

Something old, something borrowed: admixture and adaptation in human evolution

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The sequencing of ancient DNA from archaic humans — Neanderthals and Denisovans — has revealed that modern and archaic humans interbred at least twice during the Pleistocene. The field of human paleogenomics has now turned its attention towards understanding the nature of this genetic legacy in the gene pool of present-day humans. What exactly did modern humans obtain from interbreeding with Neanderthals and Denisovans? Was the introgressed genetic material beneficial, neutral or maladaptive? Can differences in phenotypes among present-day human populations be explained by archaic human introgression? These questions are of prime importance for our understanding of recent human evolution, but will require careful computational modeling and extensive functional assays before they can be answered in full. Here, we review the recent literature characterizing introgressed DNA and the likely biological consequences for their modern human carriers. We focus particularly on archaic human haplotypes that were beneficial to modern humans as they expanded across the globe, and on ways to understand how populations harboring these haplotypes evolved over time.

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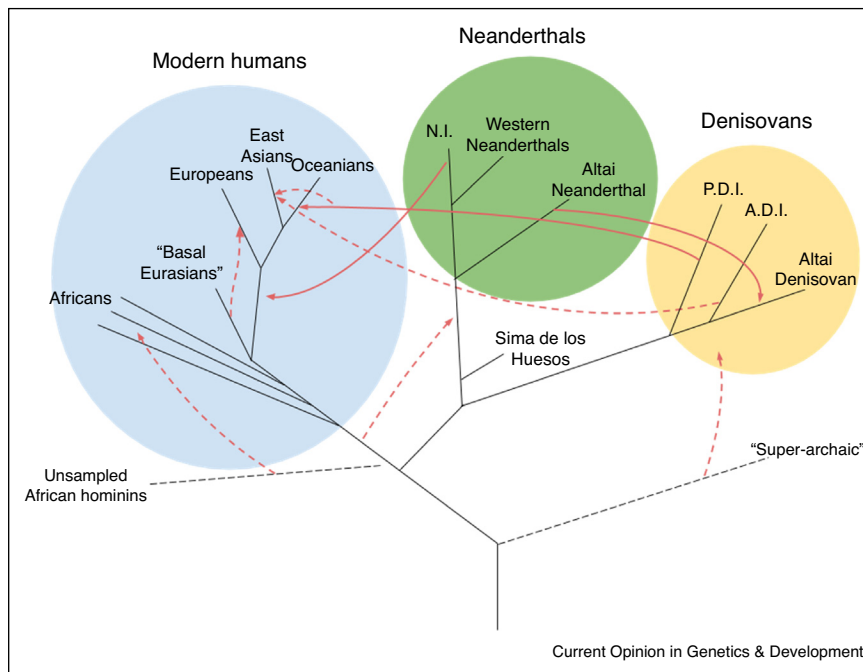
Genome-wide patterns of archaic admixture

In 2010, the first sequenced Neanderthal genome provided evidence for gene flow from Neanderthals into the ancestors of present-day non-Africans, around 50 000–60 000 years ago [1–3]. Since then, archaic human genomes have yielded ever more insightful discoveries. A few years later, a high coverage genome sequence from a

Neanderthal found in the Altai mountains allowed researchers to pin down the proportion of Neanderthal ancestry in non-Africans to be ~2% [4]. In 2017, a second high coverage genome sequence from a Neanderthal in Croatia showed that this individual was more closely related to the introgressing Neanderthal population than the Altai Neanderthal, allowing researchers to detect even slightly higher levels of Neanderthal DNA [5]. In 2018, low-coverage genomes of five additional Neanderthals living between 39 000 and 47 000 years ago allowed a first glimpse at population structure in Neanderthals and showed indications of population turnover in late Neanderthal history [6•]. But admixture between different human groups has not been limited to modern humans and Neanderthals. The genome sequence of a previously unknown group, the Denisovans (a sister group to Neanderthals), also contributed to the genomes of present-day people in Oceania, and, to a lower extent, to mainland East and South Asians [7–9,10•,11,12]. Further admixture episodes have also been suggested, including gene flow from an unsampled ‘super-archaic’ human group into Denisovans [4], from eastern Neanderthals into Denisovans [4], from modern humans into Neanderthals [13,14] and between archaic and modern human groups in Africa [15–17] (Figure 1).

Although the signals of shared ancestry between modern and archaic human groups are quite evident, the exact processes by which introgression occurred remain unclear. For example, higher levels of Neanderthal ancestry have been observed in East Asians compared to Europeans [18]. Recent work has proposed that this difference resulted from a dilution of Neanderthal ancestry in Europeans after admixture with an unsampled modern human population (‘basal Eurasians’) that had little or no Neanderthal admixture [19] (Figure 1). Others have instead suggested that the higher Neanderthal ancestry observed in East Asians is a result of additional waves of Neanderthal admixture [20–22]. Analysis of an ancient European genome has shown that at least one additional pulse of Neanderthal admixture occurred in Europe, although this modern human population does not seem to have left present-day descendants [23]. Additionally, a recent study suggests that part of the Denisovan-like ancestry found in present-day East Asians is due to an archaic group more closely related to the sequenced Denisovan genome than the Denisovan-like ancestry in South Asians and Oceanians, providing support for a two-pulse model for Denisovan-like admixture [10•] (Figure 1). In the future, more archaic human genomes may

Figure 1



Admixture events between populations of archaic and modern humans. The black tree is a highly simplified representation of the history of population splits among modern and archaic humans, including a Middle Pleistocene hominin from Sima de los Huesos, Spain, for which only limited nuclear DNA is available [77]. For the sake of simplicity, we do not include South Asians, Siberians, Native Americans and ancient modern humans — like Oase or ancient northern Eurasians — in this tree. Red arrows represent major introgression events discussed in the main text. Dashed arrows represent introgression events with only preliminary or suggestive evidence at the time of writing. N.I.: Introgressing Neanderthal population — responsible for introducing Neanderthal DNA into the ancestors of Eurasians. P.D.I.: Papuan-Introgressing Denisovan population — responsible for introducing Denisovan-like DNA into the ancestors of Oceanians (and East Asians in smaller proportions). A.D.I.: Asian-Introgressing Denisovan population — responsible for introducing Denisovan-like DNA into the ancestors of East Asians only. Figure inspired by Prüfer *et al.* 2014 [4].

help to improve our understanding of the exact complex dynamics of admixture.

Although most recent studies of modern and archaic human genomes have focused on uncovering the general history of admixture, several works have aimed to localize and characterize specific archaic human DNA tracts in present-day human genomes [22,24–28]. For example, researchers have used the genome sequence of the Altai Neanderthal to detect Neanderthal DNA in present-day non-Africans. They were thereby able to reconstruct up to 40% of the introgressing Neanderthal’s genome from the tracts remaining in present-day individuals, even though any one individual has no more than 4% Neanderthal ancestry [25–27]. They also showed that Neanderthal tracts are not uniformly distributed along the genome: large regions appear almost completely devoid of Neanderthal ancestry [25,26]. Recent work has also provided insight into the distribution of Denisovan DNA in the genomes of Oceanians [22,27], which has a similar non-uniform distribution. These ‘deserts’ — containing almost no Neanderthal or Denisovan DNA — partially overlap, and have been interpreted as evidence for

potential incompatibilities between archaic and modern human alleles. The field is now shifting its focus from finding Neanderthal and Denisovan tracts in present-day humans towards functionally characterizing them, and towards modeling their present-day distribution under different modes of natural selection in the past, with the aim of understanding the consequences of this introgression on our evolutionary history.

Admixture and purifying selection

Multiple lines of evidence suggest that negative or purifying selection against archaic human DNA has been the dominant selective force affecting the distribution of archaic human DNA in modern human genomes (Figure 2). Under a model of pure genetic drift, the average proportion of Neanderthal ancestry in modern humans should remain roughly the same over time. However, a study of late Paleolithic modern human genomes in Europe showed a progressive decrease of genome-wide archaic human ancestry with time, suggesting that archaic human haplotypes were being selectively pruned from the human gene pool over thousands of years after the introgression event [29••].

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