11. The fate of European Neanderthals: results and perspectives from ancient DNA analyses

D. SERRE
Max-Planck-Institute for Evolutionary Anthropology,
Leipzig D-04103,
Germany

Current address:
Genome Quebec Innovation Center and McGill University,
Montreal,
Canada
david.serre@mail.mcgill.ca

S. PÄÄBO
Max-Planck-Institut für Evolutionäre Anthropologie,
Leipzig D04103,
Germany
paabo@eva.mpg.de

Keywords: mtDNA, genetic diversity, population history, bottleneck, gene flow, coalescence, glaciation

Abstract

Analyses of mitochondrial DNA sequences extracted from several Neanderthal remains have provided new information on their genetic relationship with modern human individuals. However, these results have been interpreted very differently among anthropologists. Here we review these results and present additional data directly addressing the question of genetic continuity among human populations during the Late Pleistocene. An analysis of additional Neanderthal and early modern human remains from Western and Central Europe do not provide any evidence of gene flow between the two groups. We also show that under reasonable assumptions of human demography, these data rule out a major genetic contribution by Neanderthals to the modern human gene pool. Finally, we present preliminary results showing that ancient DNA studies can also contribute to unraveling aspects of Neanderthal demography. Promising avenues of research, such as the investigation of Neanderthal population genetic diversity and organization, as well as analyses of mammal populations contemporary with Neanderthals, could allow us to better understand the dynamics, and perhaps causes, of the demographic changes that occurred in Eurasia during the Late Pleistocene.

Introduction

Most researchers agree that the first hominids evolved in Africa (e.g., Campbell, 1988; Klein, 1989; Lewin, 1999) and that *Homo erectus* left Africa around two million years ago to colonize Europe and Asia as far as Indonesia (e.g., Wolpoff and Caspari, 1997; Gabunia et al., 2000; Oms et al., 2000; Wood and Richmond, 2000; Roebroeks, 2001; Balter and

K. Harvati and T. Harrison (eds.), Neanderthals Revisited: New Approaches and Perspectives, 211–220.

Gibbons, 2002; Vekua et al., 2002). However, the fate of archaic human populations that evolved regionally from this ancestral stock is much debated, especially with regards to a second wave of colonization from Africa around 100 ka. Most notably, attention has been focused on the fate of the Neanderthals,[1] the archaic humans that inhabited Europe and Western Asia during the later part of the Pleistocene (i.e., between 300 ka and 30 ka). Recent ^{14}C-dating confirms that the last Neanderthals could have co-existed with the first modern humans in Europe (Bocquet-Appel and Demars, 2000). However, it is still unclear whether this possible cohabitation influenced the gene pool of the newcomers or if, on the contrary, the Neanderthals went extinct without contributing to the gene pool of early modern humans in Europe.

In 1997, Krings extracted DNA from a humerus of the Neanderthal holotype (Krings et al., 1997). The 379 base pairs (bp) amplified from the hypervariable region of the mitochondrial (mt) genome were different from all modern human DNA sequences. Furthermore, this DNA sequence was too different from the current human sequences observed in the gene pool to be likely to be found in an individual that has not been analyzed yet. Tree reconstructions confirmed these analyses: while all human mtDNA sequences group together with a recent common ancestor,[2] the sequence retrieved from the type specimen of Neanderthal shows a much deeper separation with strong statistical support. This result is often interpreted as compelling evidence for the absence of interbreeding between Neanderthals and modern humans, or even as proof that Neanderthals and modern humans were two different species (e.g., Lindahl, 2000). However, even after the publication of two additional mtDNA sequences, very similar to that of the first individual (Krings et al., 2000; Ovchinnikov et al., 2000), many scenarios are still consistent with the data.

Two problems limit the range of the conclusions drawn from these studies: first, due to the impossibility of differentiating modern contamination from endogenous DNA sequences, a sample from a Neanderthal individual carrying a sequence similar to that of a current human could be discarded as putative contamination (Nordborg, 1998; Trinkaus, 2001). Second, the absence of early modern human DNA sequences leaves a long time span during which simple demographic processes can lead to the loss of Neanderthal sequences even with a substantial amount of admixture in the past (e.g., Relethford, 1998, 1999, 2001). Thus, the Neanderthal mtDNA could have been swamped by a continuous influx of modern human mtDNA into the Neanderthal gene pool (Enflo et al., 2001), lost by genetic drift (Nordborg, 1998), or by a population replacement much later than the Paleolithic transition, for example during the Neolithic expansion (e.g., Cavalli-Sforza et al., 1993). Here we summarize results that overcome these problems. The paper addresses the question of continuity or replacement between Neanderthals and early modern humans, as well as, more generally, aspects of what happened to the human populations during the transition from the Middle to Upper Paleolithic in Europe.

Looking for Gene Flow Between Neanderthals and Early Modern Humans

Contamination is the major problem of ancient DNA studies dealing with human remains because it is currently impossible to differentiate endogenous DNA sequences from modern contaminants present on the bones and those potentially left by excavators, curators and scientists that handled the bones. It has been shown that most ancient animal remains yield human DNA sequences when sensitive enough amplifications are used (Hofreiter et al., 2001; Wandeler et al., 2003).