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The evolutionary history of human populations in Europe

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I review the evolutionary history of human populations in Europe with an emphasis on what has been learned in recent years through the study of ancient DNA. Human populations in Europe ~430–39 kya (archaic Europeans) included Neandertals and their ancestors, who were genetically differentiated from other archaic Eurasians (such as the Denisovans of Siberia), as well as modern humans. Modern humans arrived to Europe by ~45 kya, and are first genetically attested by ~39 kya when they were still mixing with Neandertals. The first Europeans who were recognizably genetically related to modern ones appeared in the genetic record shortly thereafter at ~37 kya. At ~15 kya a largely homogeneous set of hunter-gatherers became dominant in most of Europe, but with some admixture from Siberian hunter-gatherers in the eastern part of the continent. These hunter-gatherers were joined by migrants from the Near East beginning at ~8–9 kya: Anatolian farmers settled most of mainland Europe, and migrants from the Caucasus reached eastern Europe, forming steppe populations. After ~5 kya there was migration from the steppe into mainland Europe and vice versa. Present-day Europeans (ignoring the long-distance migrations of the modern era) are largely the product of this Bronze Age collision of steppe pastoralists with Neolithic farmers.

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Introduction

The history of human populations in Europe has been studied more extensively than any other continent. Europe is the place where the earliest Neandertal specimens were discovered, pointing to the existence of people in the past who were morphologically distinct from its recent inhabitants. Many other remains were unearthed soon thereafter, and the nascent discipline of physical anthropology was applied to them, inaugurating the scientific, quantitative study of both modern and ancient human populations. During the 20th century anthropology was joined by the study of genetics. The present review focuses on the last few years of this field of study, and in particular on the insights into human history provided by ancient DNA, a powerful new tool in the kit of the prehistorian and evolutionary biologist.

Archaic Europeans

The oldest sampled nuclear DNA from Europe dates to ~430 kya from Sima de los Huesos in Spain and it was found to be more closely related to Neandertals than to Denisovans [1••], unlike mtDNA from the same population, which formed a clade with Denisovan mtDNA [2]. Genome-wide data from several European Neandertal individuals has been published down to ~39 kya [3,4]. The Neandertal population that contributed DNA to all non-Africans was more closely related to European Neandertals than to an early Neandertal from the Altai region in Siberia estimated genetically to be ~120 kya old [3]. Thus, the greater part of the history of European populations was dominated by the Neandertals and their ancestors, raising questions about why this population was replaced in what amounts to a geological blink of an eye.

The genetic divergence between modern humans and Neandertals is lower-bounded by the finding that the Sima de los Huesos hominins already belonged to the Neandertal lineage [1••] which must therefore have been already in existence by ~430 kya. Modern human and Neandertal Y-chromosomes shared a most recent common ancestor ~450–800 kya [5] also pointing to an earlier split of the two lineages. Neandertal mtDNA shared a most recent common ancestor ~270 kya, with the earliest known split represented by a specimen from Hohlenstein–Stadel in Germany [6]; the common ancestor of Neandertal and present-day human mtDNA was dated to ~300–500 kya [7], appearing to be younger than the corresponding Y-chromosome most recent common ancestor, suggesting that some gene flow may have occurred between the ancestors of modern humans and Neandertals after their separation.

A remarkable recent technical development is the discovery of the fact that mammalian mtDNA is preserved in cave sediments [8••] and its application to the study of archaic hominins from Europe and Siberia. Sediment DNA allows one to detect the presence of humans in sites where they may be archaeologically invisible and to

obtain DNA from the deep past where hominin remains may be scarce or too precious to submit to destructive sampling. A potential limitation is contamination with modern human DNA, which may make this technique more applicable to extinct lineages that could not have plausibly been contributed by modern humans.

Upper Paleolithic Europeans

It is only by ~39–36 kya that the first sample that clearly shares ancestry with Europeans but not East Asians is evident (Kostenki14 in European Russia [9]), with the earliest known such sample from western Europe at ~35–34 kya (GoyetQ116-1 from Belgium [10••]). Did these and other early Europeans [10••,11•] represent a migration into Europe post the Campanian Ignimbrite (CI) volcanic eruption [12] ~39 kya, or are they survivors of this event which set off a short period of intense cooling? The ~42–37 kya sample from Oase1 in Romania is the earliest known modern human from Europe, and may predate this event. Oase1 has an excess of 6–9% Neandertal ancestry within a genealogical timeframe of 4–6 generations and no specific affinity to Europeans [13], suggesting that at least some of the pre-CI Europeans were replaced after this event. It may be that both the modern human and Neandertal inhabitants of Europe suffered a common demise ~39 kya.

Intriguingly, GoyetQ116-1 and Kostenki14 — two of the earliest samples on the lineage leading to later Europeans — were not symmetrically related to non-Europeans, with GoyetQ116-1 being genetically closer to a ~40 kya sample from China (Tianyuan [14]). In a similar vein, mtDNA haplogroup M, rare in Europe today, but common in eastern non-Africans (East Asians, Oceanians, and Native Americans) was present in pre-Last Glacial Maximum Europeans [15]. Surprisingly, this eastern non-African affinity disappeared in samples from the Gravettian-associated 'Vestonice-cluster' ~31–26 kya which included samples from Italy, Belgium, and the Czech Republic, but partially re-appeared in the ensuing Magdalenian-associated 'El Mirón Cluster' ~19–15 kya known from sites in Spain, France, Germany, Belgium and Germany [10••].

Western European hunter-gatherers (WHG), first described in three sites of western Europe [16–19] are now known to also have lived in southeastern Europe [20,21•], Switzerland [22], the Baltics [21•,23•], and Italy [10••]; the early example from Villabruna in Italy ~15 kya [10••] has given this population the alternative name 'Villabruna cluster'. The appearance of WHG ~15 kya corresponds to the Bølling-Allerød interstadial warm period, and marked a genetic attraction of European and Near Eastern populations [10••,24••]. Was this due to migration between Europe and the Near East during this favorable climatic period or due to the expansion of related populations in Europe and the Near East that had been established there at an earlier period [10••]? WHG-like ancestry may represent a partial source of ancestry of populations bordering Europe, for example in Anatolia whose early farmers ~8 kya are genetically closer to WHG than other Near Eastern populations are [24••], or in the Atlantic where pre-colonial Guanche inhabitants of the Canary Islands had some European hunter-gatherer affinity in addition to their mainly North African origin [25]. WHG did not, however, appear to make any quantifiable genetic contribution to the Upper Paleolithic inhabitants of geographically proximate Morocco ~15 kya in North Africa [26].

Eastern European hunter-gatherers (EHG), a population of mixed WHG and Upper Paleolithic Siberian ancestry (related to the Mal'ta and AfontovaGora specimens from Lake Baikal [10••,27] ~24–17 kya) are attested in European Russia ~8 kya [28,29]. This group contributed ancestry to hunter-gatherers in Sweden ~8–5 kya [16,30], Norway [31•], the Balkans and Ukraine [20,21•], and the Baltic [21•,23•,31•,32•,33] . The spread of this ancestry across northern Europe was followed by >3.5 kya by the spread of Siberian ancestry [34] that seems to be associated with Finno-Ugrian speakers [16].

First farmers

The dominance of the WHG across much of mainland Europe was relatively short-lived, as they were largely replaced beginning in the 7th millennium BC by farmers from Anatolia via southeastern Europe [29,35•], who minimally, but variably, mixed with incoming farmers in southeastern Europe [21•] and propagated their ancestry as far as Scandinavia [30,36] and Iberia [28,37]. The WHG populations were, however, persistent, with individuals of predominantly WHG ancestry found in early Neolithic contexts in Hungary [19] and as late as the 4th millennium BC in the Blätterhöhle site in Germany [38•,39]. These WHG survivors engendered a resurgence of hunter-gatherer ancestry across Middle Neolithic Europe [28] which appears to have involved local populations of hunter-gatherers [38•] rather than a migration from a single WHG-rich area.

Over the Ice Age, Neandertal ancestry seems to have been reduced in Europe [10••] by natural selection against Neandertal variants [40,41], although these were occasionally adaptive [42]. A further reduction of Neandertal ancestry was effected during the Neolithic period by migration from the Near East whose populations descended in part from a postulated group of 'Basal Eurasians' [16,24••], a population that split off from other non-Africans before they split off from each other. Basal Eurasians are consistent with having no Neandertal ancestry at all [24••] and may have split from other non-Africans ~101–67 kya [43]. It is unknown whether the Basal Eurasians represent descendants of early modern humans in the Near East, or a later entrant into the