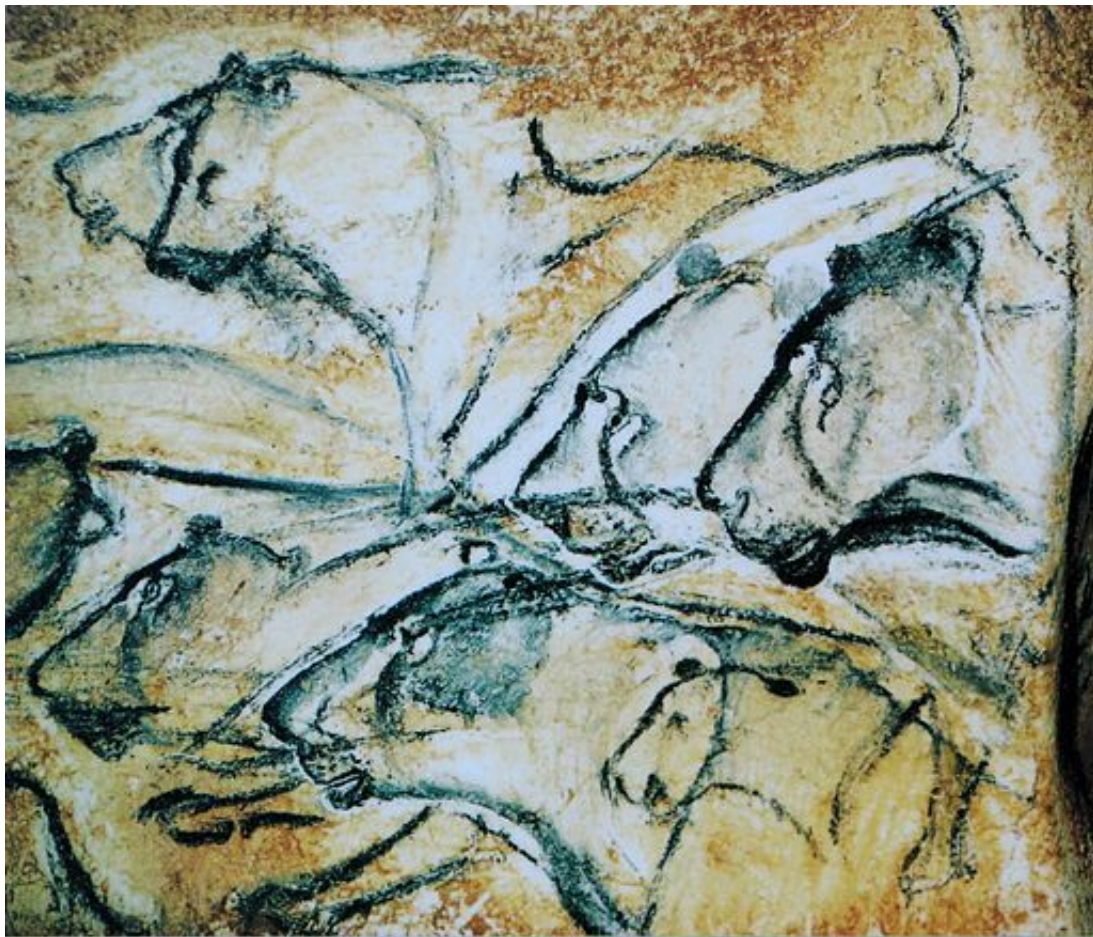


## Tracing Origins

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## Being Human

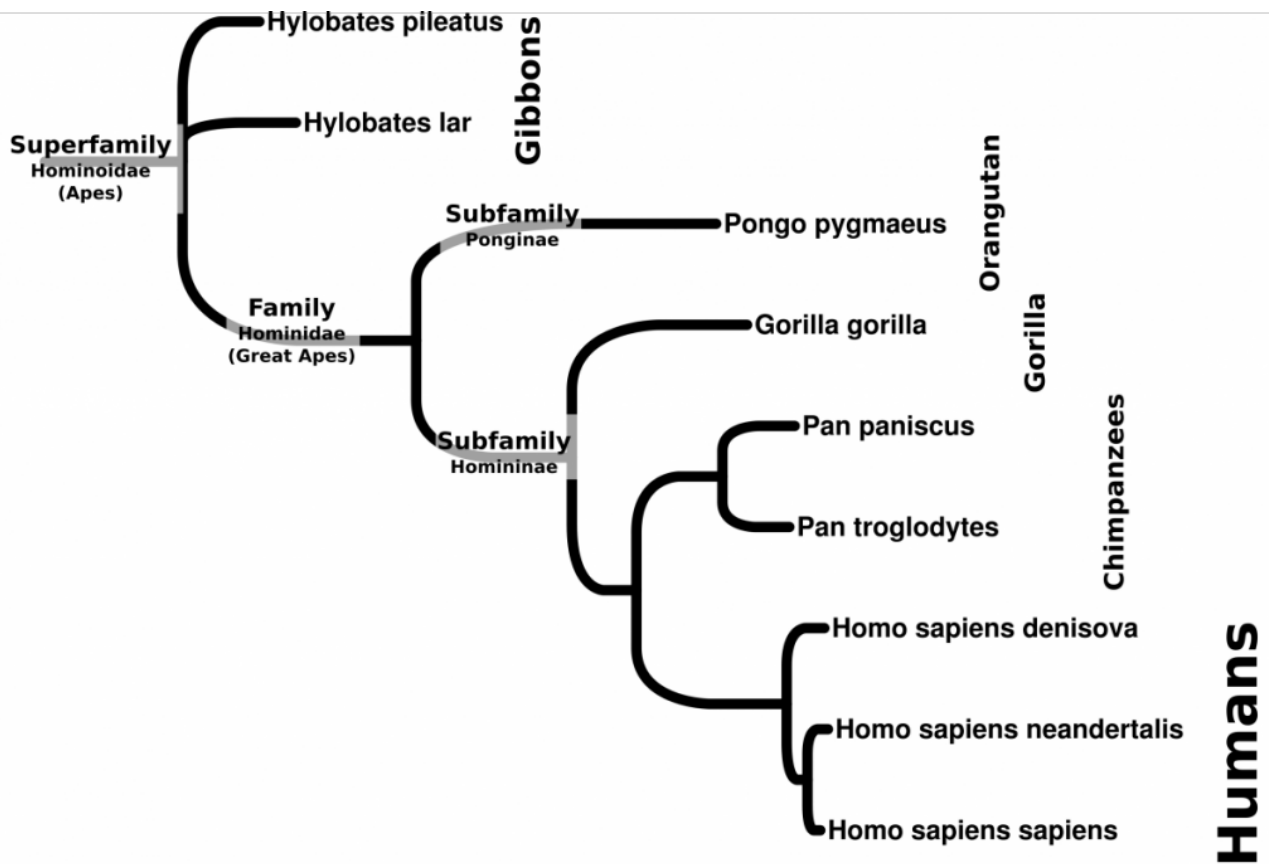


Drawings dating to approximately 30,000 years ago in the Chauvet Cave. What constitutes being human? Many will point at a cultural identity and leaving long-standing remnants of that culture. Such prehistorical artifacts like cave drawings and tools provide an anthropological framework for identifying what it is to be human, but the biological identity remains locked in the history of our DNA.



Spear points of the Clovis Culture in the Americas dating to approximately 13,000 years ago.

## The Great Apes



Phylogenetic tree generated with Cytochrome Oxidase I (COI) genes.

*Homo sapiens* represent a branch of primates in the line of Great Apes. The family of Great Apes consists of four extant genera: *Homo*, *Pan*, *Gorilla*, *Pongo*. Karyotype analysis ([Yunis et al., 1982](#)) reveals a shared genomic structure between the Great Apes. While humans have 46



chromosomes, the other Great Apes have 48. Molecular evidence at the DNA level indicates that Human Chromosome 2 is a fusion of 2 individual chromosomes. In the other Great Apes, these 2 Chromosomes are referred to as 2p and 2q to illustrate their synteny to the human counterpart.



The Pan-Homo divergence. A display at the Cradle of Humankind illuminates the skulls of two extant Hominini with a series of model fossils from the Hominina subtribe of Australopithecina and Homo

Chimpanzees (*Pan*) are the closest living relatives to modern humans. It is commonly cited that less than 2% differences in their nucleotide sequences exist with humans ([Chimpanzee Sequencing and Analysis Consortium, 2005](#)). More recent findings in comparing the complement of genes (including duplication and gene loss events) now describes the difference in genomes at about 6% ([Demuth JP, et al., 2006](#)).

## The Genus *Homo*





An underground lake at inside the Sterkfontein Cave system at the Cradle of Humankind (South Africa)

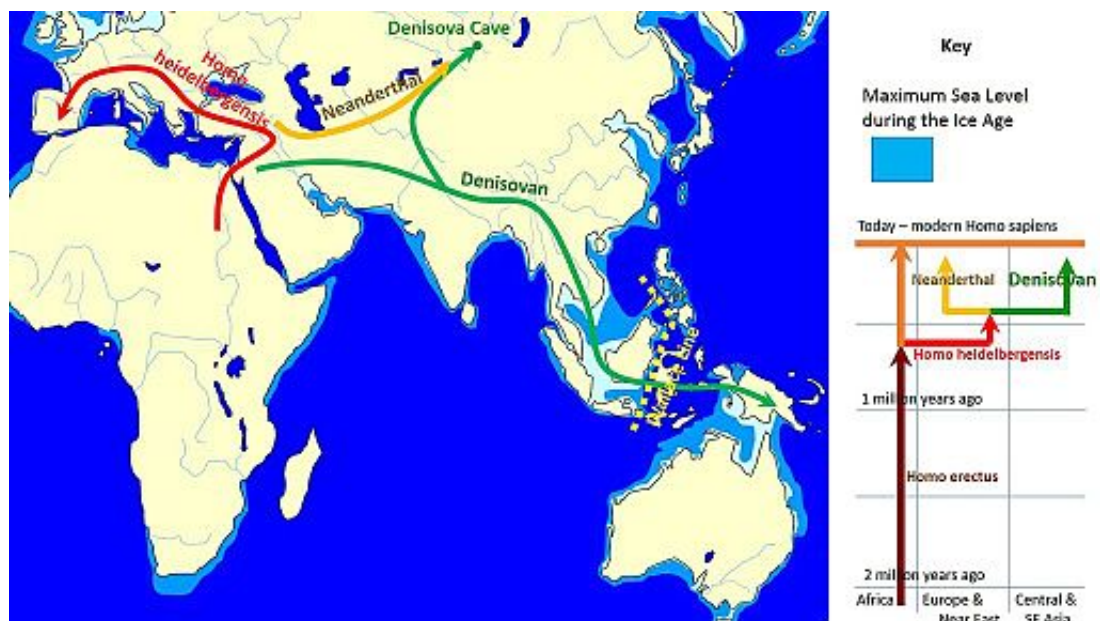
The rise of the human lineage is thought to arise in Africa. Fossils of *Australopithecus* (southern apes) found in death traps, like those at the Cradle of Humankind, reveal a historical record of organisms inhabiting the landscape. The breaks in the ceiling of the caves provide opportunities for animals to fall inside these caves to their death. The limestone deposits of the caves serve as an environment for fossilization and mineralization of their remains. An abundance of fossilized hominids in these caves including *Australopithecus africanus*, *Australopithecus prometheus*, *Paranthropus boisei*, and the newly discovered *Homo naledi* continue to reveal the natural history of the genus *Homo* from 2.6 million to 200,000 years ago.





The entrance to the archaeological site at Sterkfontein, Cradle of Humankind (South Africa).

## Ancient DNA of Humans



In 2008, a piece of a finger bone and a molar from a Siberian Cave were found that differed slightly from that of modern humans. The cave, called Denisova Cave, maintains an average temperature of 0°C year round and was suspected to contain viable soft tissue. Bones in this cave were discovered that had similarities to modern humans and Neandertals. An initial mitochondrial DNA analysis revealed that these beings represented a distinct line of humans that overlapped with them in time ([Krause et al., 2010](#)). Analysis of the full nuclear genome followed and indicated that interbreeding existed between these Denisovans, Neandertals and modern humans ([Reich et al., 2010](#)). Furthermore, analysis of DNA from a 400,000 year old femur in Spain revealed that these three lines diverged from the species *Homo heidelbergensis* and that Denisovans were closest in sequence ([Meyer et al., 2016](#)).

Between modern humans, markers found in the mtDNA can be used to trace the migrations and origins along the maternal line. Similarly, VNTRs found on the Y chromosome have revealed migration patterns along paternal lines within men. Other markers, like the insertion points of transposable elements can be used to further describe the genetics and inheritance of modern humans while providing a snapshot into evolutionary history.

## Other Resources

- [Great Ape Mitochondrial Sequences](#)
- <http://media.hhmi.org/biointeractive/click/Origins/01.html>
- [Yunis JJ, Prakash O](#). The origin of man: a chromosomal pictorial legacy. [Science](#). 1982 Mar 19;215(4539):1525-30.
- Chimpanzee Sequencing and Analysis Consortium. Initial sequence of the chimpanzee genome and comparison with the human genome. [Nature](#). 2005 Sep 1;437(7055):69-87.
- [Demuth JP, De Bie T, Stajich JE, Cristianini N, Hahn MW](#). [The evolution of mammalian gene families](#). [PLoS One](#). 2006 Dec 20;1:e85.
- [Krause, Johannes; Fu, Qiaomei; Good, Jeffrey M.; Viola, Bence; Shunkov, Michael V.; Derevianko, Anatoli P. & Pääbo, Svante \(2010\)](#), “The complete mitochondrial DNA genome of an unknown hominin from southern Siberia”, [Nature](#), **464** (7290): 894–897, doi:[10.1038/nature08976](https://doi.org/10.1038/nature08976), PMID [20336068](https://pubmed.ncbi.nlm.nih.gov/20336068/)
- [Reich, David; Green, Richard E.; Kircher, Martin; Krause, Johannes; Patterson, Nick; Durand, Eric Y.; Viola, Bence; Briggs, Adrian W. & Stenzel, Udo \(2010\)](#), “Genetic history of an archaic hominin group from Denisova Cave in Siberia”, [Nature](#), **468** (7327): 1053–1060, doi:[10.1038/nature09710](https://doi.org/10.1038/nature09710), PMID [21179161](https://pubmed.ncbi.nlm.nih.gov/21179161/)
- [Meyer M, Arsuaga JL, de Filippo C, Nagel S, Aximu-Petri A, Nickel B, Martínez I, Gracia A, Bermúdez de Castro JM, Carbonell E, Viola B, Kelso J, Prüfer K, Pääbo S](#). [Nuclear DNA sequences from the Middle Pleistocene Sima de los Huesos hominins](#). [Nature](#). 2016 Mar 24;531(7595):504-7. doi: [10.1038/nature17405](https://doi.org/10.1038/nature17405). PMID:[26976447](https://pubmed.ncbi.nlm.nih.gov/26976447/)

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