

When did genetic variations that make us human emerge?

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The study of the genomes of our closest relatives, the Neanderthals and Denisovans, has opened up new research paths that can broaden our understanding of the evolutionary history of *Homo sapiens*. A study led by the UB has made an estimation of the time when some of the genetic variants that characterize our species emerged. It does so by analyzing mutations that are very frequent in modern human populations, but not

in these other species of archaic humans.

The results, published in the journal *Scientific Reports*, show two moments in which mutations accumulated: one around 40,000 years ago, associated with the growth of the *Homo sapiens* population and its departure from Africa, and an older one, more than 100,000 years ago, related to the time of the greatest diversity of types of *Homo sapiens* in Africa.

"The understanding of the deep history of our species is expanding rapidly. However, it is difficult to determine when the genetic variants that distinguish us from other human species emerged. In this study, we have placed species-specific variants on a timeline. We have discovered how these variants accumulate over time, reflecting events such as the point of divergence between *Homo sapiens* and other human species around 100,000 years ago," says Alejandro Andirkó, first author of this article, which was part of his doctoral thesis at the UB.

The study, led by Cedric Boeckx, ICREA research professor in the section of General Linguistics and member of the Institute of Complex Systems of the UB (UBICS), included the participation of Juan Moriano, UB researcher, Alessandro Vitriolo and Giuseppe Testa, experts from the University of Milan and the European Institute of Oncology, and Martin Kuhlwilm, researcher at the University of Vienna.

Predominance of behavioral and facial-related variations

The results of the research study also show differences between evolutionary periods. Specifically, they highlight the predominance of genetic variants related to behavior and facial structure—key characteristics in the differentiation of our species from other human

species—more than 300,000 years ago, a date that coincides with the available fossil and [archaeological evidence](#). "We have discovered sets of genetic variants which affect the evolution of the face and which we have dated between 300,000 and 500,000 years ago, the period just prior to the dating of the earliest fossils of our species, such as the ones discovered at the Jebel Irhoud archaeological site in Morocco," notes Andirkó.

The researchers also analyzed variants related to the brain, the organ that can best help explain key features of the rich repertoire of behaviors associated with *Homo sapiens*. Specifically, they dated variants which [medical studies](#) conducted in present-day humans have linked to the volume of the cerebellum, corpus callosum and other structures. "We found that brain tissues have a particular genomic expression profile at different times in our history; that is, certain genes related to neural development were more highly expressed at certain times," says the researcher.

Supporting the mosaic nature of the evolution of *Homo sapiens*

These results complement an idea that is dominant in evolutionary anthropology: that there is no linear history of [human species](#), but that different branches of our evolutionary tree coexisted and often intersected. "The breadth of the range of human diversity in the past has surprised anthropologists. Even within *Homo sapiens* there are fossils, such as the ones I mentioned earlier from Jebel Irhoud, which, because of their features, were thought to belong to another species. That's why we say that human beings have lived a mosaic evolution," he notes.

"Our results," the researcher continues, "offer a picture of how our genetics changed, which fits this idea, as we found no evidence of

evolutionary changes that depended on one or a several key mutations," he says.

Application of machine learning techniques

The methodology used in the study was based on a Genealogical Estimation of Variant Age method, developed by researchers at the University of Oxford. Once they had this estimation, they applied a [machine learning tool](#) to predict which genes have changed the most in certain time windows and which tissues these genes may have impacted. Specifically, they used ExPecto, a deep learning tool that uses a convolutional network—a type of computational model—to predict gene expression levels and function from a DNA sequence.

"Since there are no data on the genomic expression of variants in the past, this tool is an approach to a problem that has not been addressed until now. Although the use of machine learning prediction is increasingly common in the clinical world, as far as we know, nobody has tried to predict the consequences of genomic changes over time," notes Andirkó.

The importance of the perinatal phase in the brain development of our species

In a previous study, the same UB team, together with the researcher Raül Gómez Buisán, used genomic information from archaic humans. In that study they analyzed genomic deserts, regions of the genome of our species where there are no genetic fragments of Neanderthals or Denisovans, and which, moreover, have been subjected to positive pressure in our species: that is, they have accumulated more mutations than would have been expected by neutral evolution. The researchers studied the expression of genes—i.e., which proteins code for different

functions—found in desert regions throughout [brain development](#), from prenatal to adult stages, covering sixteen brain structures. The results showed differences in gene expression in the cerebellum, striatum and thalamus. "These results bring into focus the relevance of brain structures beyond the neocortex, which has traditionally dominated research on the evolution of the human brain," says Juan Moriano.

Moreover, the most striking differences between brain structures were found at prenatal stages. "These findings add new evidence to the hypothesis of a [species](#)-specific trajectory of brain development taking place at perinatal stages—the period from 22 weeks to the end of the first four weeks of neonatal life—that would result in a more globular head shape in modern humans, in contrast to the more elongated shape seen in Neanderthals," concludes Moriano.

More information: Alejandro Andirkó et al, Temporal mapping of derived high-frequency gene variants supports the mosaic nature of the evolution of Homo sapiens, *Scientific Reports* (2022). [DOI: 10.1038/s41598-022-13589-0](#)

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