Sequencing of DNA in a thighbone of one of the earliest modern humans, discovered in 2008 in Siberia, shows interbreeding occurred sometime between 50,000 and 60,000 years ago, they say.*

The article added, "A small imprint of Neanderthal DNA, about 2 percent, exists in all humans today except in Africans, evidence of interbreeding between the two species only after the 'Out of Africa' scenario of human development, researchers say."

A *BBC* report on the 2014 Paabo study dramatically stated that Paabo's finding "is rewriting the story of humanity" and that it "raises the possibility that the human line first emerged millions of years earlier than current estimates." The report even added this attention-grabbing quote from Paabo himself: "We seem to have caught evolution red-handed."

On the basis of a comparison of fragmentary DNA in a 45,000-year-old Neanderthal bone with DNA of some people living today, the BBC writer made a long leap in logic: "This raises the possibility that the very first species of the human line separated from apes 10 or 11 million years

ago—rather than the five or six million years ago that genetic evidence had previously suggested."

No, maybe there was not inbreeding

A Neanderthal DNA report with more nuance (which is always welcomed, though rarely encountered, in the media) appeared in *The Guardian* in August 2012. It was based on a study, published in *Proceedings of the National Academy of Sciences*, that reanalyzed the DNA inbreeding data reported by Paabo's team in 2010.

> *When scientists discovered a few years ago that modern humans shared swaths of DNA with long-extinct Neanderthals, their best explanation was that at some point the two species must have interbred.*
>
> *Now a study by scientists at the University of Cambridge has questioned this conclusion, hypothesising instead that the DNA overlap [described by Paabo's team in 2010] is a remnant of a common ancestor of both Neanderthals and modern humans.*

According to a model developed by Cambridge evolutionary ecologists Andrea Manica and Anders Eriksson, "There was an ancestor of both Neanderthal and modern humans—some archaeologists would call that *Homo heidelbergensis*—that would have covered Africa and Europe about half a million years ago."

In other words, just because some people today apparently share certain segments of DNA with extinct Neanderthals, that does not necessarily mean that interbreeding occurred. Yes, it is perfectly legitimate for different researchers to interpret scientific data differently—especially when the application of the technology involved in the research is relatively new.

Limitations should be acknowledged

All media reports on science should always be offered to the public with the appropriate qualifiers—such as noting that the findings are possible or preliminary, and continued studies are necessary to provide more details and clarification. The public should continually be reminded that this is how science works.

The media needs to convey to the public that science is a never-ending, complicated search for answers, with many detours and dead ends along the way. But, imperfect as it is, this is the best way we know of to get answers to complicated questions about nature. So stay tuned for updates. In addition, a little old-fashioned journalistic cynicism could go a long way toward furthering public understanding of complex scientific topics. Journalists should ask tough questions of scientists, just like they do of politicians. (But then, they don't often do that anymore either, do they?) It would also help if scientists themselves (or their public relations representatives) used less hyperbole in their press releases and other communications with the public.

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Richard Wilson • 2 years ago

No mention of the numerous problems with the Cann et al paper published in Nature that sought to lend support to the ridiculous notion that there was an African origin for so-called 'modern humans'.

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Laurence Evans ➚ Richard Wilson • 2 years ago

The African origin of humans and prevalence of Neanderthal genes only in the migrants out of Africa (while not in Africans) provides mutual support and evidence for both hypotheses. Sorry Richard, your ancestors are African.

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Richard Wilson ➚ Laurence Evans • 2 years ago

"The African origin of humans"? The haplogroup split from an LCA (of undetermined origin) around 160,000 years ago into African and Non-African groups does not imply that my ancestors are Out of Africa. Repeating the erroneous conclusions of Cann and others only illustrates the depth of the problem. Please read "reconsidering out of Africa" by Klyosov 2014 and then explain your conclusion, thanks.

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Valko Petrov • 2 years ago

The refusal of the theory of a common African origin of man leads to the acceptance of the multiregional hypothesis, which is as plausible as the creationism. The latter, however, is not considered a scientific theory by the majority of scientists. The same is valid for the multiregional theory too. It was not accepted by the majority of scientists. Sorry!

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