Many of us are fascinated by narratives regarding the origin and evolution of our species. Who are we? How did we people the world? Answers to these simple questions remain elusive even though researchers have been quite successful in describing past human morphology and culture using evidence from anthropology, archaeology, history, sociology and linguistics. However, when they address human migrations, archaeologists are somewhat restricted to surviving artifactual evidence and limited to descriptions of culture expansions, which may have occurred by the movement of either ideas or people. The advent of genomics, by which one can sequence whole or part of an individual's DNA, provided a powerful means to dig into past human demographic history. Notably, the coalescent theory posits that individuals in a population share genetic variants that originated from a common ancestor. This powerful theory is the basis for a number of bioanalytical innovations that utilize genetic data to reconstruct human movements around the world.

**From classical archaeology to ancient DNA**

During the last decade, the application of high-throughput DNA sequencing technologies to DNA extracted from archaeological remains has further revolutionized our understanding of human expansion and recent evolution. So-called ancient DNA (aDNA) provides accurate genetic snapshots of populations at a given time and place, most of which are confounded in modern-day populations with subsequent demographic events such as admixture, population collapse or migration. Crucially, aDNA also enables accurately calibrating the molecular clock of human evolution. Not only can we identify splits between populations and infer migrations, we can also provide precise temporal context to these events.

**Figure 1.** Early movements of modern humans after they left Africa. Red dots are archaeological sites mentioned in the text, and arrows are only an approximation of the true migration process. The insets represent Beringia (top) and Sahul (bottom) with mainland extensions at the time of first human presence in these regions in light grey. LGM, Last Glacial Maximum.
Equipped with the aDNA toolbox, researchers have now explored most regions of the world and tested hypotheses laid out by archaeologists, anthropologists and linguists. In all cases, aDNA findings were quite surprising (Figures 1 and 2).

Out of Africa and early non-African divergences

Africans have the highest levels of genetic diversity of any contemporary human population, and the oldest splits between human populations are found in sub-Saharan Africans. This evidence suggests that the origin of all anatomically modern humans (hereafter simply referred to as modern humans) is in Africa, although the exact location is still unknown.

Genetic studies of contemporary populations support an expansion of modern humans out of Africa around 65–55 thousand years ago (65–55 ka). Ancient genome sequences from extinct Neanderthals—archaic humans present in western and central Eurasia until 40 ka—revealed that all non-African populations have ~2% Neanderthal ancestry, supporting a single out-of-Africa event rapidly followed by admixture with Neanderthals. Furthermore, some populations in Asia and Oceania carry 3–6% Denisovan ancestry—Asian archaic humans identified through aDNA only. Admixture with archaic humans reveals the earliest split among non-African populations 60–45 ka: one lineage colonized mainland Eurasia, the other colonized New Guinea and Australia. The genomes of early Eurasian individuals such as Ust’Ishim (45 ka), Kostenki (37 ka) and Tianyuan (40 ka) further constrain a split between western Eurasia and East Asia/Oceania to 55–45 ka.

Eurasia

Modern humans were widely distributed in Europe as early as 45 ka, but this early Palaeolithic European ancestry is virtually absent in contemporary populations. Three subsequent migration waves contributed to the modern-day European genetic makeup. First, Palaeolithic populations were replaced by Mesolithic hunter-gatherers coming from southern European and central Eurasian refugia ~14 ka, after the cold and arid conditions of the last Ice Age (also known as the Last Glacial Maximum (LGM); 28–18 ka). Second, the Neolithic farming populations of Anatolia, the Levant and northern Iran expanded throughout Europe ~8.5 ka and admixed with the Mesolithic hunter-gatherers. The Neolithic lifestyle (animal husbandry, agriculture and sedentarism) helped increase population size, but health became poorer. Third, Bronze Age herding populations migrated from the Eurasian steppe to central Europe ~5 ka. These Yamnaya and Afanasievo people already had a mixed ancestry related to various Russian and Caucasus hunter-gatherers. The Steppe migration revolutionized European technology with the notable introduction of the wheel and may have spread Indo-European languages.

aDNA from central and South Asia revealed complex patterns of migrations similar to Europe, where local Upper Palaeolithic hunter-gatherers admixed
with, or were replaced by, subsequent migrants from western Eurasia, including the Steppe expansion ~5 ka. In East and Southeast Asia, current Austroasiatic speakers exhibit a mixture of East Asian agriculturalist and eastern Eurasian hunter-gatherer ancestry. Present-day Japanese people result from an admixture between the indigenous Jomon population and later East Asian agriculturalists.

**Island Southeast Asia and Oceania**

Modern humans had probably crossed the Wallace line—the biogeographical zone that separates Asian and Oceanian ecoregions—by 55 ka in an initial single wave to colonize Island Southeast Asia and Sahul (a prehistoric continent that consisted of New Guinea and Australia). These ancestors of Australo-Melanesian speakers (Papuans, Aboriginal Australians, Torres Strait Islanders, Melanesians and Island Southeast Asia Negrito populations) underwent further genetic diversification and adaptation to new environments. Papuans and Australians represent some of the oldest continuous indigenous cultures on earth due to isolation until European arrival. Island Southeast Asians west of the Wallace line further admixed with ancestors of Austroasiatic speakers from a mainland East Asian expansion ~10 ka. Then, an expansion associated with the Lapita culture spread the Austronesian languages and agriculture into Island Southeast Asia and Polynesian islands ~5–3 ka, further expanding as far as Hawaii and Easter Island. aDNA from Lapita individuals revealed they are highly related to present-day Taiwanese and have no Australo-Melanesian ancestry.

**Siberia and Beringia**

Ancestral North Siberians (represented by an individual from the Yana River, ~31 ka) diversified ~38 ka, soon after the basal split between western Eurasians and East Asians. The population did not contribute to present-day Siberians or Native Americans. During the LGM, Beringia was the land bridge exposed by lowered sea levels that connected northeastern Siberia and Alaska. Because northern Siberia was uninhabited due to adverse climatic conditions, humans who ventured into that region of the world were driven to a refugium in southeastern Beringia. However, the ice sheets that covered northern North America obstructed their dispersal further east, impeding access to the American continent. aDNA shows the isolated population had an Ancestral North Siberians ancestry and admixed with East Asians. This admixture resulted in two different lineages that diverged around 20 ka: the Ancient Palaeo-Siberians and the Ancestral Native Americans. The genetic legacy of Ancient Palaeo-Siberians is currently restricted to populations in northeastern Siberia, whereas the direct ancestors of present-day Siberians were the Neo-Siberians, a population related to East Asians that expanded northwards ~10 ka.

**America**

The scenario of a southeastern Beringian refugium during the LGM is compatible with the Beringian Standstill Hypothesis, which proposes that Ancestral Native Americans were isolated for 2400–9000 years, giving rise to most of the non-Arctic Native American ancestry. Ancestral Native Americans entered North America ~25–15 ka, when ice sheets started to melt. This population split into two lineages either in Beringia or in North America ~17–15 ka: Northern Native Americans (represented by the Anzick individual—~12 ka—associated with the Clovis culture). The peopling of America was swift and humans reached southern South America as early as ~14 ka (Monte Verde, Chile). Furthermore, a Siberian-related population described as Palaeo-Eskimos expanded into the Arctic ~5–4 ka. Palaeo-Eskimos were replaced by Neo-Eskimos, ancestors of the current Inuits, around 700 years ago.

**Africa**

Due to poor preservation conditions, aDNA studies have been challenging in Africa. Arguably, the main post-Out-of-Africa demographic event was the agricultural expansion of the Bantu-language population from the Highlands of Nigeria and Cameroon into sub-Saharan Africa around 4 ka. Bantu migrants generally admixed with local hunter-gatherers and in some places totally replaced the population. Additionally, two previous streams contributed to shaping the current African substructure: the migration of pastoralist populations from South Sudan to East and Central Africa ~7 ka, and the migration of agropastoralists from Ethiopia to Kenya and Tanzania ~5 ka.

**aDNA and detection of adaptation**

Genetic variants that confer better survival and reproduction tend to increase in frequency in a population and dominate over other variants. These adaptive variants leave distinctive genomic selection signatures in the form of unusually long genomic regions of low genetic diversity, where the variant and its surrounding genomic region increase in frequency in the population (Figure 3, left panel). Demographic processes such as admixture and bottlenecks may
potentially mask selection signatures (Figure 3, right panel), but aDNA is a powerful way to examine genomes for adaptive variants before known selection pressures, admixture and other demographic events occur.

As modern humans left Africa and peopled the world, they encountered new climatic conditions, food sources and pathogens. The Neolithic transition (notably the domestication of animals that carry zoonotic pathogens), the more recent Western Colonization and Industrial Revolution have also created drastic cultural and environmental changes. A major example of adaptation is the evolution of light skin pigmentation in western Europe. Originally, Palaeolithic Eurasians and Mesolithic Western Europeans carried variants linked to darker skin. Thanks to aDNA studies, we now know that some variants linked to lighter pigmentation were introduced by Neolithic farmers and Bronze Age herders. These only became widespread in Europe in the last 5000 years as a likely adaptation to counteract vitamin D deficiency due to lower levels of UV radiation at higher latitudes.

Another very notable example is the adaptation for lactase persistence in Europe, which enables milk digestion after weaning and into adulthood. This variant shows one of the strongest signatures of selection in European populations. aDNA studies found this allele was rare (although present) in populations from Early Neolithic until the Bronze Age, and then very rapidly increased in frequency to ~70% in present-day northern Europe. The sudden rise in frequency comes at least 3000 years later than the archaeological record of dairy practices.

The evolution of height and the immune system are also the target of aDNA researchers, although these traits are challenging to study due to their highly polygenic nature.

**Limitations and future of aDNA**

aDNA research is limited by the availability of well-preserved human remains, and there are obvious ethical considerations regarding the destruction of samples for DNA studies. aDNA research also suffers from evolutionary genetics theoretical restrictions: divergence dates of genetic lineages can be inferred up to their most recent common ancestor, but we are blind to potentially extinct lineages that might have split earlier. For that reason, archaeological evidence of very early human presence in some regions of the world (Africa, Australia, Sumatra, China and the Arabian Peninsula), which seems

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**Figure 3.** Left panel: detecting genetic signatures of selection in a ‘perfect’ world, i.e., in the absence of demographic processes that may break up the hallmarks of selection. A genetic variant (red and yellow star on a black genomic background) is selected for. The variant and its surrounding genomic sequence reach fixation in the contemporary population, so that all individuals have reduced diversity in this genomic region.

Right panel: detecting signatures in the ‘real’ world is complicated by demographic processes. A population bottleneck reduces overall genome-wide diversity, making it more difficult to identify a region under selection (i.e., with low diversity). Admixture introduces new genomic backgrounds, also breaking up the signs of reduced diversity, even if the admixing population carries the same selected variant. aDNA allows observation of selection signatures occurring before demographic processes may disrupt the signal.
to predate diversification of currently known genetic lineages, might simply represent extinct populations that did not contribute ancestry to subsequent populations.

Ancient genetic data generated fascinating and often surprising discoveries about our past, but, as is the case for genetic results in general, great care must be taken when interpreting findings. While genetic studies involve grouping people into populations and observing the genetic differences between them, this cannot support racial or nationalistic ideologies. Nevertheless, aDNA is an invaluable tool to describe indigenous genetic diversity before the impact of European colonialism and more recent globalization. Beyond providing irrefutable genetic evidence of colonization-linked indigenous population collapse such as in South America, aDNA also provides useful evidence to repatriate indigenous remains locked in museums to where they belong. With the advent of personalized medicine, aDNA data may also help improve the health of indigenous people.

Exciting complementary disciplines that inform human evolution have recently emerged from the study of aDNA. Palaeomicrobiology, where genomes of ancient pathogens and microbes can be sequenced, provides invaluable insights about historical pandemics (such as the Black Death), but also the general health and lifestyle of past people. Similarly, palaeoepigenetics—which investigates DNA modifications that regulate gene expression—can contribute knowledge about morphology, age, lifestyle and eventually the environment of past individuals. There is definitely a future to aDNA research.

Further reading


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