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Around the World in Four Millennia

How Prehistoric Humans Spanned the Globe

By Megan Bartlett

Traditional genetics research centered around the detailed study of a few model organisms, coloring the public imagination with images of overflowing greenhouses of peas, thickly spread plates of *E. coli*, and jars of oddly shaped fruit flies. This sharp focus enabled scientists to elucidate the fundamental concepts of genetics, but the questions that geneticists are tackling now require a larger scope of study. The population geneticists and paleontologists who map the prehistoric migrations of modern humans that led to our global colonization have expanded their evidence to include such diverse items as mummified remains found in peat bogs, the Burushaski language isolate spoken in Pakistan, the painted caves of Altamira, Spain, and blood collected from Bantu tribesmen. Vastly improved techniques for genetic analysis have allowed researchers to use small sequences of DNA to extrapolate far beyond the sparse fossil record, replacing former speculation with solid biological evidence.

Genes from Mother, Genes from Father

The human genome has over 10 mil-

lion sites where genetic composition can vary, yet most genes tell us little about the connection between their carriers. Nonetheless there are two types of genetic material can be traced because they are only passed on in one simple path: mitochondrial DNA (mtDNA) and the Y chromosome.

In addition to the DNA in the cell nucleus that controls most biological processes, there are also small amounts of DNA inside the mitochondria, the cellular organelles that produce the cell's energy (Figure 1). Mitochondria are present in egg cells, where they are passed on to the offspring, but they are not transferred by sperm. As a result, all of the offspring's mitochondria are derived entirely from those donated by the mother, creating a genetic lineage traced through the female line (1).

The fitness impact of mtDNA on the organism is so small that it is almost free from evolutionary selective pressures, a state known as neutral selection. However, random mistakes in copying accumulate over time and produce changes in the mtDNA sequence, differentiating some lineages from others. Because these changes do not affect the survival of the organism, they are pre-

served in the mtDNA lineage and can be sequenced and compared. Groups that are separated and do not interbreed will have different mtDNA mutations, while members of the same group will share mutations. So, lineages that share more sequences of mtDNA have had been separated for less time, indicating that they shared a female ancestor more recently.

The male lineage can be traced similarly through the Y-chromosome, which is only passed from fathers to sons. The principle behind both is the same: both types of genetic material accumulate random changes over time with a fairly constant rate of mutation, enabling scientists to estimate the time required for differentiating mutations to have taken place; from that, the most recent date when two subjects shared an ancestor, as in Figure 2, can be predicted (2). By collecting data from many subjects and comparing their relatedness, divergences in the prehistoric human population can be detected and used to trace the migrations that brought early humans out of Africa and across the globe.

In the Beginning

Before the era of genetic analysis of-

credit: adapted from Ref. 10

ferred concrete proof, paleontologists and anthropologists were caught in a vicious stalemate on the question of where modern humans evolved. Those supporting the “out of Africa” theory held that the modern human species *Homo sapiens* had evolved in Africa, then spread outward to the rest of the world. Those pushing forth the multi-region hypothesis suggested that an earlier hominid species, *Homo erectus*, developed in Africa and spread outward to the other continents, and the separated populations either evolved to *Homo sapiens* or became extinct.

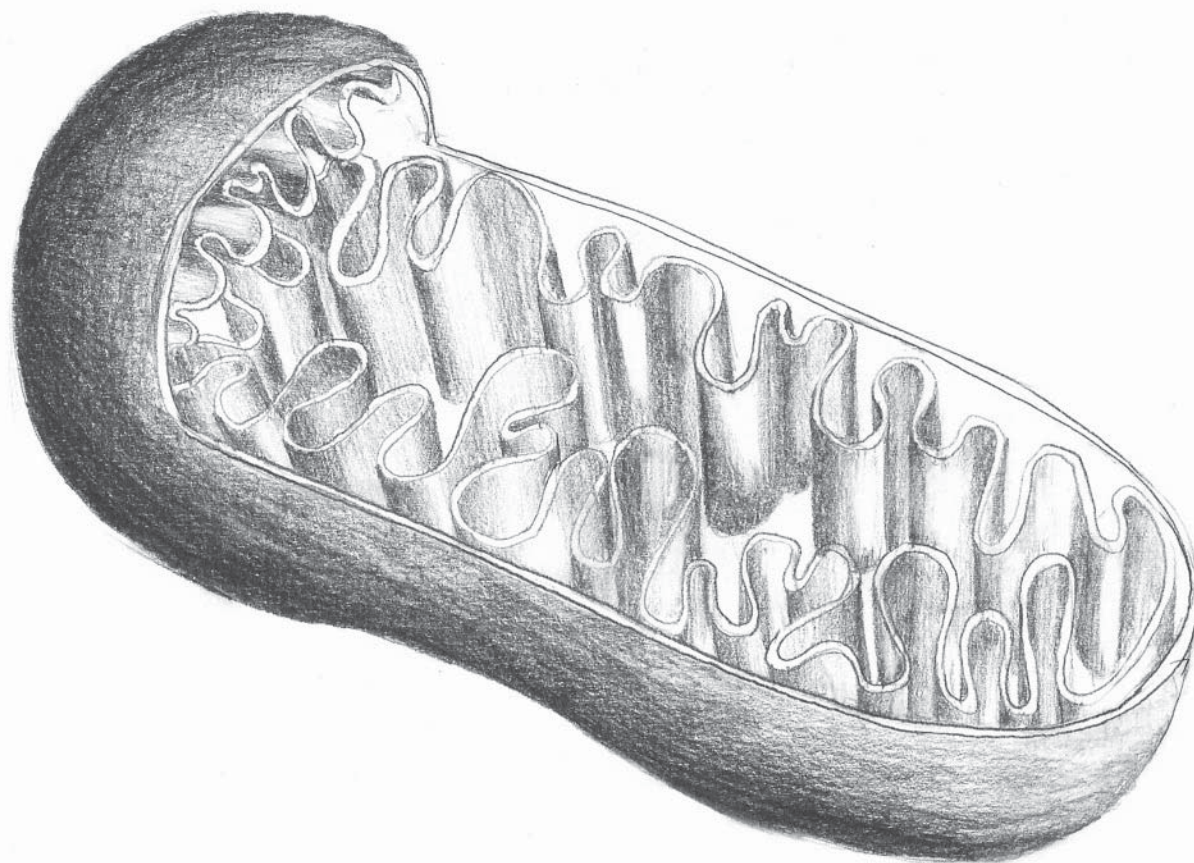
Genetic studies beginning in the 1970s threw new evidence into the debate, tipping the balance of proof far to the side of the Africa hypothesis. The European Neanderthals were rejected as the ancestors of modern humans, Native Americans were shown to have derived from a prehistoric *Homo sapiens* population in Asia, and perhaps most surprisingly at the time, large-scale

genetic sampling showed far more genetic variation within races (85% of total variation) than between them (7%), violently overturning the possibility of multiple isolated appearances of the modern human species (4). The site of this original evolution was chosen to be Africa because the oldest known hominid fossils have been discovered there. Furthermore, Africa contains only 13% of the world’s population, but 40% of its genetic diversity, suggesting a far longer period of human divergence and evolution than the rest of the world (5).

The “out of Africa” hypothesis drastically restructured the human pedigree, allowing every human alive today to be traced back to a common ancestor in Africa. Analysis of mtDNA and the Y chromosome have produced two ancestors, known as Mitochondrial Eve and Adam. Mitochondrial Eve is the most recent common female ancestor of all living humans, traced through 150,000

years of mitochondrial transfer from mothers to daughters. She was certainly not the only human female alive at her time, but she was the only one who can claim everyone alive today as a descendant (5). In complement to Mitochondrial Eve, studies of the Y chromosome have revealed an Adam, but Adam is placed in Africa only 59,000 years ago – “Adam” and “Eve” do not represent the first humans, and they certainly never met (6)!

Why do these dates conflict? Different parts of the human genome evolved separately from each other, with unique mutation rates and patterns of inheritance. The lineage of one gene does not necessarily match the lineage of another, or of a species as a whole, allowing mtDNA and the Y chromosome to converge at disparate times as a result of “thousands of years of sexual politics” (4). Men have more variation than women in their reproductive success, ranging from no children to hundreds,



Credit: Chelsea Gordon

Figure 1. Mitochondria in which Mitochondrial DNA is found.

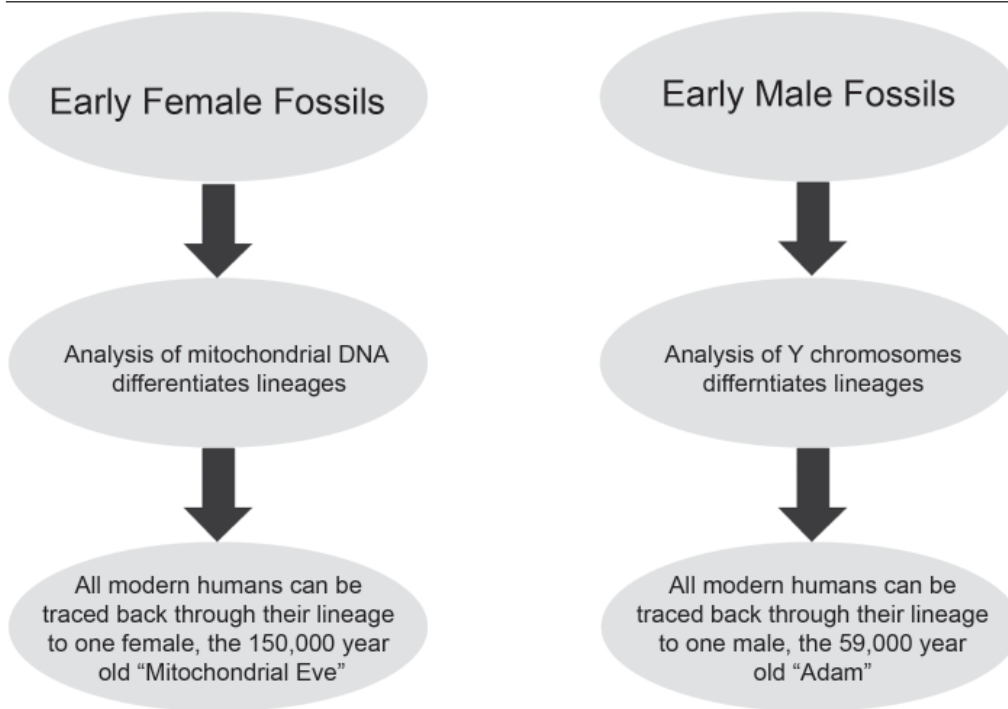


Figure 2. Analysis of mitochondrial DNA and Y chromosomes from early fossils help scientists trace humans back to their common ancestors.

making it easy for some Y chromosome mutations to dominate while others are eliminated (7).

Modern Humans

The *Homo erectus* populations that remained in Africa slowly evolved, with *Homo sapiens* diverging as a separate species by 200,000-130,000 years ago (2,8). However, the *Homo sapiens* population crashed from 100,000 individuals at 200,000 years ago to only 10,000 individuals at 100,000 years ago, barely surviving an episode where some scientists argue “they were as endangered as pygmy chimpanzees or mountain gorillas are today” (3). The blame for this devastation has been pinned on climate change, with a dramatic peak in cold temperatures sealing more water in the ice caps and reducing precipitation, expanding the Sahara and Kalahari Deserts while shrinking the human-inhabited grasslands. The impassibility of the deserts, coupled with the extremely low population, trapped the early humans in Africa for millennia. Ironically, the inhospitable weather may have forced the early humans to develop the technology and social

structures that would enable them to adapt to harsher conditions, leaving them well prepared for the return of warm, moist climate that enabled them to leave Africa (3,9).

By 50,000 years ago, some early humans in Africa had found a more welcoming habitat than the central grasslands – the seacoast, rich in wildlife, vegetation, and seafood. The move to the coast allowed a dramatic population expansion and opened up a new pattern for migration (10, 11). Rather than slog through inland deserts, forests, or mountains, adapting to each new environment in turn, early humans followed the relatively unchanging coastal regions to new continents. Scientists have followed their movements through DNA markers, culminating in what is known as the coastal migration model.

All Eurasian mtDNA lineages can be traced to one woman in Africa who lived 50-60 thousand years ago, known as L3, or less prosaically, Eurasian Eve. She has a parallel in her contemporary Eurasian Adam, known as M168, tracked through the Y-chromosome. It is the divergences in their descen-

dants that tell us the most about how humans migrated: the L3 lineage splits into M and non-M mtDNA types, while the M168 lineage separates into M130 and non-M130. M is never found in the Middle East and Europe, but it occurs in 20% of Indians and virtually all Australians, while M130 is confined to eastern Asia and America, and totally absent west of the Caspian Sea (4).

This points to two lineages that migrated at different times. The M and M130 lineages left Africa first. They hugged the coast through the Middle East, to the southernmost tip of India, then split to either travel north into China or navigate Indonesia and Malaysia to reach Australia as early as 60,000 years ago (12).

Their passage can be traced by the remarkable physical and genetic similarity between Africans and some isolated populations like those on the Andaman Islands in the Bay of Bengal (9). However, their dependence on the coast limited the range of their genetic distribution to Australia and scattered parts of southeast Asia.

Most humans are descended from the second lineage that left Africa thousands of years later, known as M89. These humans entered the Sahara Desert during a wet, warm climate period, and were forced to emigrate when conditions became dry, pushing them into the Middle East and trapping them there (6). From the group in the Middle East, another lineage (M9) split off to venture into Central Asia. However, the immense mountain ranges that cut across central Asia divided this lineage into M20, which moved south to India and Pakistan, and M45, which traveled north of the Hindu Kush mountains to spread through central Asia (4). The M45 line then gave rise to the M175 marker, which is found only in East Asia, and accounts for 60-90% of Asian men living east of the Himalayas. However, many of the remaining

genetic lineages in East Asia are M130 – the coastal migrants. It is likely that the M130 coastal migrants changed direction to travel north along the coast or spread inland from southern India, and mixed with the M175 humans when they later arrived (4).

Europe

It seems logical that the first Europeans would have come from a group of Middle Eastern humans that moved northwest. However, the Middle Eastern lineages are extremely rare in Europe. Instead, most isolated populations of Europeans (who would be more likely to retain the older Paleolithic markers), such as the Celts and Basques, carry M173, which is derived from M45. Members of the central Asian clan spread into Europe from the steppes of Siberia about 30,000 years ago, perhaps because they needed the thousands of years of cultural evolution in the harsh Siberian environment to survive the climate of Europe (4). Both the Y chromosome and mtDNA attribute about 80% of Europe's genetic frequency to Paleolithic migration, and mtDNA analysis segregates modern European lineages into seven clusters of female ancestors, known as the Seven Daughters of Eve. The Paleolithic clan mothers are known as (in order of appearance of their genetic markers): Ursula, comprising 11% of Europeans, mostly Britain and Scandinavia; Xenia, 6% of Europeans with three distinct branches in eastern, central, and western Europe; Helena, 47% with descendants all over continental Europe; Velda, 5%, in western and far northern Europe; Tara, 9%, Mediterranean, Britain, and Ireland; and Katrine, 6%, around the Mediterranean (5). The seventh lineage is the one Neolithic mtDNA lineage, and possibly responsible for bringing agriculture to Europe. This clan mother is known as Jasmine, and her descendants account for 17% of Europe, following a definite pathway along the Mediterranean coast to Spain,

then up to western Britain.

The Americas

The ancestry of the Native Americans has proven to be relatively straightforward – however, it is still a mystery how and when those ancestors entered the Americas. The M45 lineage in Siberia gave rise to the M242 marker, which is the oldest marker in the Americas, about 20,000 years ago. This clan could easily have followed the steppes to the eastern edge of Asia, and trailed behind migrating herds of animals across a land bridge to the Americas created by lowered sea levels (3). The gentler grasslands of the Americas would have allowed the migrating clans to reach the tip of South America in as few as 1,000 years (4). The timing for this migration is under heated debate, ranging from 15-50 thousand years ago, as is the occurrence of later migrations from Asia or Australia. Even the genetic evidence does not give a clear picture, with the genetic distance between Asians and Native Americans producing an estimate of 32,000 years for the time of migration, while the presence of the M242 marker in Siberia 20,000 years ago projects the date farther into the future to about 15,000 years ago, requiring more precise genetic calibration and better archaeological evidence (6).

Genetic evidence shows there were multiple migrations from Asia. The three language families of the Americas; Na-Dene, Amerind, and Eskimo-Aleut, correlate with different genetic compositions. Amerind, the most widespread language family, has high frequencies of M242 and M3 (a marker found only in Native Americans) in both North America and South America. Its large range and homology with Siberian M242 lineages suggest that it is the oldest lineage, and most likely to be the lineage that reached America via the Bering Strait. The second-most widespread language family is Na-Dene, which contains a high frequency of the coastal marker M130, but is not present in South America.

It is made up of humans from coastal China (originating in Siberia) who traveled by boat to North America within the last 10,000 years, after the Bering Strait became submerged. They would have followed the coast along the Pacific Rim, reaching as far south as California. The Eskimo-Aleut speakers do not have a distinct genetic composition, and are most likely a subset of the Amerind clan that settled along the northern coast (4).

The Big Picture

Reconstructing the prehistoric past is always fraught with uncertainties, but genetic tracing has allowed scientists to fill in the sparse fossil record with a broad representation of human migrations, and supplied new insights into how humans evolved. It is a powerful tool, but not flawless – the lineage of a specific gene, marker, or chromosome may not reflect the lineage of an entire species, and the evolutionary paths of different genes may contradict. However, between the Y-chromosome, mtDNA, and nuclear DNA, a clearer picture of how humans came to spread across the globe has emerged. **H**

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