Earth Sciences

How Ancient Genomics Unveils the Prehistory of Humans

Since a decade ago, the field of ancient DNA has flourished, with unprecedented success in sequencing genome-wide data from ancient specimens. The majority of the research has focused on past humans, both modern ones with a close relationship to one or more present-day populations as well as archaic ones that no longer live today, such as Neanderthals.

With a growing pool of genomic data from past humans from many different times and locations, it is increasingly possible to study the large-scale patterns of human prehistory and associated trends over time and space. A new review [1] published in *Trends in Genetics* (https://doi.org/10.1016/j.tig.2017.11.008), produced by Professor FU Qiaomei and Dr. Melinda A. Yang at the Molecular Paleontology Lab of the Chinese Academy of Sciences Institute of Vertebrate Paleontology and Paleoanthropology, contextualizes much of the research to date, providing a comprehensive understanding of major features of human genetic prehistory.

Their review covers the genetic patterns and relationships that have been uncovered regarding human prehistory over the last few years, focusing on ancient genetic studies of ~45,000- to 7,500-year-old humans from primarily pre-agricultural cultures.

Analysis of ancient DNA from ~45,000- to 35,000-year-old modern humans show how diverse ancient humans of Eurasia were. These, the oldest modern humans that have been sequenced thus far, illustrate that by this time, multiple distinct populations were present. Some contributed to present-day populations – individuals from Belgium and Russia show the closest relationship to present-day Europeans and an individual from northern China shows the closest relationship to present-day East Asians. However, other individuals highlight lost populations – at least two individuals from this time period represent populations that did not contribute to any present-day Eurasians.
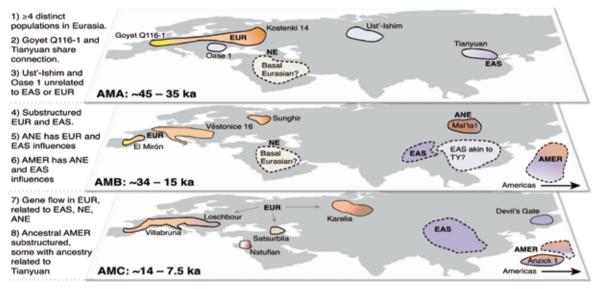
In the period bracketing the Last Glacial Maximum, population subdivision, movement and partial or complete replacements are a common theme in regional human prehistory. In West Eurasia, this is directly shown

using specimens from ~35,000 to 14,000 years ago. An individual found in Belgium ~35,000 years ago possesses ancestry that is not observed in the immediately succeeding European individuals sampled, but that does recur in partial amounts in some Europeans from ~19,000 to14,000 years ago. In East Eurasia, individuals with very early European-like ancestry are found in the Lake Baikal region ~24,000 to 17,000 years ago, and while this population no longer exists today, a large proportion of Native American ancestry derives from these "North Eurasians."

As climatic conditions warmed after the Last Glacial Maximum, by ~14,000 years ago, more connections appeared across Eurasia. Ancient Europeans show connections to present-day populations in East Asia and the Near East, some ancient eastern European individuals possess partial "North Eurasian" ancestry, and individuals in the Caucasus begin to show partial ancestry related to a "Basal Eurasian" population for which no direct samples are available, but whom separated very early from other non-Africans and are partially represented in all recent Europeans and Near Easterners.

Research focused on the phenotypes associated with different genetic variants also reveals some surprising results, such as Europeans as recent as 7,500 years ago still possessing dark skin, and blue eyes occurring as early as 14,000 years ago. Also, the ~7,500-year-old individual of European ancestry with dark skin from Spain also possesses blue eyes, a combination not observed today.

The most comprehensive research thus far is in West and North Eurasia, with a small but growing pool of research focused on ancient humans from East Asia and the African and American continents. A common thread across all the research is that human prehistory in different regions of the world have a complex history of movement, separation and interaction that can be discerned from the genetics of ancient humans.

On a different trajectory, the authors also ask what the study of the genetics of ancient modern humans has contributed to our understanding of the relationship and interactions between modern and archaic humans.

Schematic from the Yang and Fu review depicting the sampled and proposed populations in Eurasia (and the Americas) from ~45 to 7.5 thousand years ago (ka), separated into three time periods: Ancient Modern A, B and C (AMA, AMB, AMC). Colors represent distinct ancestries; EUR refers to European-related ancestry, NE refers to Near East-related ancestry, EAS refers to East Asian-related ancestry and AMER refers to the Native American-related ancestry. Dashed lines indicate no samples are available, and other labels refer to key specimens.

Several early modern humans have been used to confirm interbreeding between modern and archaic humans did occur. By searching for chunks of Neanderthal sequence in these early modern humans, and studying their lengths, multiple studies have confirmed a date of gene flow from Neanderthals to all non-African modern humans around 60,000 to 50,000 years ago.

Studies of the genetics of archaic and modern humans highlight that multiple archaic admixture events occurred in human prehistory. A stunning find in Romania of an ~39,000-year-old individual with a Neanderthal ancestor only 4 to 6 generations previous indicates an additional archaic admixture event, and a connection between some present-day Oceanian populations and the Denisovans, an archaic human closely related to but distinct from Neanderthals, is yet another. Varying levels of archaic Denisovan and/or Neanderthal ancestry in different present-day populations hint at the possibility of even more archaic admixture events.

While analysis of a few ancient individuals is incredibly useful, the review also emphasizes the value of studying many ancient individuals from several different points in time. The review highlights earlier work showing that the archaic admixture proportion in Eurasians gradually declines over time, suggesting most archaic DNA in modern humans is under negative selection and is slowly removed from modern human populations by purifying selection.

Just as important as highlighting the patterns that have been observed thus far studying the genetics of ancient modern humans, their review also emphasizes where sampling is needed and asks key questions. Greater sampling in regions outside West Eurasia will help to better characterize the human population dynamics and types of archaic admixture across the globe. Increasing samples from the same region and time period allows study of the frequency of variants in past populations, which will aid in understanding population continuity and adaptation to their environment. Past ancestry represented in partial amounts in recent populations hint that sampling past specimens will uncover where and when those populations lived. While the fruits of labor have been plentiful, as demonstrated by this review, there is yet a long but fulfilling road to walk towards understanding the genetic prehistory of humans.

References

Yang, M.A., and Fu, Q. (2018). Insights into modern human prehistory using ancient genomes. *Trends in Genetics*.