



**Prof. Paweł Golik,
PhD, DSc**

is a geneticist and evolutionary biologist, a Professor at the Faculty of Biology, University of Warsaw. Previously, he worked at the Center for Molecular Medicine at Emory University in Georgia, USA, and at the Centre de Génétique Moléculaire CNRS in Gif-sur-Yvette, France. His research focuses on the mechanisms of gene function, as well as projects at the intersection of genetics and evolutionary theory. In collaboration with the Faculty of "Artes Liberales" at the University of Warsaw, he also participates in projects that combine biology with the humanities and arts. He is an avid popularizer of science and serves as the Chairman of the Council for the Promotion of Science, Polish Academy of Sciences.
p.golik@uw.edu.pl

MIGRATIONS RECORDED IN OUR DNA

Ancient humans led a nomadic lifestyle, moving about in search of food and gradually populating different continents.



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A gorilla – our close relative from the hominid family

Paweł Golik

Faculty of Biology, University of Warsaw
PAS Institute of Biochemistry and Biophysics,
Warsaw

Since ancient times, humans have been very curious about our own origins. To understand who we are today, we need to know where we came from and how our ancestors lived. In the mid-nineteenth century, Charles Darwin provided a broad answer to these fundamental questions: our species, like all others, evolved as part of the tree of life on Earth through the process of biological evolution. Paleontological research, which continues to this day, has since then given us an ever-clearer picture of how our long evolutionary journey unfolded. About seven million years ago, our evolutionary line split off from that of our closest living relatives: chimpanzees. Today, the only surviving branch of that lineage is our species, which we proudly (and perhaps somewhat optimistically) call *Homo sapiens*, or “wise man.”

Historians study the most recent chapters of our story. What came before, or *prehistory* (typically understood as the time between the emergence of the first *Homo* species and the invention of writing), has long intrigued us, while stubbornly holding on to many of its secrets.

Until not long ago, our understanding of early human prehistory came solely from studying fossil remains and artifacts left behind, like tools, tombs, and later, works of art. These findings revealed that the hominin evolutionary story began millions of years ago in Africa. The first species of the *Homo* genus, such as *Homo habilis* – or “handy man” – emerged around 2.5 million years ago. In contrast, our own species appeared much more recently, around 300,000 years ago, which is quite recent in evolutionary terms.

Roughly two million years ago (well before *Homo sapiens* appeared), earlier members of our genus, like *Homo habilis* and later *Homo erectus*, started venturing out of Africa. They reached regions in Asia, such as the Caucasus, modern-day China, and Indonesia, as well as parts of Europe. From around 400,000 years ago, parts of Europe and western Asia, stretching to the Altai Mountains, were home to the Neanderthals – a species that resembled us quite closely but disappeared around 40,000 years ago, soon after modern humans arrived in the same areas. Fossil evidence alone, however, does not provide a clear answer as to how these early human wanderers were related to



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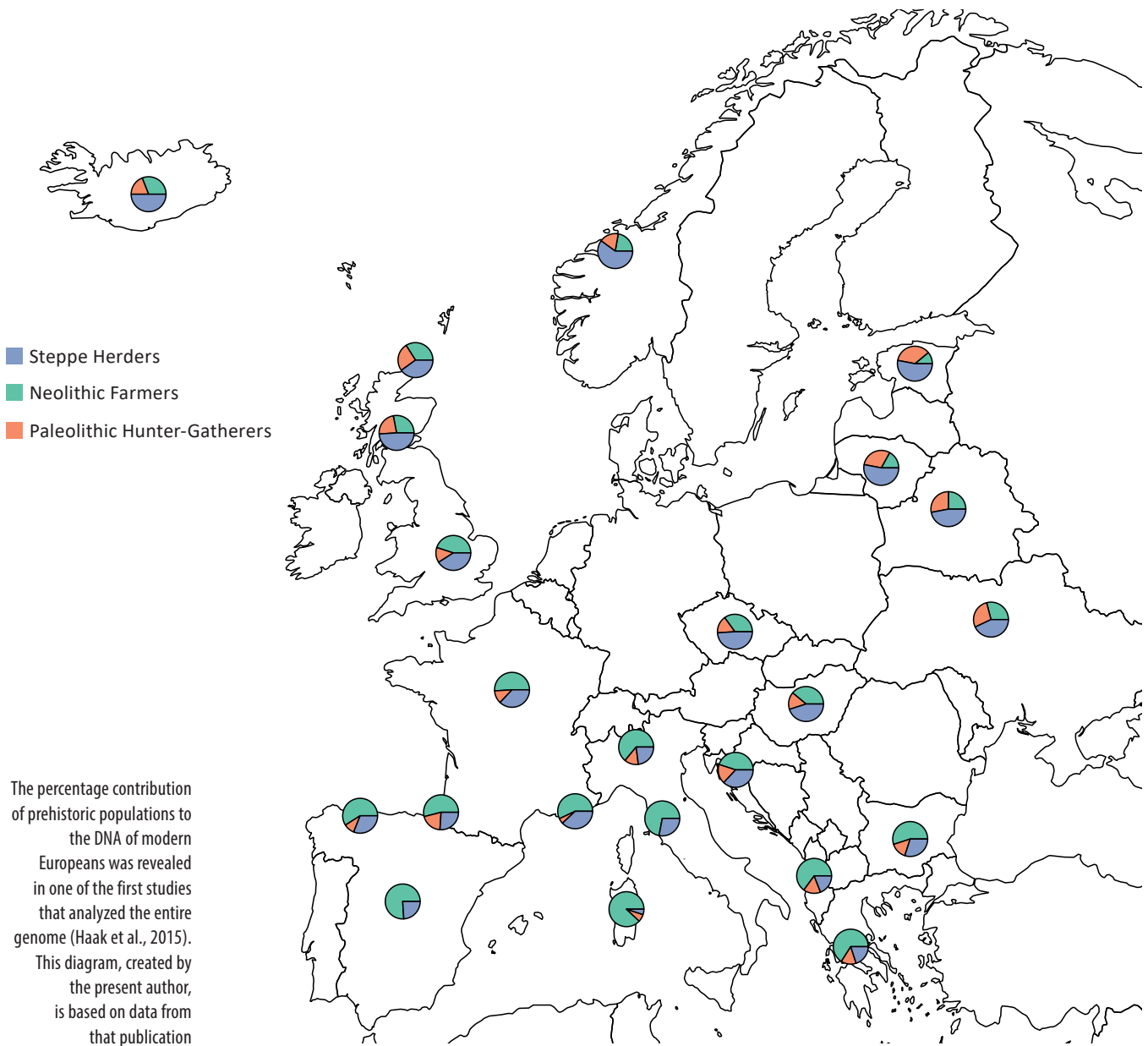
us. Were they our direct ancestors, gradually evolving into modern humans across a vast expanse that included areas far beyond Africa?

The Most Recent Migration

New approaches to answering questions about our prehistory – beyond just studying fossil remains – have emerged only in recent decades. In the late 1970s and early 1980s, geneticists developed gene sequencing methods, which involves reading off the order of nucleotides (the “letters”) in DNA chains. This was a groundbreaking development, not only for understanding how cells and organisms function but also for studying evolution. Essentially, evolution involves changes in DNA sequences that are passed down from one generation to the next, following certain principles that can even be modeled mathematically. By comparing the DNA of living organisms – be they from the same species or from distant branches of the tree of life – scientists can retrace their evolutionary relationships. Interestingly, this idea is not unique to biology. In the nineteenth century, linguists used a similar method to trace out the history of modern languages and even to reconstruct the sounds of long-forgotten ones. In some ways, the process in biology is simpler, as the rules governing DNA evolution tend to be more systematic and consistent.

By the late 1980s, the first studies comparing the DNA of people from different parts of the world were published. These pioneering efforts aimed to reconstruct the prehistory of our species using one specific part of the genome – the *mitochondrial DNA* (or mtDNA). Unlike the chromosomes in the cell nucleus, which are inherited from both parents, mtDNA gets passed down solely from the mother. This simplifies

A human finger bone from Denisova Cave, Russia. The genome of these ancient relatives of both modern humans and Neanderthals – so far found only in such fragmentary skeletal remains – was successfully sequenced in 2012



evolutionary analysis because it avoids the genetic shuffling that occurs when chromosomes are recombined from both parents. Instead, mtDNA allows us to track changes along an unbroken maternal line.

The results of these first mtDNA studies confirmed what fossil research had already suggested: the evolution of our species began in Africa. The root of the human mtDNA tree is situated there, with all non-African mtDNA representing just a branch of one of the three main African lineages. But the next discovery was unexpected. By estimating the rate at which changes accumulate in mtDNA, researchers calculated that the ancestors of all modern humans lived in Africa around 200,000 years ago, forming a relatively small population. These ancestors were already members of our own species, *H. sapiens*. Ear-

lier hominins like *H. erectus* and Neanderthals, who had left Africa much earlier and spread across Eurasia, were therefore not our direct ancestors. Instead, they were earlier offshoots of the evolutionary tree. After modern humans themselves left Africa in a later migration, they replaced and eventually outcompeted these earlier species. We Europeans are descendants not of the earliest waves of *H. sapiens* migration, but rather of the most recent one, which began less than 100,000 years ago.

Disappearing DNA

This theory gained additional support at the end of the twentieth century. In addition to mtDNA, researchers began to study another part of the human genome



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these genetic stories disappear. New tools to solve this issue have only recently become available.

Australopithecus afarensis

A wealth of data

The early twenty-first century brought significant advancements in DNA sequencing, particularly with the introduction of *next-generation sequencing* (NGS). These new methods allowed scientists to make rapid progress, moving from the monumental task of decoding the first human genome (completed in 2000–2001) to sequencing 1,000 complete genomes from people around the world (by 2015). By 2022, there were databases containing the full DNA sequences of hundreds of thousands of people established in the UK and the US, and researchers had sequenced the genomes of

inherited directly from one parent – the Y chromosome. Passed from father to son, Y chromosomes also turned out to trace back to Africa about 200,000 years ago. Meanwhile, advances in DNA isolation, amplification, and sequencing allowed scientists to analyze ancient DNA (aDNA) from fossil remains. Early studies of aDNA in the late 1990s placed European Neanderthals on a branch of the evolutionary tree running parallel to that of modern humans, and further from modern Europeans than African branches. This seemed to further corroborate the idea that Neanderthals were not direct ancestors of *Homo sapiens*, but a side branch that was ultimately replaced.

By the turn of the twenty-first century, thanks to increasingly detailed studies of both mtDNA and Y chromosomes, scientists were able to map out the major migration routes of early *Homo sapiens*. These maps revealed that, around 50,000 years ago, early humans reached what is now Australia by moving along the coasts of Asia. A group known as the basal Eurasian population migrated west to Europe (arriving about 45,000 years ago) and east to Asia. From there, some crossed the Bering Strait – then a land bridge – into the Americas, some 15,000–20,000 years ago. Private companies even began offering genetic tests based on mtDNA and Y chromosome analysis, promising to trace customers' ancestry back to these early human lineages.

However, there was a major limitation to these early studies. The focus on mtDNA and the Y chromosome – genetic material inherited only from one parent – meant that much of the genetic story was left out. Think of a modern family: children do inherit mtDNA from their mother, passed down through the maternal line, and the Y chromosome (if male) from their father, passed through the paternal line. But this leaves out the maternal grandfather and paternal grandmother, whose genes do continue into the next generation, albeit not via the mtDNA or Y chromosome. The further back in time we go, the more of

Whole-genome research, particularly with ancient DNA, has also led to surprising discoveries about our relationship with Neanderthals.

over 1,000 individuals from Poland. This explosive growth in human genomics has not only revolutionized biomedical research but has also transformed our understanding of human prehistory and evolution.

The past two decades have also seen remarkable progress in ancient DNA research. By combining next-generation sequencing, highly refined DNA iso-

Reconstruction of *Homo habilis*, Rama 9 Museum, Bangkok



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lation techniques, and powerful new methods of computational analysis, scientists can now study genomes from prehistoric times. For example, well-preserved remains in Arctic permafrost have allowed researchers to sequence the entire genome of mammoths that lived over a million years ago. However, because humans did not inhabit the far north that long ago, whereas Africa's climate is not conducive to preserving DNA, much of what we know about early human prehistory comes from remains found in Eurasia. Between 2006 and 2010, a team led by Svante Pääbo in Leipzig reconstructed nearly the complete genome of Neanderthals, dating back tens of thousands of years – about as old as the oldest complete DNA from our own species, which is about 45,000 years old.

The abundance of modern and ancient whole-genome sequences has now given us an unprecedented view of our prehistory, far beyond what was possible with studies limited to mtDNA and the Y chromo-

some. Alongside DNA sequencing, the development of advanced mathematical tools for analyzing genetic similarities and differences has played a key role. These tools account for the fact that genetic material from both parents is shuffled before being passed down to the next generation. The impact of these new techniques on the study of human prehistory is perhaps best demonstrated by the deep insights they have provided into the ancient history of Europe.

The colonization of Europe

Humans first arrived in Europe about 45,000 years ago; there they encountered Neanderthals who had been living outside Africa for hundreds of thousands of years. These early Europeans were nomadic hunter-gatherers from various Paleolithic cultures. They are known as the *Western Hunter-Gatherers* (WHG) and their story was complex – during the Last Glacial Maximum, they retreated to the warmer southern regions of Europe, only to recolonize the continent once the ice retreated. Another key event in European history was the spread of agriculture, which began arriving from the Near East (the Fertile Crescent) around 9,000 years ago. Next, over 5,000 years ago, at the transition from the Stone Age to the Bronze Age, people from the Pontic Steppe, located between Eastern Europe and Asia, migrated into Europe, bringing with them the Indo-European languages.

Early studies focusing only on mtDNA and Y chromosomes suggested that the vast majority of Europeans descended from this last steppe migration, whereas traces of earlier populations could perhaps be sought only among the Basques. However, this view shifted in 2015, when studies analyzing the complete genomes of hundreds of modern Europeans, along with ancient DNA from Stone and Bronze Age skeletons, revealed a much more intricate story. Rather than one wave replacing another, it turns out that each wave of migration mixed with the people already living there, and today's Europeans carry DNA from all these ancient groups.

For example, when the first Neolithic farmers (known as the *Early European Farmers*, or EEF) arrived in Europe and met the earlier WHG hunter-gatherers, they initially lived side by side but gradually began to intermix over centuries. Similarly, when the steppe herders arrived during the Bronze Age, they did not completely replace the earlier inhabitants – they too blended with them. In fact, the steppe herders were themselves the product of earlier mixtures, combining genes from Eastern European hunter-gatherers, populations from Asia and the Caucasus, as well as influences from the Near East (modern-day Iran) and Siberia. The DNA of modern Europeans still carries traces of all these migrations and the repeated blend-

Skulls of various *Homo* species, including *Homo antecessor*, one of the earliest human ancestors whose traces have been discovered in Europe



Homo neanderthalis



Homo antecessor



Homo sapiens



Homo erectus



Reconstruction of *Homo erectus*, Natural History Museum in Vienna

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ing of populations, a process known as *admixture*. The percentage of genetic material from these ancient groups varies depending on where on the continent someone’s ancestors came from. However, these differences do not form clear boundaries; instead, they form gradients that still reflect the major migrations of the past.

An intertwined history

Recent years have brought new studies that continue to illuminate the prehistory of different parts of the world. While the specifics may differ, the overall pattern is the same: waves of migration led to the mixing of various populations. Until recently, it was thought that the indigenous peoples of southern Africa’s deserts were an example of a “pure” population – genetically unchanged since the dawn of our species, untouched by later migrations. However, new research in recent years, exploring Africa’s vast human diversity for the first time, has shown that even in this region, people carry traces of ancient migrations and the intermingling of different groups.

Whole-genome studies, especially with ancient DNA, have also changed our understanding of our relationship with Neanderthals. While it is still true that our and their evolutionary lines split apart long before *Homo sapiens* left Africa, it turns out that once our ancestors migrated out of Africa, they encountered Neanderthals and interbred with them (cross-

breeding between closely related species is a common phenomenon in zoology). Today, people of Eurasian descent carry the genetic legacy of these encounters—about 2% to 4% of their DNA comes from Neanderthals (in African populations, Neanderthal DNA is much rarer). In Asia, early humans also interbred with another, lesser-known species whose remains were found in the Altai Mountains, known as Denisovans. What is more, Denisovans and Neanderthals also interbred, and traces of this have been found in DNA extracted from skeletons nearly 100,000 years old. Ongoing research aims to determine whether these ancient genetic exchanges are simply evolutionary curiosities or whether they might have an impact on our health today. It is no surprise that Svante Pääbo won the 2022 Nobel Prize in Physiology or Medicine for his groundbreaking work.

The use of genomic analysis to study prehistory – known as *archaeogenetics* – is still a young and fast-growing field. It is also highly interdisciplinary. DNA only tells part of the story; the rest needs to be filled in by archaeological and anthropological research. Undoubtedly, there are many surprises still in store for us. But one thing we know for certain: migration has been part of our species’ story from the very beginning, shaping the very essence of who we are – discoverers and explorers, open to new encounters. We cannot be divided up into isolated groups; our genes carry the intertwined history of people from all corners of the world. ■

Further reading:

Haak W. et al., Massive migration from the steppe was a source for Indo-European languages in Europe, *Nature* 2015, vol. 522.

Pääbo S., *Neanderthal Man: In Search of Lost Genomes*, 2014.

Reich D., *Who We Are and How We Got Here: Ancient DNA and the New Science of the Human Past*, 2018.

Rutherford A., *A Brief History of Everyone Who Ever Lived: The Stories in Our Genes*, 2016.