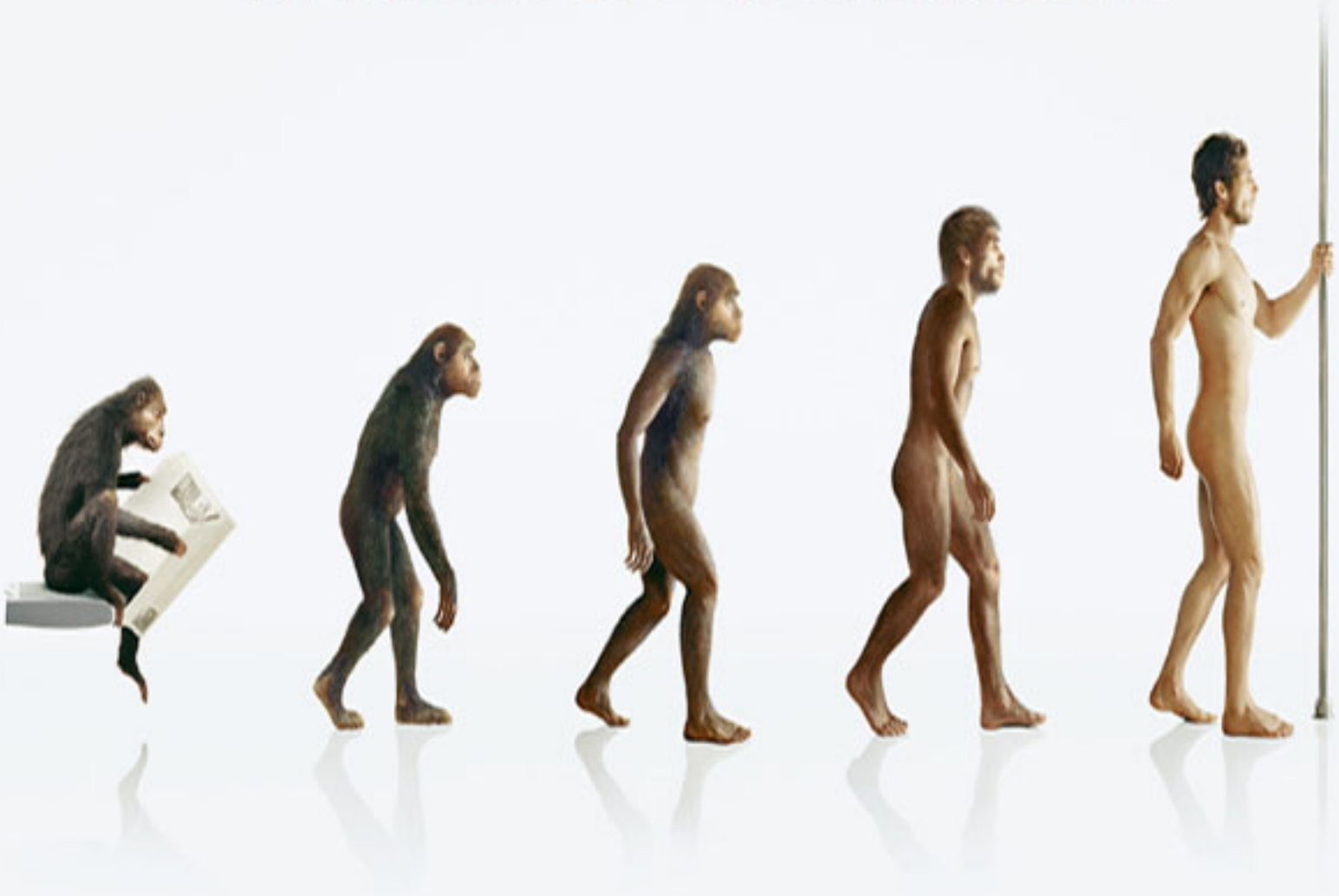
An Introduction to Human Evolution



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First Edition, 2012

ISBN 978-81-323-4608-1

Published by:
The English Press
4735/22 Prakashdeep Bldg,
Ansari Road, Darya Ganj,
Delhi - 110002
Email: info@wtbooks.com

Table of Contents

Chapter- 1

Introduction to Human Evolution

Human evolution, or *anthropogeny*, is the origin and evolution of *Homo sapiens* as a distinct species from other hominids, great apes and placental mammals. The study of human evolution uses many scientific disciplines, including physical anthropology, primatology, archaeology, linguistics and genetics.

The term "human" in the context of human evolution refers to the genus *Homo*, but studies of human evolution usually include other hominids, such as the Australopithecines, from which the genus *Homo* had diverged by about 2.3 to 2.4 million years ago in Africa. Scientists have estimated that humans branched off from their common ancestor with chimpanzees about 5–7 million years ago. Several species and subspecies of *Homo* evolved and are now extinct. These include *Homo erectus*, which inhabited Asia and *Homo sapiens neanderthalensis*, which inhabited Europe. Archaic *Homo sapiens* evolved between 400,000 and 250,000 years ago.

The dominant view among scientists concerning the origin of anatomically modern humans is the "Out of Africa" or recent African origin hypothesis, which argues that *Homo sapiens* arose in Africa and migrated out of the continent around 50,000 to 100,000 years ago, replacing populations of *Homo erectus* in Asia and *Homo neanderthalensis* in Europe. Scientists supporting the alternative multiregional hypothesis argue that *Homo sapiens* evolved as geographically separate but interbreeding populations stemming from a worldwide migration of *Homo erectus* out of Africa nearly 2.5 million years ago. This theory has been contradicted by recent evidence, although it has been suggested that non Homo sapiens Neanderthal genomes may have contributed about 4% of non African heredity and the recently discovered Denisova hominin may have contributed 6% of the genome of Melanesians.

History of ideas

The word *homo*, the name of the biological genus to which humans belong, is Latin for "human". It was chosen originally by Carolus Linnaeus in his classification system. The word "human" is from the Latin *humanus*, the adjectival form of *homo*. The Latin "homo" derives from the Indo-European root, *dhghem*, or "earth".

Carolus Linnaeus and other scientists of his time also considered the great apes to be the closest relatives of humans due to morphological and anatomical similarities. The

possibility of linking humans with earlier apes by descent only became clear after 1859 with the publication of Charles Darwin's *On the Origin of Species*. This argued for the idea of the evolution of new species from earlier ones. Darwin's book did not address the question of human evolution, saying only that "Light will be thrown on the origin of man and his history".

Fossil Hominid Evolution Display at The Museum of Osteology, Oklahoma City, USA.

The first debates about the nature of human evolution arose between Thomas Huxley and Richard Owen. Huxley argued for human evolution from apes by illustrating many of the similarities and differences between humans and apes and did so particularly in his 1863 book *Evidence as to Man's Place in Nature*. However, many of Darwin's early supporters (such as Alfred Russel Wallace and Charles Lyell) did not agree that the origin of the mental capacities and the moral sensibilities of humans could be explained by natural

selection. Darwin applied the theory of evolution and sexual selection to humans when he published *The Descent of Man* in 1871.

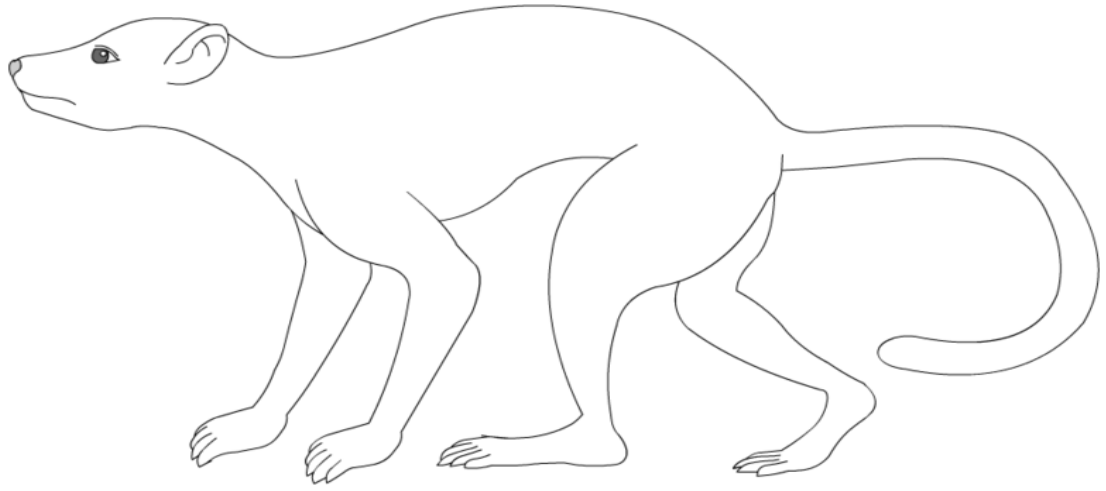
A major problem was the lack of fossil intermediaries. It was only in the 1920s that such fossils were discovered in Africa. In 1925, Raymond Dart described *Australopithecus africanus*. The type specimen was the Taung Child, an Australopithecine infant discovered in a cave. The child's remains were a remarkably well-preserved tiny skull and an endocranial cast of the individual's brain. Although the brain was small (410 cm^3), its shape was rounded, unlike that of chimpanzees and gorillas and more like a modern human brain. Also, the specimen showed short canine teeth and the position of the foramen magnum was evidence of bipedal locomotion. All of these traits convinced Dart that the Taung baby was a bipedal human ancestor, a transitional form between apes and humans.

The classification of humans and their relatives has changed considerably over time. The gracile Australopithecines are now thought to be ancestors of the genus *Homo*, the group to which modern humans belong. Both Australopithecines and *Homo sapiens* are part of the tribe Hominini. Recent data suggests Australopithecines were a diverse group and that *A. africanus* may not be a direct ancestor of modern humans. Reclassification of Australopithecines that originally were split into either gracile or robust varieties has put the latter into a family of its own, *Paranthropus*. Taxonomists place humans, Australopithecines and related species in the same family as other great apes, in the Hominidae.

Note: 1e+06 years = 1 × 10^6 years = 1 million years ago = 1 Ma

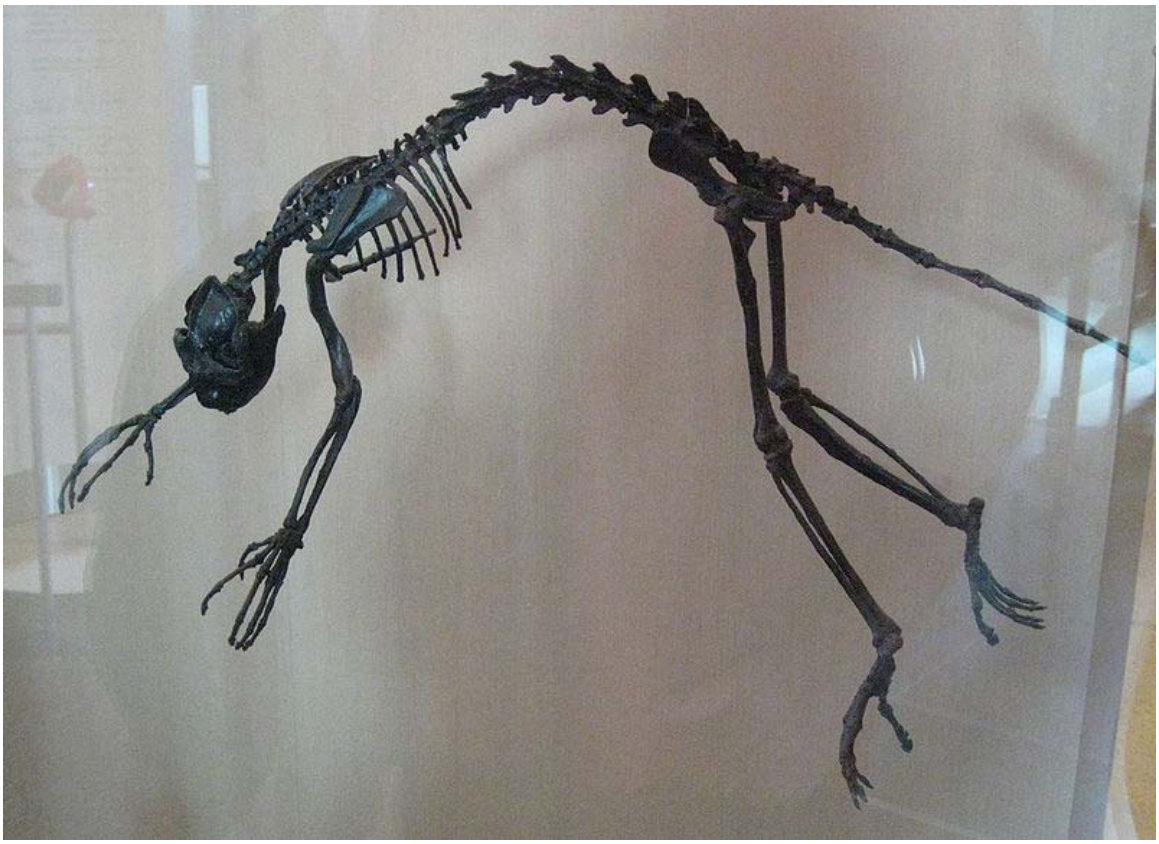
Before Homo

Evolution of the great apes



Plesiadapis

The evolutionary history of the primates can be traced back 65 million years, as one of the oldest of all surviving placental mammal groups. The oldest known primate-like mammal species, the Plesiadapis, come from North America, but they were widespread in Eurasia and Africa during the tropical conditions of the Paleocene and Eocene.



Notharctus

The beginning of modern climates was marked by the formation of the first Antarctic ice in the early Oligocene around 30 million years ago. A primate from this time was *Notharctus*. Fossil evidence found in Germany in the 1980s was determined to be about 16.5 million years old, some 1.5 million years older than similar species from East Africa and challenging the original theory regarding human ancestry originating on the African continent.

David Begun says that these primates flourished in Eurasia and that the lineage leading to the African apes and humans— including Dryopithecus—migrated south from Europe or Western Asia into Africa. The surviving tropical population, which is seen most completely in the upper Eocene and lowermost Oligocene fossil beds of the Fayum depression southwest of Cairo, gave rise to all living primates—lemurs of Madagascar, lorises of Southeast Asia, galagos or "bush babies" of Africa and the anthropoids; platyrrhines or New World monkeys and catarrhines or Old World monkeys and the great apes and humans.

The earliest known catarrhine is *Kamoyapithecus* from uppermost Oligocene at Eragaleit in the northern Kenya Rift Valley, dated to 24 million years ago. Its ancestry is generally thought to be species related to *Aegyptopithecus*, *Propliopithecus* and *Parapithecus* from the Fayum, at around 35 million years ago. In 2010, *Saadanius* was described as a close relative of the last common ancestor of the crown catarrhines and tentatively dated to 29–28 million years ago, helping to fill an 11-million-year gap in the fossil record.

Reconstructed tailless *Proconsul* skeleton

In the early Miocene, about 22 million years ago, the many kinds of arboreally adapted primitive catarrhines from East Africa suggest a long history of prior diversification. Fossils at 20 million years ago include fragments attributed to *Victoriapithecus*, the earliest Old World Monkey. Among the genera thought to be in the ape lineage leading up to 13 million years ago are *Proconsul*, *Rangwapithecus*, *Dendropithecus*, *Limnopithecus*, *Nacholapithecus*, *Equatorius*, *Nyanzapithecus*, *Afropithecus*, *Heliopithecus* and *Kenyapithecus*, all from East Africa. The presence of other generalized non-cercopithecids of middle Miocene age from sites far distant—*Otavipithecus* from cave deposits in Namibia and *Pierolapithecus* and *Dryopithecus* from France, Spain and Austria—is evidence of a wide diversity of forms across Africa and the Mediterranean basin during the relatively warm and equable climatic regimes of the early and middle Miocene. The youngest of the Miocene hominoids, *Oreopithecus*, is from 9 million year old coal beds in Italy.

Molecular evidence indicates that the lineage of gibbons (family Hylobatidae) became distinct from Great Apes between 18 and 12 million years ago and that of orangutans (subfamily Ponginae) became distinct from the other Great Apes at about 12 million years; there are no fossils that clearly document the ancestry of gibbons, which may have originated in a so-far-unknown South East Asian hominoid population, but fossil proto-orangutans may be represented by *Ramapithecus* from India and *Griphopithecus* from Turkey, dated to around 10 million years ago.

Divergence of the human lineage from other Great Apes

Species close to the last common ancestor of gorillas, chimpanzees and humans may be represented by *Nakalipithecus* fossils found in Kenya and *Ouranopithecus* found in Greece. Molecular evidence suggests that between 8 and 4 million years ago, first the gorillas and then the chimpanzees (genus *Pan*) split off from the line leading to the humans; human DNA is approximately 98.4% identical to that of chimpanzees when comparing single nucleotide polymorphisms. The fossil record of gorillas and chimpanzees is quite limited. Both poor preservation (rain forest soils tend to be acidic and dissolve bone) and sampling bias probably contribute to this problem.

Other hominines likely adapted to the drier environments outside the equatorial belt, along with antelopes, hyenas, dogs, pigs, elephants and horses. The equatorial belt contracted after about 8 million years ago. Fossils of these hominans - the species in the human lineage following divergence from the chimpanzees - are relatively well known.

The earliest are *Sahelanthropus tchadensis* (7 Ma) and *Orrorin tugenensis* (6 Ma), followed by:

- *Ardipithecus* (5.5–4.4 Ma), with species *Ar. kadabba* and *Ar. ramidus*;
- *Australopithecus* (4–1.8 Ma), with species *Au. anamensis*, *Au. afarensis*, *Au. africanus*, *Au. bahrelghazali*, *Au. garhi* and *Au. sediba*;
- *Kenyanthropus* (3–2.7 Ma), with species *Kenyanthropus platyops*;
- *Paranthropus* (3–1.2 Ma), with species *P. aethiopicus*, *P. boisei* and *P. robustus*;
- *Homo* (2 Ma–present), with species *Homo habilis*, *Homo rudolfensis*, *Homo ergaster*, *Homo georgicus*, *Homo antecessor*, *Homo cepranensis*, *Homo erectus*, *Homo heidelbergensis*, *Homo rhodesiensis*, *Homo neanderthalensis*, *Homo sapiens idaltu*, *Archaic Homo sapiens*, *Homo floresiensis*.

Genus Homo

Homo sapiens is the only extant species of its genus, *Homo*. While some other, extinct, *Homo* species might have been ancestors of *Homo sapiens*, many were likely our "cousins", having speciated away from our ancestral line. There is not yet a consensus as to which of these groups should count as separate species and which as subspecies. In some cases this is due to the dearth of fossils, in other cases it is due to the slight differences used to classify species in the *Homo* genus. The Sahara pump theory

(describing an occasionally passable "wet" Sahara Desert) provides an explanation of the early variation in the genus *Homo*.

Based on archaeological and paleontological evidence, it has been possible to infer, to some extent, the ancient dietary practices of various *Homo* species and to study the role of diet in physical and behavioral evolution within *Homo*.

H. habilis

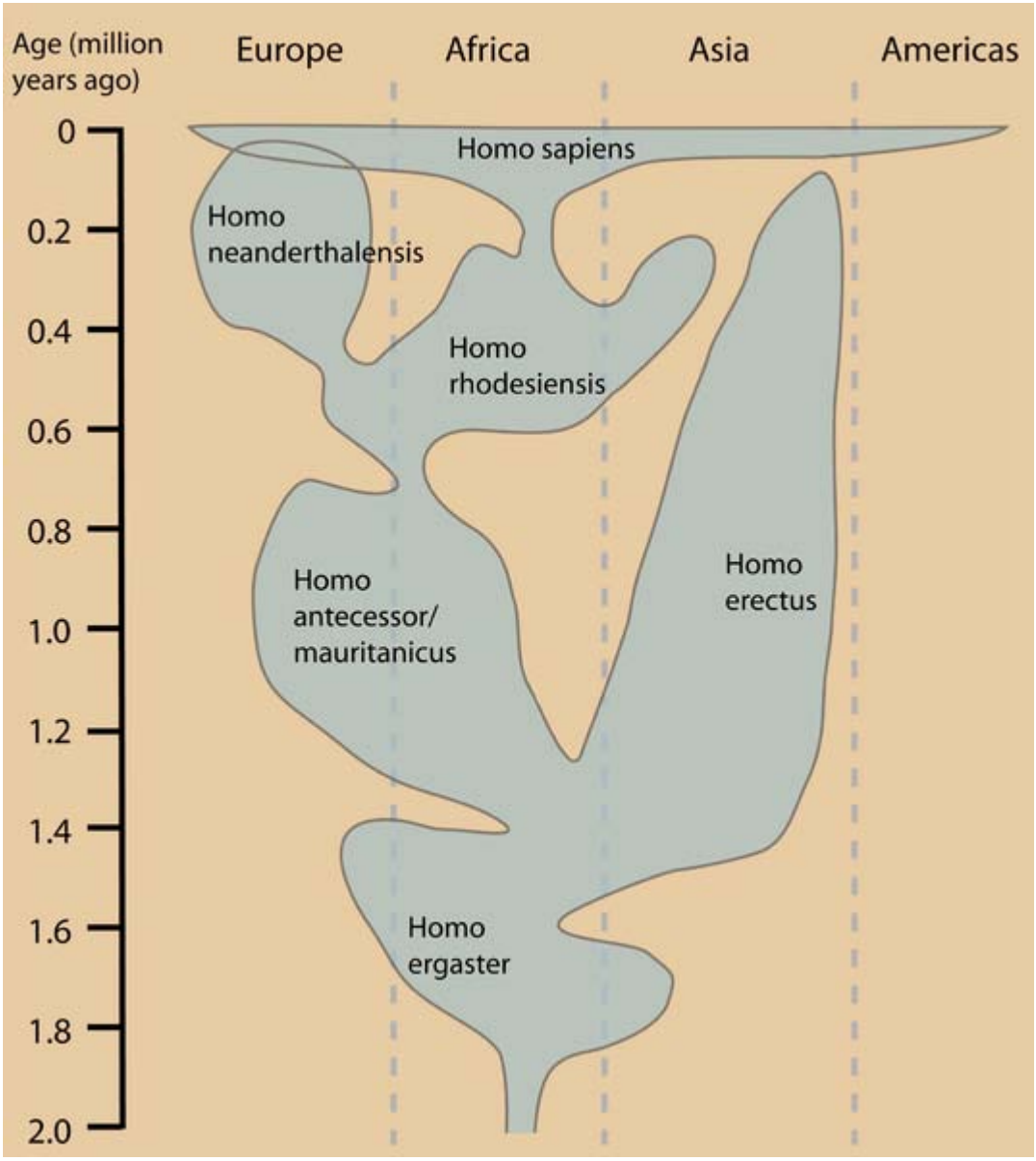
Homo habilis lived from about 2.4 to 1.4 Ma. *Homo habilis*, the first species of the genus *Homo*, evolved in South and East Africa in the late Pliocene or early Pleistocene, 2.5–2 Ma, when it diverged from the Australopithecines. *Homo habilis* had smaller molars and larger brains than the Australopithecines and made tools from stone and perhaps animal bones. One of the first known hominids, it was nicknamed 'handy man' by its discoverer, Louis Leakey due to its association with stone tools. Some scientists have proposed moving this species out of *Homo* and into *Australopithecus* due to the morphology of its skeleton being more adapted to living on trees rather than to moving on two legs like *Homo sapiens*.

H. rudolfensis and *H. georgicus*

These are proposed species names for fossils from about 1.9–1.6 Ma, the relation of which with *Homo habilis* is not yet clear.

- *Homo rudolfensis* refers to a single, incomplete skull from Kenya. Scientists have suggested that this was another *Homo habilis*, but this has not been confirmed.
- *Homo georgicus*, from Georgia, may be an intermediate form between *Homo habilis* and *Homo erectus*, or a sub-species of *Homo erectus*.

H. ergaster and *H. erectus*



One current view of the temporal and geographical distribution of hominid populations. Other interpretations differ mainly in the taxonomy and geographical distribution of hominid species.

The first fossils of *Homo erectus* were discovered by Dutch physician Eugene Dubois in 1891 on the Indonesian island of Java. He originally gave the material the name *Pithecanthropus erectus* based on its morphology that he considered to be intermediate between that of humans and apes. *Homo erectus* (H erectus) lived from about 1.8 Ma to about 70,000 years ago (which would indicate that they were probably wiped out by the Toba catastrophe; however, *Homo erectus soloensis* and *Homo floresiensis* survived it). Often the early phase, from 1.8 to 1.25 Ma, is considered to be a separate species, *Homo ergaster*, or it is seen as a subspecies of *Homo erectus*, *Homo erectus ergaster*. In the

early Pleistocene, 1.5–1 Ma, in Africa, Asia and Europe, some populations of *Homo habilis* are thought to have evolved larger brains and made more elaborate stone tools; these differences and others are sufficient for anthropologists to classify them as a new species, *Homo erectus*. In addition *Homo erectus* was the first human ancestor to walk truly upright. This was made possible by the evolution of locking knees and a different location of the foramen magnum (the hole in the skull where the spine enters). They may have used fire to cook their meat.

A famous example of *Homo erectus* is Peking Man; others were found in Asia (notably in Indonesia), Africa and Europe. Many paleoanthropologists now use the term *Homo ergaster* for the non-Asian forms of this group and reserve *Homo erectus* only for those fossils that are found in Asia and meet certain skeletal and dental requirements which differ slightly from *H. ergaster*.

H. cepranensis and *H. antecessor*

These are proposed as species that may be intermediate between *H. erectus* and *H. heidelbergensis*.

- *H. antecessor* is known from fossils from Spain and England that are dated 1.2 Ma–500 ka.
- *H. cepranensis* refers to a single skull cap from Italy, estimated to be about 800,000 years old.

H. heidelbergensis

H. heidelbergensis (Heidelberg Man) lived from about 800,000 to about 300,000 years ago. Also proposed as *Homo sapiens heidelbergensis* or *Homo sapiens paleohungaricus*.

H. rhodesiensis and the Gawis cranium

- *H. rhodesiensis*, estimated to be 300,000–125,000 years old. Most current experts believe Rhodesian Man to be within the group of *Homo heidelbergensis*, though other designations such as Archaic Homo sapiens and Homo sapiens rhodesiensis have also been proposed.
- In February 2006 a fossil, the Gawis cranium, was found which might possibly be a species intermediate between *H. erectus* and *H. sapiens* or one of many evolutionary dead ends. The skull from Gawis, Ethiopia, is believed to be 500,000–250,000 years old. Only summary details are known and no peer reviewed studies have been released by the finding team. Gawis man's facial features suggest its being either an intermediate species or an example of a "Bodo man" female.

H. neanderthalensis

Le Ferrassie Neanderthal skull (cast)

H. neanderthalensis lived from 400,000 to about 30,000 years ago. Also proposed as *Homo sapiens neanderthalensis*. Evidence from sequencing mitochondrial DNA indicated that no significant gene flow occurred between *H. neanderthalensis* and *H. sapiens* and, therefore, the two were separate species that shared a common ancestor about 660,000 years ago. In 1997, Mark Stoneking stated: "These results [based on mitochondrial DNA extracted from Neanderthal bone] indicate that Neanderthals did not contribute mitochondrial DNA to modern humans… Neanderthals are not our ancestors". Subsequent investigation of a second source of Neanderthal DNA supported these findings.

However, the 2010 sequencing of the Neanderthal genome indicated that Neanderthals did indeed interbreed with *H. sapiens circa* 75,000 BC (after *H. sapiens* moved out from Africa, but before they separated into Europe, the Middle East and Asia). Nearly all modern humans have 1% to 4% of their DNA derived from Neanderthal DNA. This 1–4% bit of DNA is only present in non-African humans. However, supporters of the multiregional hypothesis point to recent studies indicating non-African nuclear DNA heritage dating to one Ma, although the reliability of these studies has been questioned. Competition from *Homo sapiens* probably contributed to Neanderthal extinction. They could have coexisted in Europe for as long as 10,000 years.

Comparative table of *Homo* species

Comparative table of *Homo* species

Species	Lived when (Ma)	Lived where	Adult height	Adult mass	Cranial capacity (cm³)	Fossil record	Discovery / publication of name
H. antecessor	1.2 – 0.8	Spain	1.75 m (5.7 ft)	90 kg (200 lb)	1,000	2 sites	1997
H. cepranensis	0.9 – 0.8?	Italy			1,000	1 skull cap	1994/2003
H. erectus	1.5 – 0.2	Africa, Eurasia (Java, China, India, Caucasus)	1.8 m (5.9 ft)	60 kg (130 lb)	850 (early) – 1,100 (late)	Many	1891/1892
H. ergaster	1.9 – 1.4	Eastern and Southern Africa	1.9 m (6.2 ft)		700–850	Many	1975
H. floresiensis	0.10? – 0.012	Indonesia	1.0 m (3.3 ft)	25 kg (55 lb)	400	7 individuals	2003/2004
H. gautengensis	>2 – 0.6	South Africa	1.0 m (3.3 ft)			1 individual	2010/2010
H. georgicus	1.8	Georgia			600	4 individuals	1999/2002
H. habilis	2.3 – 1.4	Africa	1.0–1.5 m (3.3–4.9 ft)	33–55 kg (73–120 lb)	510–660	Many	1960/1964
H. heidelbergensis	0.6 – 0.35	Europe, Africa, China	1.8 m (5.9 ft)	60 kg (130 lb)	1,100–1,400	Many	1908
H. neanderthalensis	0.35 – 0.03	Europe, Western Asia	1.6 m (5.2 ft)	55–70 kg (120–150 lb) (heavily built)	1,200–1,900	Many	(1829)/1864
H. rhodesiensis	0.3 – 0.12	Zambia			1,300	Very few	1921

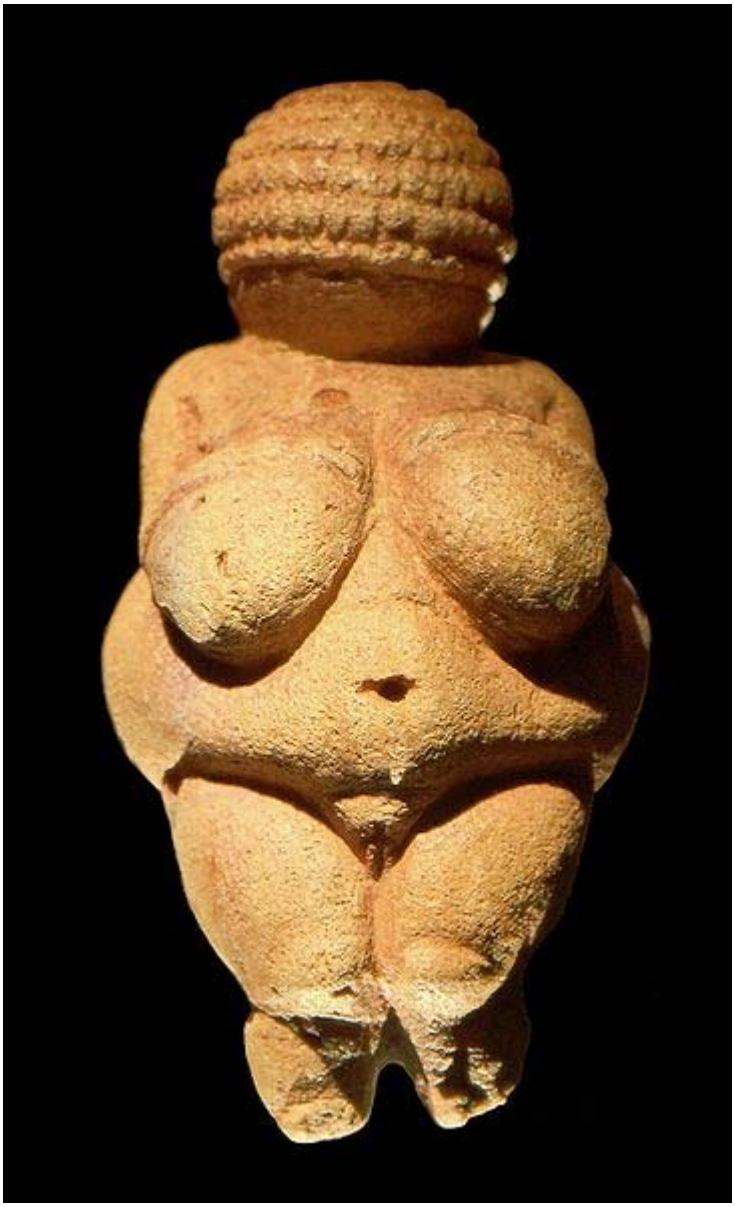
H. rudolfensis	1.9	Kenya				1 skull	1972/1986
H. sapiens idaltu	0.16 – 0.15	Ethiopia			1,450	3 craniums	1997/2003
H. sapiens sapiens (modern humans)	0.2 – present	Worldwide	1.4–1.9 m (4.6–6.2 ft)	50–100 kg (110–220 lb)	1,000–1,850	Still living	—/1758

Use of tools

"A sharp rock", an Oldowan pebble tool, the most basic of human stone tools

Fire, one of the greatest human discoveries

An Acheulean hand axe, the pinnacle of *Homo erectus* stone working



Venus of Willendorf, an example of Paleolithic art

Using tools has been interpreted as a sign of intelligence and it has been theorized that tool use may have stimulated certain aspects of human evolution—most notably the continued expansion of the human brain. Paleontology has yet to explain the expansion of this organ over millions of years despite being extremely demanding in terms of energy consumption. The brain of a modern human consumes about 20 watts (400 kilocalories per day), which is one fifth of the energy consumption of a human body. Increased tool use would allow hunting for energy-rich meat products and would enable processing more energy-rich plant products. Researchers have suggested that early hominids were thus under evolutionary pressure to increase their capacity to create and use tools.

Precisely when early humans started to use tools is difficult to determine, because the more primitive these tools are (for example, sharp-edged stones) the more difficult it is to decide whether they are natural objects or human artifacts. There is some evidence that the australopithecines (4 Ma) may have used broken bones as tools, but this is debated.

It should be noted that many species make and use tools, but it is the human species that dominates the areas of making and using more complex tools. The oldest known tools are the "Oldowan stone tools" from Ethiopia. It was discovered that these tools are from 2.5 to 2.6 million years old, which predates the earliest known "Homo" species. There is no known evidence that any "Homo" specimens appeared by 2.5 Ma. A Homo fossil was found near some Oldowan tools and its age was noted at 2.3 million years old, suggesting that maybe the Homo species did indeed create and use these tools. It is surely possible, but not solid evidence. Bernard Wood noted that "Paranthropus" coexisted with the early Homo species in the area of the "Oldowan Industrial Complex" over roughly the same span of time. Although there is no direct evidence that points to Paranthropus as the tool makers, their anatomy lends to indirect evidence of their capabilities in this area. Most paleoanthropologists agree that the early "Homo" species were indeed responsible for most of the Oldowan tools found. They argue that when most of the Oldowan tools were found in association with human fossils, Homo was always present, but Paranthropus was not.

In 1994, Randall Susman used the anatomy of opposable thumbs as the basis for his argument that both the Homo and Paranthropus species were toolmakers. He compared bones and muscles of human and chimpanzee thumbs, finding that humans have 3 muscles that chimps lack. Humans also have thicker metacarpals with broader heads, making the human hand more successful at precision grasping than the chimpanzee hand. Susman defended that modern anatomy of the human thumb is an evolutionary response to the requirements associated with making and handling tools and that both species were indeed toolmakers.

Stone tools

Stone tools are first attested around 2.6 Ma, when *H. habilis* in Eastern Africa used so-called pebble tools, choppers made out of round pebbles that had been split by simple strikes. This marks the beginning of the Paleolithic, or Old Stone Age; its end is taken to be the end of the last Ice Age, around 10,000 years ago. The Paleolithic is subdivided into the Lower Paleolithic (Early Stone Age, ending around 350,000–300,000 years ago), the Middle Paleolithic (Middle Stone Age, until 50,000–30,000 years ago) and the Upper Paleolithic.

The period from 700,000–300,000 years ago is also known as the Acheulean, when *H. ergaster* (or *erectus*) made large stone hand-axes out of flint and quartzite, at first quite rough (Early Acheulian), later "retouched" by additional, more subtle strikes at the sides of the flakes. After 350,000 BP (Before Present) the more refined so-called Levallois technique was developed. It consisted of a series of consecutive strikes, by which scrapers, slicers ("racloirs"), needles and flattened needles were made. Finally, after about

50,000 BP, ever more refined and specialized flint tools were made by the Neanderthals and the immigrant Cro-Magnons (knives, blades, skimmers). In this period they also started to make tools out of bone.

Modern humans and the "Great Leap Forward" debate

Until about 50,000–40,000 years ago the use of stone tools seems to have progressed stepwise. Each phase (*H. habilis*, *H. ergaster*, *H. neanderthalensis*) started at a higher level than the previous one, but once that phase started further development was slow. These *Homo* species were culturally conservative, but after 50,000 BC modern human culture started to change at a much greater speed. Jared Diamond, author of *The Third Chimpanzee* and some anthropologists characterize this as a "Great Leap Forward".

Modern humans started burying their dead, making clothing out of hides, developing sophisticated hunting techniques (such as using trapping pits or driving animals off cliffs) and engaging in cave painting. As human culture advanced, different populations of humans introduced novelty to existing technologies: artifacts such as fish hooks, buttons and bone needles show signs of variation among different populations of humans, something that had not been seen in human cultures prior to 50,000 BP. Typically, *H. neanderthalensis* populations do not vary in their technologies.

Among concrete examples of Modern human behavior, anthropologists include specialization of tools, use of jewellery and images (such as cave drawings), organization of living space, rituals (for example, burials with grave gifts), specialized hunting techniques, exploration of less hospitable geographical areas and barter trade networks. Debate continues as to whether a "revolution" led to modern humans ("the big bang of human consciousness"), or whether the evolution was more gradual.

Models of human evolution

Today, all humans belong to one population of *Homo sapiens sapiens,* undivided by species barrier. However, according to the "Out of Africa" model this is not the first species of hominids: the first species of genus *Homo*, *Homo habilis*, evolved in East Africa at least 2 Ma and members of this species populated different parts of Africa in a relatively short time. *Homo erectus* evolved more than 1.8 Ma and by 1.5 Ma had spread throughout the Old World.

Anthropologists have been divided as to whether current human population evolved as one interconnected population (as postulated by the Multiregional Evolution hypothesis), or evolved only in East Africa, speciated, then migrated out of Africa and replaced human populations in Eurasia (called the "Out of Africa" Model or the "Complete Replacement" Model).

Out of Africa

According to the Out of Africa model, developed by Chris Stringer and Peter Andrews, modern *H. sapiens* evolved in Africa 200,000 years ago. *Homo sapiens* began migrating from Africa between 70,000 – 50,000 years ago and eventually replaced existing hominid species in Europe and Asia. Out of Africa has gained support from research using mitochondrial DNA (mtDNA). After analysing genealogy trees constructed using 133 types of mtDNA, researchers concluded that all were descended from a woman from Africa, dubbed Mitochondrial Eve. Out of Africa is also supported by the fact that mitochondrial genetic diversity is highest among African populations.

There are differing theories on whether there was a single exodus or several. A multiple dispersal model involves the Southern Dispersal theory, which has gained support in recent years from genetic, linguistic and archaeological evidence. In this theory, there was a coastal dispersal of modern humans from the Horn of Africa around 70,000 years ago. This group helped to populate Southeast Asia and Oceania, explaining the discovery of early human sites in these areas much earlier than those in the Levant. A second wave of humans dispersed across the Sinai peninsula into Asia, resulting in the bulk of human population for Eurasia. This second group possessed a more sophisticated tool technology and was less dependent on coastal food sources than the original group. Much of the evidence for the first group's expansion would have been destroyed by the rising sea levels at the end of the Holocene era. The multiple dispersal model is contradicted by studies indicating that the populations of Eurasia and the populations of Southeast Asia and Oceania are all descended from the same mitochondrial DNA lineages, which support a single migration out of Africa that gave rise to all non-African populations.

The broad study of African genetic diversity headed by Dr. Sarah Tishkoff found the San people to express the greatest genetic diversity among the 113 distinct populations sampled, making them one of 14 "ancestral population clusters". The research also located the origin of modern human migration in south-western Africa, near the coastal border of Namibia and Angola.

According to the Toba catastrophe theory to which some anthropologists and archeologists subscribe, the supereruption of Lake Toba on Sumatra island in Indonesia roughly 70,000 years ago had global consequences, killing most humans then alive and creating a population bottleneck that affected the genetic inheritance of all humans today.

Recent and current human evolution

Natural selection is being observed in contemporary human populations, with recent findings demonstrating the population which is at risk of the severe debilitating disease kuru has significant over-representation of an immune variant of the prion protein gene G127V versus non-immune alleles. Scientists postulate one of the reasons for the rapid selection of this genetic variant is the lethality of the disease in non-immune persons. Other reported evolutionary trends in other populations include a lengthening of the reproductive period, reduction in cholesterol levels, blood glucose and blood pressure.

In their 2009 book *The 10,000 Year Explosion*, Gregory Cochran and Henry Harpending argue that human evolution has accelerated since and as a result of the development of agriculture and civilisation since some 50,000 years ago and that there are consequently substantial genetic differences between different current human populations.

Notable human evolution researchers

- Robert Broom, a Scottish physician and palaeontologist whose work on South Africa led to the discovery and description of the Paranthropus genus of hominins and of "Mrs. Ples"
- Raymond Dart, an Australian anatomist and palaeoanthropologist, whose work at Taung, in South Africa, led to the discovery of *Australopithecus africanus*
- Charles Darwin, a British naturalist who documented considerable evidence that species originate through evolutionary change
- Henry McHenry, an American anthropologist who specializes in studies of human evolution, the origins of bipedality and paleoanthropology
- Donald Johanson, credited with the discovery of *Australopithecus afarensis*
- Jeffrey Laitman, an American anatomist and physical anthropologist whose work has explored the evolution of the vocal tract and speech
- Louis Leakey, an African archaeologist and naturalist whose work was important in establishing human evolutionary development in Africa
- Mary Leakey, a British archaeologist and anthropologist whose discoveries in Africa include the Laetoli footprints
- Richard Leakey, an African paleontologist and archaeologist, son of Louis and Mary Leakey
- Svante Pääbo, a Swedish biologist specializing in evolutionary genetics
- David Pilbeam, a paleoanthropologist, researcher and writer on a range of topics involving human and primate evolution.
- Jeffrey H. Schwartz, an American physical anthropologist and professor of biological anthropology
- Chris Stringer, anthropologist, leading proponent of the recent single origin hypothesis
- Alan Templeton, geneticist and statistician, proponent of the multiregional hypothesis
- Philip V. Tobias, a South African palaeoanthropologist is one of the world's leading authorities on the evolution of humankind
- Erik Trinkaus, a prominent American paleoanthropologist and expert on Neanderthal biology and human evolution
- Milford H. Wolpoff, an American paleoanthropologist who is the leading proponent of the multiregional evolution hypothesis.

Chapter- 2

Denisova Hominin

The **Denisova hominin** is the name given to the remains of a member of the genus *Homo* that may be a previously unknown species based on an analysis of its mitochondrial DNA (mtDNA). In March 2010, discovery was announced of bone fragments of a juvenile that lived about 41,000 years ago found in Denisova Cave (Altai Krai, Russia), a region also inhabited at about the same time by Neanderthals and modern humans. The mtDNA of the Denisova hominin is distinct from the mtDNAs of Neanderthals and modern humans. In December 2010, an international team of scientists determined the sequence from the nuclear genome of this group (known as the **Denisovans**) from this finger bone. According to their analysis, this group shares a common origin with the Neanderthals and interbred with the ancestors of modern Melanesians.

Anatomy and lineage

Little is known of the precise anatomical features of the Denisovans since the only physical remains discovered thus far are the finger bone from which only mitochondrial genetic material was gathered. A tooth found in Denisova Cave carries a mtDNA very similar to that of the finger bone and shares no derived morphological features with Neanderthal or modern humans. The Siberian bone's mtDNA differs from that of modern humans by 385 bases (nucleotides) in the mtDNA strand out of approximately 16,500, whereas the difference between modern humans and Neanderthals is around 202 bases. In contrast, the difference between chimpanzees and modern humans is approximately 1,462 mtDNA base pairs. Analysis of the specimen's genome shows it to be due to a common branch of ancestors with Neanderthal lineage, but, after they diverged from one another, Denisovans and Neanderthals had largely separated population histories.

Discovery

In 2008, Russian archeologists working at the site of Denisova Cave in the Altai Mountains of Siberia uncovered a small bone fragment from the fifth finger of a juvenile hominin, dubbed the "X-woman" (referring to the maternal descent of mitochondrial DNA), or the Denisova hominin. Artifacts, including a bracelet, excavated in the cave at the same level were carbon dated to around 40,000 BP.

A team of scientists led by Johannes Krause and Swedish biologist Svante Pääbo from the Max Planck Institute for Evolutionary Anthropology in Leipzig, Germany, sequenced

mtDNA extracted from the fragment. Because of the cool climate in the location of the Denisova Cave, the discovery benefited from DNA's ability to survive for longer periods at lower temperatures. The analysis indicated that modern humans, Neanderthals and the Denisova hominin last shared a common ancestor around 1 million years ago. Some studies suggest that modern humans coexisted with Neanderthals in Europe and the discovery raises the possibility that Neanderthals, modern humans and the Denisovan hominin may have co-existed.

The DNA analysis further indicated that this new hominin species was the result of an early migration out of Africa, distinct from the later out-of-Africa migrations associated with Neanderthals and modern humans, but also distinct from the earlier African exodus of *Homo erectus*. Professor Chris Stringer, human origins researcher at London's Natural History Museum and one of the leading proponents of the recent single-origin hypothesis, remarked: "This new DNA work provides an entirely new way of looking at the still poorly understood evolution of humans in central and eastern Asia." Pääbo noted that the existence of this distant branch creates a much more complex picture of humankind during the Late Pleistocene.

In 2010, a second paper from the Svante Pääbo group reported the prior discovery, in 2000, of a third upper molar from a young adult, dating from about the same time (the finger was from level 11 in the cave sequence, the tooth from level 11.1). The tooth differed in several aspects from those of Neanderthals while having archaic characteristics similar to the teeth of *Homo erectus*. They again performed mitochondrial DNA analysis on the tooth and found it to have a different but similar sequence to that of the finger bone, indicating a divergence time about 7,500 years before and suggesting it belonged to a different individual from the same population.

Nuclear genome analysis

In the same 2010 paper, the authors report the isolation and sequencing of nuclear DNA from the Denisova finger bone. This specimen showed an unusual degree of DNA preservation and low level of contamination. They were able to achieve near-complete genomic sequencing, allowing a detailed comparison with Neanderthal and modern humans. From this analysis, they concluded that in spite of the apparent divergence of their mitochondrial sequence, the Denisova population along with Neanderthal shared a common branch from the lineage leading to modern African humans. The estimated time of divergence between Denisovans and Neanderthals is 640,000 years ago and that between both these groups and modern Africans is 804,000 years ago. They suggest that the divergence of the Denisova mtDNA results either from the persistence of a lineage purged from the other branches of humanity through genetic drift or else an introgression from an older hominin lineage.

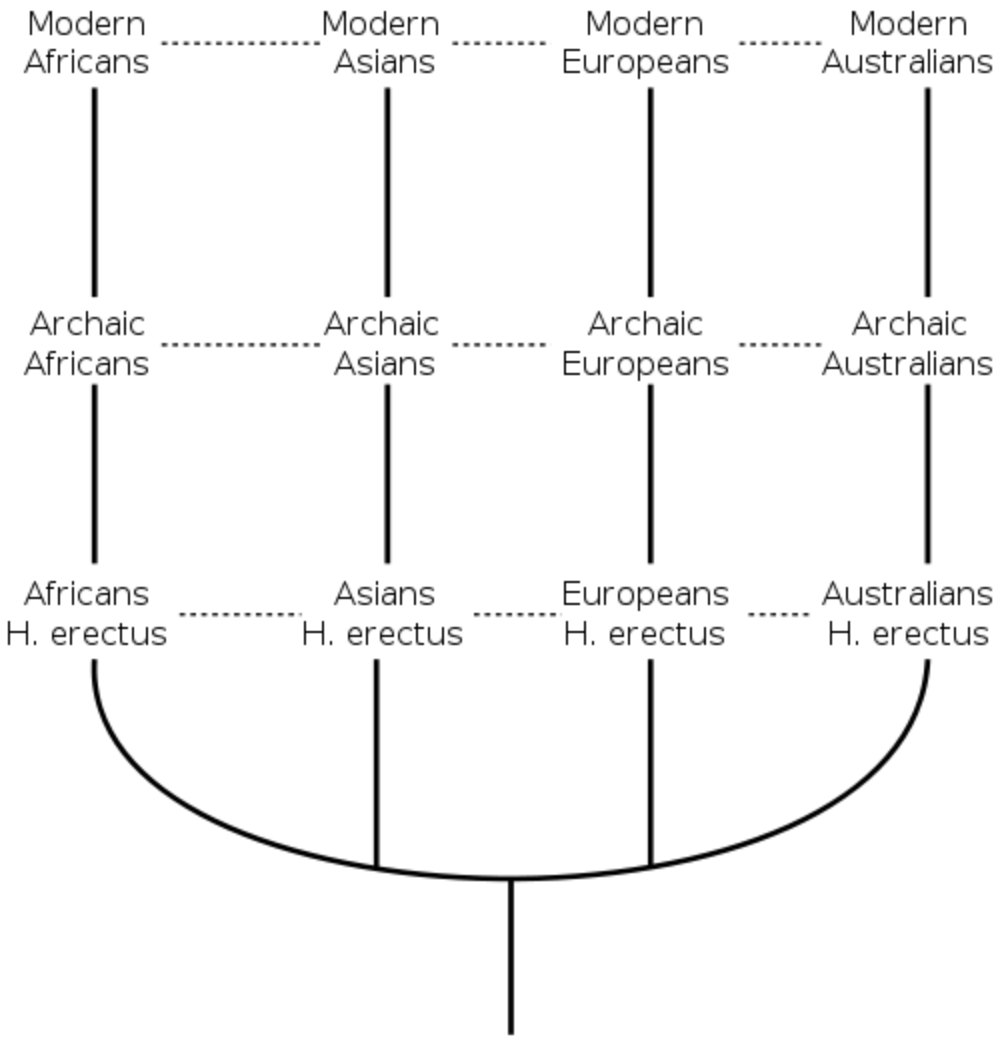
Interbreeding with modern humans

In addition to genetic studies linking approximately 4% of non-African modern human DNA to Neanderthals, these tests comparing the Denisova hominin genome with those of

six modern humans whose genome has been sequenced, a !Kung from South Africa, a Nigerian, a French person, a Papua New Guinean, a Bougainville Islander and a Han Chinese showed that between 4% and 6% of the genome of Melanesians (represented by the Papua New Guinean and Bougainville Islander) derives from a Denisovan population, possibly introduced during the early migration of the ancestors of Melanesians into Southeast Asia. This history of interaction suggests that Denisovans once ranged widely over eastern Asia.

Chapter- 3

Multiregional Origin of Modern Humans



A graph detailing the evolution to modern humans using the Multiregional theory of human evolution. The horizontal lines represent 'multiregional evolution' gene flow between regional lineages.

The **multiregional hypothesis** is a scientific model that provides an explanation for the pattern of human evolution. The hypothesis holds that the evolution of humanity from near the beginning of the Pleistocene two million years ago to the present day has been within a single, continuous human species. This species encompasses archaic human forms such as *Homo erectus* and Neanderthals as well as modern forms, which are held to be subspecies and evolved worldwide to the diverse populations of modern *Homo sapiens sapiens*. The theory contends that there was some human genetic continuity in various regions of the world as well as gene interchange between the regions. Proponents of multiregional origin point to fossil and genomic evidence as support for their hypothesis.

The primary competing hypothesis is recent African origin of modern humans (also known as "Out of Africa"), which contends that modern humans arose in Africa around 100-200,000 years ago, moving out of Africa around 50-60,000 years ago to replace the other human forms without interbreeding.

Regional continuity

The term "multiregional hypothesis" was first coined in the early 1980s by Milford H. Wolpoff and colleagues as an explanation for the apparent similarities seen in *Homo erectus* and *Homo sapiens* fossils from the same region, what they called *regional continuity*.

Wolpoff rejected the earlier proposal by Coon of parallel evolution and proposed a theory based on clinal variation that would allow for the necessary balance between local selection and a global species. He proposed that *Homo erectus*, Neanderthals, *Homo sapiens* and other humans were a single species. This species arose in Africa two million years ago as *H. erectus* and then spread out over the world, developing adaptations to regional conditions. It was proposed that for periods of time some populations became isolated, developing in a different direction, but through continuous interbreeding, replacement, genetic drift and selection, adaptations that were an advantage anywhere on earth would spread, keeping the development of the species in the same overall direction, while maintaining adaptations to regional factors. Eventually, the more unusual local varieties of the species would have disappeared in favor of modern humans, retaining some regional adaptations, but with many common features.

Fossil evidence

Replica of Sangiran 17 *Homo erectus* skull from Indonesia showing obtuse face to vault angle determined by fitting of bones at brow

Cast of anatomically modern human Kow Swamp 1 skull from Australia with a face to vault angle matching that of Sangiran 17

Proponents of the multiregional hypothesis see regional continuity of certain morphological traits from archaic humans to modern humans, demonstrating regional genetic continuity, even as changes in other traits occur in parallel over time across all regions, demonstrating lateral genetic exchange. For example, in 2001 Wolpoff and colleagues published an analysis of character traits of the skulls of early modern human fossils in Australia and central Europe. They concluded that the diversity of these recent humans could not "result exclusively from a single late Pleistocene dispersal" and implied dual ancestry from Javan *Homo erectus* for Australia and from Neanderthals for Central Europe.

Southeast Asia

Alan Thorne held that there was regional continuity in the human fossils in southeast Asia. Wolpoff, initially skeptical, became convinced when reconstructing the Sangiran 17 *Homo erectus* skull from Indonesia, when he was surprised that the skull's face to vault angle matched that of the Australian modern human Kow Swamp 1 skull. Wolpoff had expected the skull to match that of the *Homo erectus* specimens from China like the Dali skull, but instead, the face to vault angle seemed to be retained regionally over time, even while the fossils in the two regions showed parallel increases in brain case size and parallel reductions in masticatory structures over the intervening approximately 750,000 years.

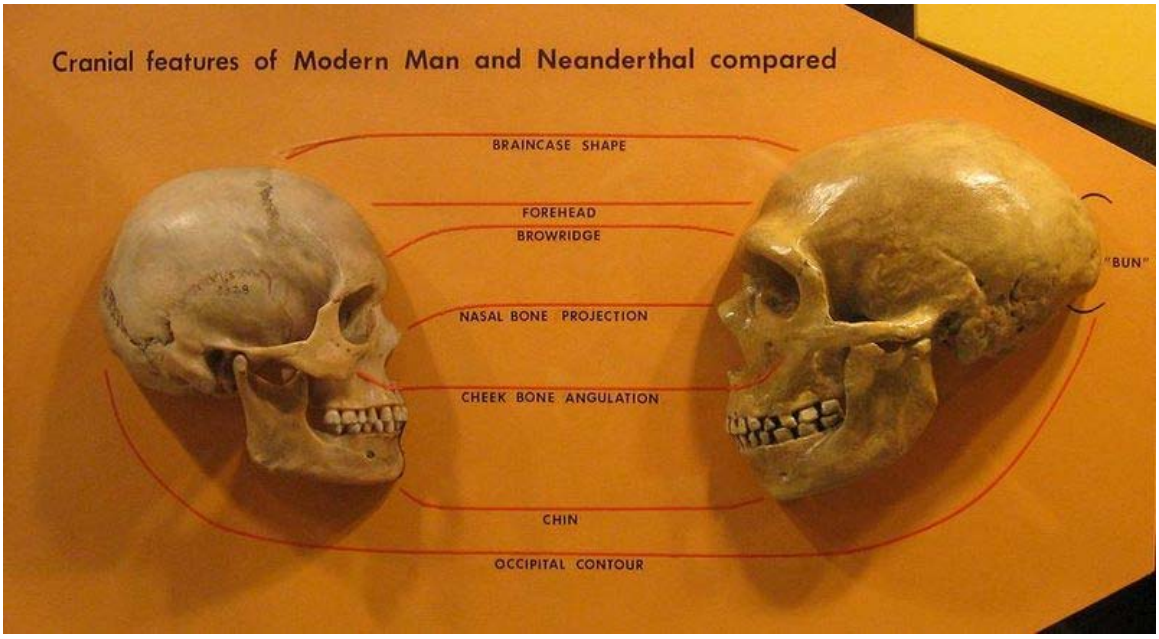
China

Replica of *Homo erectus* ("Peking man") skull from China

Franz Weidenreich, who oversaw the excavations of numerous "Peking man" *Homo erectus* fossils at Zhoukoudian in the early 20th century, believed the fossil record demonstrated certain unique features linking prehistoric and modern human populations in China. Many subsequent Chinese paleoanthropologists, such as Wu Xinzhi, were also disposed to favor the multiregional hypothesis for the same reason.

More recent finds provide additional support for regional human continuity in China. The *Tianyuan 1* specimen unearthed in 2003 in Tianyuan Cave, Zhoukoudian and Carbon 14 dated to 42-39 kya exhibits a series of typical modern human features such as a distinct chin. However, the skeleton also has archaic traits such as low anterior to posterior dental proportions indicating relatively large molars and certain leg bone proportions typical of archaic forms such as Neanderthals. Shang *et. al.* conclude that this combination of modern and archaic traits "implies that a simple spread of modern humans from Africa is unlikely." A jaw bone found in 2008 and dated to 110,000 kya may also exhibit a mixture of archaic and modern human traits.

Europe



Comparison of modern human and neanderthal skull

Proponents of the multiregional hypothesis argue for regional continuity in Europe on the basis of skeletal anatomy, morphology and genetics of speech and the archaeology of the middle to upper paleolithic transition, which they believe to be inconsistent with the possibility of complete replacement of the Neanderthals in Europe without interbreeding.

Some detractors of the theory have argued, in contrast, that the morphological differences between Neanderthals and early and modern humans indicate that they are different species, based on skull differences more distinct than between any subspecies pairs examined except for the two subspecies of gorilla, implying limited or no interbreeding.

Many of the multiregional claims regarding skeletal morphology in Europe center on forms with both archaic Neanderthal traits and modern traits, to provide evidence of interbreeding rather than replacement. Examples include the *Lapedo child* found in Portugal and the *Oase 1* mandible from Peştera cu Oase, Romania, though the *Lapedo child* example is disputed by some. In a 2007 paper examining numerous samples from European early modern humans, later European humans from the Gravettian period and the earlier Neanderthal and east African populations from whom the first two populations could have descended, Erik Trikaus identified numerous features in the later European samples which were absent from the African sample, but present in the Neanderthal sample. These features included various aspects of skull and mandible shape, tooth shape and size and shapes and proportions of other bones. Trinkaus concluded that early modern Europeans had predominant African ancestry with a substantial degree of admixture from the Neanderthals then indigenous to Europe.

Genetic evidence

Genetic evidence from the late 1980s on the mitochondrial genome indicated that all living humans had as an ancestor a single female living in African about 200,000 years ago. This led to a hypothesis that around that time period, a small founder population of humans left Africa and eventually replaced all archaic humans then living outside of Africa without interbreeding, contrary to the multiregional hypothesis. However, as data on the far larger autosomal DNA genome started to became available, evidence mounted that genetic contributions from archaic human populations from around the world and not just from Africa, also persist in modern humans. Recent analyses of DNA taken directly from Neanderthal and denisovan specimens indicates that those populations also contributed to the genome of living humans, as predicted by the multiregional hypothesis.

Mitochondrial DNA

A 1987 analysis of mitochondrial DNA from 147 people from around the world indicated that their mitochondrial lineages all traced to a common ancestor in Africa about 200,000 years ago. The analysis suggested that this reflected the worldwide expansion of modern humans as a new species, replacing rather than mixing with local archaic humans. Later analysis of mitochondrial DNA from neanderthals and from the denisova hominin indicated that those mitochondrial strains had diverged from the living human mitochondrial line long before 200,000 years ago, consistent with lack of interbreeding between early modern and archaic humans.

The original mitochondrial DNA results and the resulting recent African replacement theory posed a serious challenge to the multiregional hypothesis. Mitochondrial DNA alone, however, could not entirely rule out interbreeding between early modern and archaic humans, since archaic human mitochondrial strains from such interbreeding could have been lost due to genetic drift. Indeed, later analysis of autosomal DNA from both modern and archaic humans was to show results very different from those from mitochondrial DNA.

Autosomal and X chromosome DNA

By analysing haplotype data, Alan Templeton found support for three waves of human migration out of Africa, the first being 1.9 million years ago and concluded that it was impossible that existing Eurasian populations had not interbred with African migrants.

Studies on past population bottlenecks that can be inferred from molecular data have led multiregionalists to conclude that the recent single-origin hypothesis is untenable because there are no population size bottlenecks affecting all genes that are more recent than 2 million years ago.

- CMAH CMP-N-acetylneuraminic acid hydroxylase pseudogene show 2.9 Mya coalescence time.

- NAT2 SNPs lineages cluster in sub-Saharan Africa, Europe and East Asia, with genetic distances scaling with geographic distances. The NAT1 lineage tree is rooted in Eurasia with a coalescence time of 2.0 Mya that cannot be explained by balancing selection and the NAT1*11A haplotype absent from subsaharan Africa.

- ALMS1 suggest ancient and complex evolutionary history with a coalescence time of about 2 Mya.

- Analyses of a region of RRM2P4 (ribonucleotide reductase M2 subunit pseudogene 4) showed a coalescence time of about 2 Mya, with a clear root in Asia.

- PDHA1 (pyruvate dehydrogenase) locus on X chromosome has estimated coalescent-time depth of 1.86 Ma, although the worldwide lineage pattern is unlike other autosomal sites and consistent with recent dispersal from Africa.

- MAPT locus 17q21.3 split into deep genetic lineages H1 and H2. H2 lineage in European population suggest inheritance from Neanderthals.

- ASAH1. Related to mental activity N-Acylsphingosine Amidohydrolase gene two V and M deep genetic lineages have TMRCA 2.4±.4 Ma. Linkage disequilibrium 62% and small nucleotide diversity 0.05% indicate a signature of positive Darwinian selection for the V lineage. The M lineage is attributed to ancient population structure of humans in Africa.

- X-chromosome genes DMD44, APXL, AMELX, TNFSF5 show S-N heterogeneous patterns of variation. While these genes do show the greatest diversity in Africa, consistent with recent African replacement, the genes' varying diversity by region cannot be explained by simple expansion from Africa, which should have resulted in similar diversity patterns for all these genes.

- Genome polymorphism: Inversion polymorphism: known 5-million-base pair (Mbp) 8p23.1, 1-Mbp on 17q21.3 and novel 1.2-Mbp on 15q24, 2.1-Mbp 15q13, 1.7-Mbp 17q12. In the sample of 8 gnomes from worldwide sample including Yuruba Kidd&al group found 4 million SNPs and 796,273 small indels (1−100 bp in size); 15 large regions of excess nucleotide variation 500 kbp to 3 Mbp. Two of variable sites are described detailed above.

- Microcephalin D allele introgressed into the modern human gene pool points to the Neanderthal lineage as possible source and compelling evidence of admixture among the human loci.

Proponents of the multiregional hypothesis show genetic sequences of several loci in the human genome with million year old genealogy. Those data of deep genetic lineages are explained in the multiregional theory framework as a result of heredity from *structured ancestral population*. The data are not interpreted in light of the RAO hypothesis postulating recent replacement where separated million years ago genetic lineages are at best unpredicted.

DNA from archaic humans

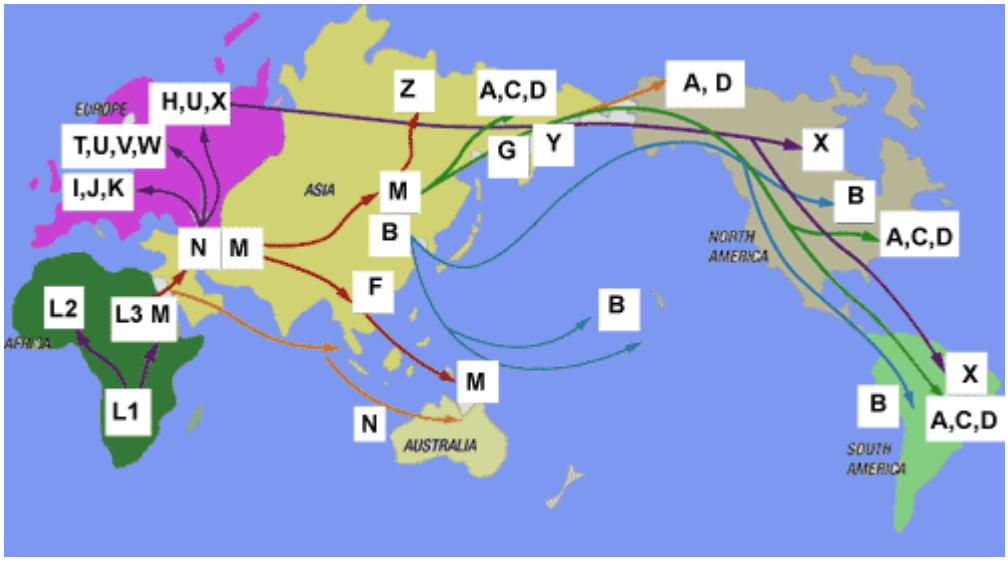
By 2006, extraction of DNA directly from some archaic human samples was becoming possible. The earliest analyses were of neanderthal DNA and indicated that the neanderthal contribution to modern human genetic diversity was no more than 20%, with a most likely value of 0%. By 2010, however, detailed DNA sequencing of the neanderthal specimens from Europe indicated that the contribution was nonzero, with neanderthals sharing 1-4% more genetic variants with living nonafricans than with living humans in subsaharan Africa, supporting regional continuity outside of Africa. In late 2010, a recently discovered nonneanderthal archaic human, the denisova hominin from southern siberia, was found to share 4-6% of its genome with living melanesian humans and with no other living group, supporting lateral gene transfer between two regions outside of Africa. The combination of regional continuity inside and outside of Africa and lateral gene transfer between various regions around the world supports the multiregional hypothesis.

Alternate hypotheses

Polygenism

The polygenic hypothesis for human origins, popular in the 19th and early 20th centuries, proposed that human populations had evolved independently and in parallel in the various regions of the world, without gene interchange. This differs from multiregional evolution in that the multiregional model posits significant lateral gene flow through migrations or interbreeding between populations, while polygenism envisions none. The polygenic hypothesis is no longer considered a viable scientific hypothesis since separately evolving species could not develop the interfertility of modern human populations.

Recent African replacement



Map of early human migrations according to mitochondrial population genetics

In paleoanthropology, the **recent African origin of modern humans** is the mainstream model describing the origin and early dispersal of anatomically modern humans. The theory is called the *(Recent) Out-of-Africa* model in the popular press and academically the *recent single-origin hypothesis* (*RSOH*), *Replacement Hypothesis* and *Recent African Origin* (*RAO*) model. The hypothesis that humans have a single origin (monogenesis) was published in Charles Darwin's *Descent of Man* (1871). The concept was speculative until the 1980s, when it was corroborated by a study of present-day mitochondrial DNA, combined with evidence based on physical anthropology of archaic specimens. According to genetic and fossil evidence, archaic *Homo sapiens* evolved to anatomically modern humans solely in Africa, between 200,000 and 100,000 years ago, with members of one branch leaving Africa by 60,000 years ago and over time replacing earlier human populations such as Neanderthals and *Homo erectus*.

The recent single origin of modern humans in East Africa was the near-consensus position held within the scientific community until 2010. However, recent sequencing of autosomal DNA from neanderthals and from an archaic human from denisova suggest that these populations, which were already outside of Africa at the posited time of the recent African human origin, also contributed to the modern human gene pool.

The competing hypothesis is the multiregional origin of modern humans. Some push back the original "out of Africa" migration—in this case, by *Homo erectus*, not by *Homo sapiens*—to two million years ago.

History of the theory

With the development of anthropology in the early 19th century, scholars disagreed vigorously about different theories of human development. Those such as Johann Friedrich Blumenbach and James Cowles Pritchard held that since the creation, the various human races had developed as different varieties sharing descent from one people (monogenism). Their opponents, such as Louis Agassiz and Josiah C. Nott, argued for polygenism, or the separate development of human races as separate species, or had developed as separate species through transmutation of species from apes, with no common ancestor.

Charles Darwin was one of the first to propose common descent of living organisms and among the first to suggest that all humans had in common ancestors who lived in Africa. In the *Descent of Man*, he speculated that humans had descended from apes which still had small brains but walked upright, freeing their hands for uses which favoured intelligence. Further, he thought such apes were African:

In each great region of the world the living mammals are closely related to the extinct species of the same region. It is, therefore, probable that Africa was formerly inhabited by extinct apes closely allied to the gorilla and chimpanzee; and as these two species are now man's nearest allies, it is somewhat more probable that our early progenitors lived on the African continent than elsewhere. But it is useless to speculate on this subject, for an ape nearly as large as a man, namely the Dryopithecus of Lartet, which was closely allied to the anthropomorphous Hylobates, existed in Europe during the Upper Miocene period; and since so remote a period the earth has certainly undergone many great revolutions and there has been ample time for migration on the largest scale.
—Charles Darwin, *Descent of Man*

The prediction was insightful because at the time, in 1871, there were hardly any human fossils of ancient hominids available. Almost fifty years later, Darwin's speculation was supported when anthropologists began finding numerous fossils of ancient small-brained hominids in several areas of Africa (list of hominina fossils).

The debate in anthropology had swung in favour of monogenism by the mid-20th century. Isolated proponents of polygenism held forth in the mid-20th century, such as Carleton Coon, who hypothesized as late as 1962 that *Homo sapiens* arose five separate times from *Homo erectus* in five separate places. The "Recent African origin" of modern humans means "single origin" (monogenism) and has been used in various contexts as an antonym to polygenism.

With the advent of archaeogenetics in the 1990s, scientists were able to date the "out of Africa" migration with some confidence. In 2000, the mitochondrial DNA (mtDNA) sequence of "Mungo Man" of ancient Australia was published. This work was later questioned and explained by W. James Peacock, leader of the team who sequenced Mungo man's ancient mtdna.

The question of whether there was inheritance of other typological (not *de facto*) *Homo* subspecies into the *Homo sapiens* genetic pool remains under debate.

Early Homo sapiens

Anatomically modern humans originated in Africa about 250,000 years ago. The trend in cranial expansion and the acheulean elaboration of stone tool technologies which occurred between 400,000 years ago and the second interglacial period in the Middle Pleistocene (around 250,000 years ago) provide evidence for a transition from *H. erectus* to *H. sapiens*. In the Recent African Origin (RAO) scenario, migration within and out of Africa eventually replaced the earlier dispersed *H. erectus*.

Homo sapiens idaltu, found at site Middle Awash in Ethiopia, lived about 160,000 years ago. It is the oldest known anatomically modern human and classified as an extinct subspecies. Fossils of early *Homo sapiens* were found in Qafzeh cave in Israel and have been dated to 80,000 to 100,000 years ago. However these humans seem to have either become extinct or retreated back to Africa 70,000 to 80,000 years ago, possibly replaced by south bound Neanderthals escaping the colder regions of ice age Europe. Hua Liu & al. analyzing autosomal microsatellite markers dates to c. 56,000±5,700 years ago mtDNA evidence. He interprets the paleontological fossil of early modern human from Qafzeh cave as an isolated early offshoot that retracted back to Africa.

All other fossils of fully modern humans outside Africa have been dated to more recent times. The oldest well dated fossils found outside Africa are from Lake Mungo, Australia and have been dated to about 42,000 years ago. The Tianyuan cave remains in Liujiang region China have a probable date range between 38,000 and 42,000 years ago. They are most similar in morphology to Minatogawa Man, modern humans dated between 17,000 and 19,000 years ago and found on Okinawa Island, Japan. However, others have dated Liujang Man to 111,000 to 139,000 years before the present.

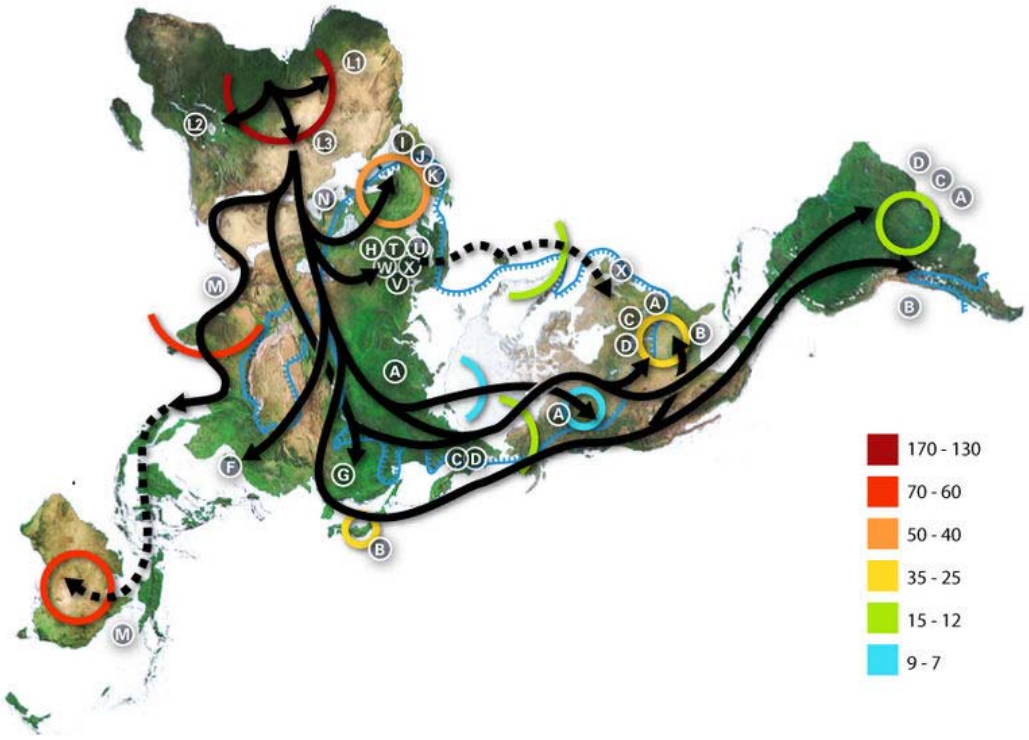
Beginning about 100,000 years ago evidence of more sophisticated technology and artwork begins to emerge and by 50,000 years ago fully modern behaviour becomes more prominent. By this time the ritual burying of the dead is noted. Stone tools show regular patterns that are reproduced or duplicated with more precision while tools made of bone and antler appear for the first time.

Genetic reconstruction

Two pieces of the human genome are quite useful in deciphering human history: mitochondrial DNA and the Y chromosome. These are the only two parts of the genome that are not shuffled about by the evolutionary mechanisms that generate diversity with each generation: instead, these elements are passed down intact. According to the hypothesis, all humans alive today inherited their mitochondria from one woman who lived in Africa about 160,000 years ago; she has been named Mitochondrial Eve. All men today have inherited their Y chromosomes from a man who lived 60,000 years ago, probably in Africa. He has been named Y-chromosomal Adam. It is now believed that

more men participated in the out of Africa exodus of early humans than women based on comparing non-sex-specific chromosomes with sex-specific ones.

Mitochondrial DNA



One model of human migration based on Mitochondrial DNA

The first lineage to branch off from Mitochondrial Eve is L0. This haplogroup is found in high proportions among the San of Southern Africa, the Sandawe of East Africa. It is also found among the Mbuti people. These groups branched off early in human history and have remained relatively genetically isolated since then. Haplogroups L1, L2 and L3 are descendents of L1-6 and are largely confined to Africa. The macro haplogroups M and N, which are the lineages of the rest of the world outside Africa, descend from L3.

Y-chromosomal DNA

The mutations defining macro-haplogroup CT (all Y haplogroups except A and B) predate the "Out of Africa" migration, its descendent macro-group DE being confined to Africa. The mutations that distinguish Haplogroup C from all other descendants of CR have occurred some 60,000 years ago, shortly after the first Out of Africa migration.

Haplogroup F originated some 45,000 years ago, either in North Africa (in which case it would point to a second wave of out-of-Africa migration) or in South Asia. More than 90% of males not native to Africa are descended in direct male line from the first bearer of haplogroup F.

Autosomal DNA

Analysis of 53 populations from genome-wide SNP data from 1138 unrelated individuals revealed that the population groups studied fell into just three genetic groups: Africans, Eurasians (which includes natives of Europe and the Middle East and Southwest Asians east to present-day Pakistan) and East Asians, which includes natives of Asia, Japan, Southeast Asia, the Americas and Oceania. The study determined that most ethnic group differences can be attributed to genetic drift, with modern African populations having greater genetic diversity than the other two genetic groups consistent with human origin in Africa.

Other analyses of autosomal DNA from modern humans, however, indicated a likelihood of recent admixture from archaic human populations from outside of Africa. In 2010, analysis of two sources of archaic human autosomal DNA provided evidence for specific instances of admixture, in the form of a 1-4% shared genome between neanderthals and nonafrican modern humans not shared by modern Africans and a 4-6% shared genome between an archaic human recently discovered in denisova and modern melanesians not shared by other modern humans.

Exodus from Africa

Red Sea crossing

Some 70 millennia ago, a part of the bearers of mitochondrial haplogroup L3 migrated from East Africa into the Near East.

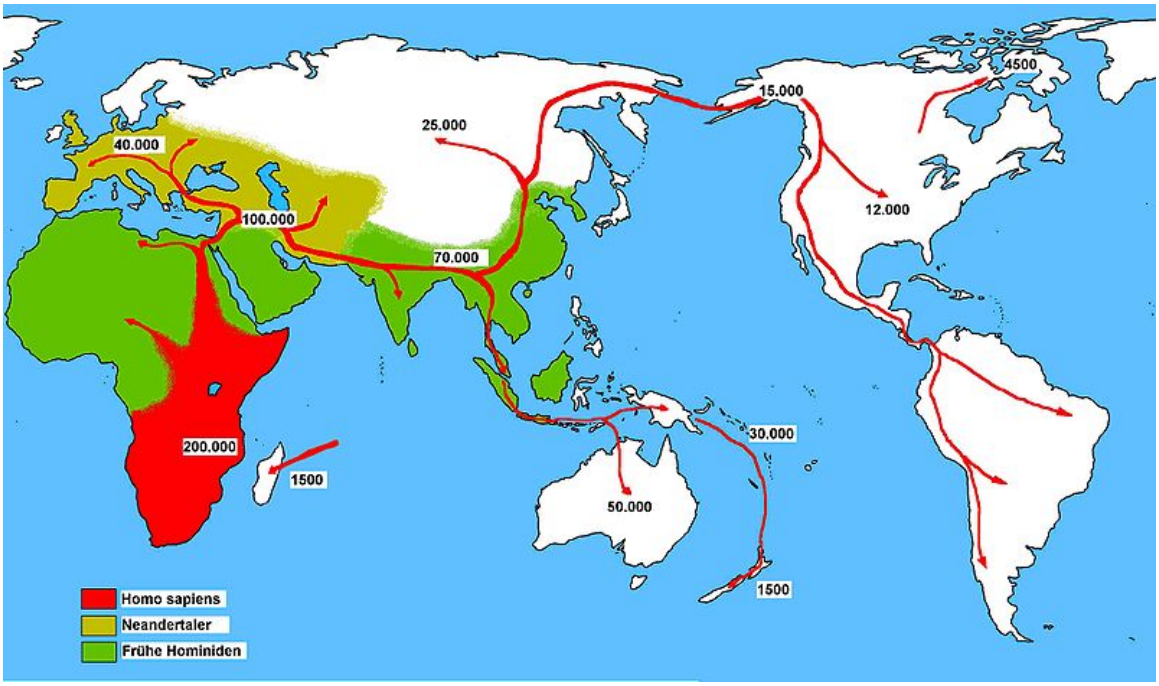
Some scientists believe that only a few people left Africa in a single migration that went on to populate the rest of the world, based in the fact that only descents of L3 are found outside Africa. From that settlement, some others point to the possibility of several waves of expansion. For example, Wells says that the early travelers followed the southern coastline of Asia, crossed about 250 kilometers [155 miles] of sea and colonized Australia by around 50,000 years ago. The Aborigines of Australia, Wells says, are the descendants of the first wave of migrations.

It has been estimated that from a population of 2,000 to 5,000 in Africa, only a small group of possibly 150 people crossed the Red Sea. This is because, of all the lineages present in Africa, only the daughters of one lineage, L3, are found outside Africa. Had there been several migrations one would expect more than one African lineage outside Africa. L3's daughters, the M and N lineages, are found in very low frequencies in Africa (although haplogroup M1 is very ancient and diversified in North and Northeast Africa) and appear to be recent arrivals. A possible explanation is that these mutations occurred in East Africa shortly before the exodus and by the founder effect became the dominant haplogroups after the exodus from Africa. Alternatively, the mutations may have arisen shortly after the exodus from Africa.

Other scientists have proposed a Multiple Dispersal Model, in which there were two migrations out of Africa, one across the Red Sea travelling along the coastal regions to India (the Coastal Route), which would be represented by Haplogroup M. Another group of migrants with Haplogroup N followed the Nile from East Africa, heading northwards and crossing into Asia through the Sinai. This group then branched in several directions, some moving into Europe and others heading east into Asia. This hypothesis attempts to explain why Haplogroup N is predominant in Europe and why Haplogroup M is absent in Europe. Evidence of the coastal migration is hypothesized to have been destroyed by the rise in sea levels during the Holocene epoch. Alternatively, a small European founder population that initially expressed both Haplogroup M and N could have lost Haplogroup M through random genetic drift resulting from a bottleneck (i.e. a founder effect).

Today at the Bab-el-Mandeb straits the Red Sea is about 12 miles (20 kilometres) wide, but 50,000 years ago it was much narrower and sea levels were 70 meters lower. Though the straits were never completely closed, there may have been islands in between which could be reached using simple rafts. Shell middens 125,000 years old have been found in Eritrea, indicating the diet of early humans included seafood obtained by beachcombing.

Subsequent expansion



Map of early human migrations

From the Near East, these populations spread east to South Asia by 50,000 years ago and on to Australia by 40,000 years ago, *Homo sapiens* for the first time colonizing territory never reached by *Homo erectus*. Europe was reached by Cro-Magnon some 40,000 years ago. East Asia (Korea, Japan) was reached by 30,000 years ago. It is disputed whether subsequent migration to North America took place around 30,000 years ago, or only considerably later, around 14,000 years ago.

The group that crossed the Red Sea travelled along the coastal route around the coast of Arabia and Persia until reaching India, which appears to be the first major settling point. M is found in high frequencies along the southern coastal regions of Pakistan and India and it has the greatest diversity in India, indicating that it is here where the mutation may have occurred. Sixty percent of the Indian population belong to Haplogroup M. The indigenous people of the Andaman Islands also belong to the M lineage. The Andamanese are thought to be offshoots of some of the earliest inhabitants in Asia because of their long isolation from mainland Asia. They are evidence of the coastal route of early settlers that extends from India along the coasts of Thailand and Indonesia all the way to Papua New Guinea. Since M is found in high frequencies in highlanders from New Guinea as well and both the Andamanese and New Guineans have dark skin and Afro-textured hair, some scientists believe they are all part of the same wave of migrants who departed across the Red Sea ~60,000 years ago in the Great Coastal Migration. Notably, the findings of Harding et al. (2000, p. 1355) show that, at least with regard to dark skin color, the haplotype background of Papua New Guineans at MC1R (one of a number of genes involved in melanin production) is identical to that of Africans (barring a single silent mutation). Thus, although these groups are distinct from Africans

at other loci (due to drift, bottlenecks, etc), it is evident that selection for the dark skin color trait likely continued (at least at MC1R) following the exodus. This would support the hypothesis that suggests that the original migrants from Africa resembled pre-exodus Africans (at least in skin color) and that the present day remnants of this ancient phenotype can be seen among contemporary Africans andamanese and New Guineans. Others suggest that their physical resemblance to Africans could be the result of convergent evolution.

From Arabia to India the proportion of haplogroup M increases eastwards: in eastern India, M outnumbers N by a ratio of 3:1. However, crossing over into East Asia, Haplogroup N reappears as the dominant lineage. M is predominant in South East Asia but amongst Indigenous Australians N reemerges as the more common lineage. This discontinuous distribution of Haplogroup N from Europe to Australia can be explained by founder effects and population bottlenecks.

Competing hypotheses

The multiregional hypothesis, initially proposed by Milford Wolpoff, holds that the evolution of humans from *H. erectus* at the beginning of the Pleistocene 1.8 million years BP to the present day has been within a single, continuous worldwide population. Proponents of multiregional origin reject the assumption of an infertility barrier between ancient Eurasian and African populations of *Homo*. Multiregional proponents point to the fossil record and genetic evidence in chromosomal DNA. One study suggested that at least 5% of the human modern gene pool can be attributed to ancient admixture, which in Europe would be from the Neanderthals.

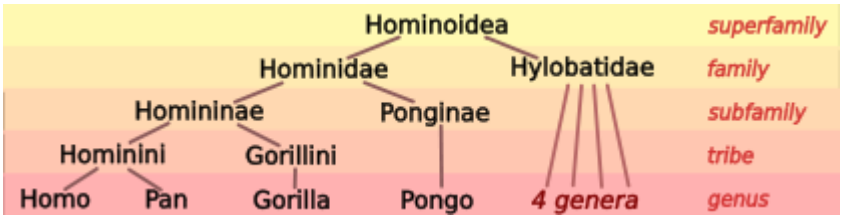
A recently discovered fossilized mandible that is putatively a hybrid between *Homo sapiens* and an earlier hominid, that is likely to be 110,000 years old, has been interpreted as a challenge to the recent out-of-Africa hypothesis. However, some scholars doubt that the fossil represents a Homo sapiens hybrid.

Chapter- 4

Human Evolutionary Genetics

Human evolutionary genetics studies how one human genome differs from the other, the evolutionary past that gave rise to it and its current effects. Differences between genomes have anthropological, medical and forensic implications and applications. Genetic data can provide important insight into human evolution.

Origin of apes

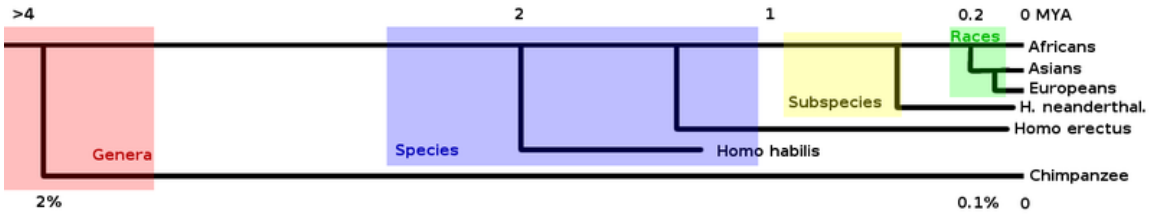


Taxonomic relationships of hominoids

Biologists classify humans, along with only a few other species, as great apes (species in the family Hominidae). The Hominidae include two distinct species of chimpanzee (the bonobo, *Pan paniscus* and the common chimpanzee, *Pan troglodytes*), two species of gorilla (the western gorilla, *Gorilla gorilla* and the eastern gorilla, *Gorilla graueri*) and two species of orangutan (the Bornean orangutan, *Pongo pygmaeus* and the Sumatran orangutan, *Pongo abelii*).

Apes, in turn, belong to the primates order (>400 species). Data from both mitochondrial DNA (mtDNA) and nuclear DNA (nDNA) indicates that primates belong to the group of Euarchontoglires, together with Rodentia, Lagomorpha, Dermoptera and Scandentia. This is further supported by Alu-like short interspersed nuclear elements (SINEs) which have been found only in members of the Euarchontoglires.

Cladistics



A phylogenetic tree like the one shown above is usually derived from DNA or protein sequences from populations. Often mitochondrial DNA or Y chromosome sequences are used to study ancient human demographics. These single-locus sources of DNA do not recombine and are almost always inherited from a single parent, with only one known exception in mtDNA (Schwartz and Vissing 2002). Individuals from the various continental groups tend to be more similar to one another than to people from other continents. The tree is rooted in the common ancestor of chimpanzees and humans, which is believed to have originated in Africa. Horizontal distance in the diagram corresponds to two things:

1. **Genetic distance**. Given below the diagram, the genetic difference between humans and chimps is less than 2%, or 20 times larger than the variation among modern humans.
2. **Temporal remoteness** of the most recent common ancestor. Rough estimates are given above the diagram, in millions of years. The mitochondrial most recent common ancestor of modern humans lived roughly 200,000 years ago, latest common ancestors of humans and chimps between four and seven million years ago.

Chimpanzees and humans belong to different genera, indicated in red. Formation of species and subspecies is also indicated and the formation of "races" is indicated in the green rectangle to the right (note that only a very rough representation of human phylogeny is given). Note that vertical distances are not meaningful in this representation.

Speciation of humans and the African apes

The separation of humans from their closest relatives, the African apes (chimpanzees and gorillas), has been studied extensively for more than a century. Five major questions have been addressed:

- Which apes are our closest ancestors?
- When did the separations occur?
- What was the effective population size of the common ancestor before the split?
- Are there traces of population structure (subpopulations) preceding the speciation or partial admixture succeeding it?
- What were the specific events (including fusion of chromosomes 2a and 2b) prior to and subsequent to the separation?

General observations

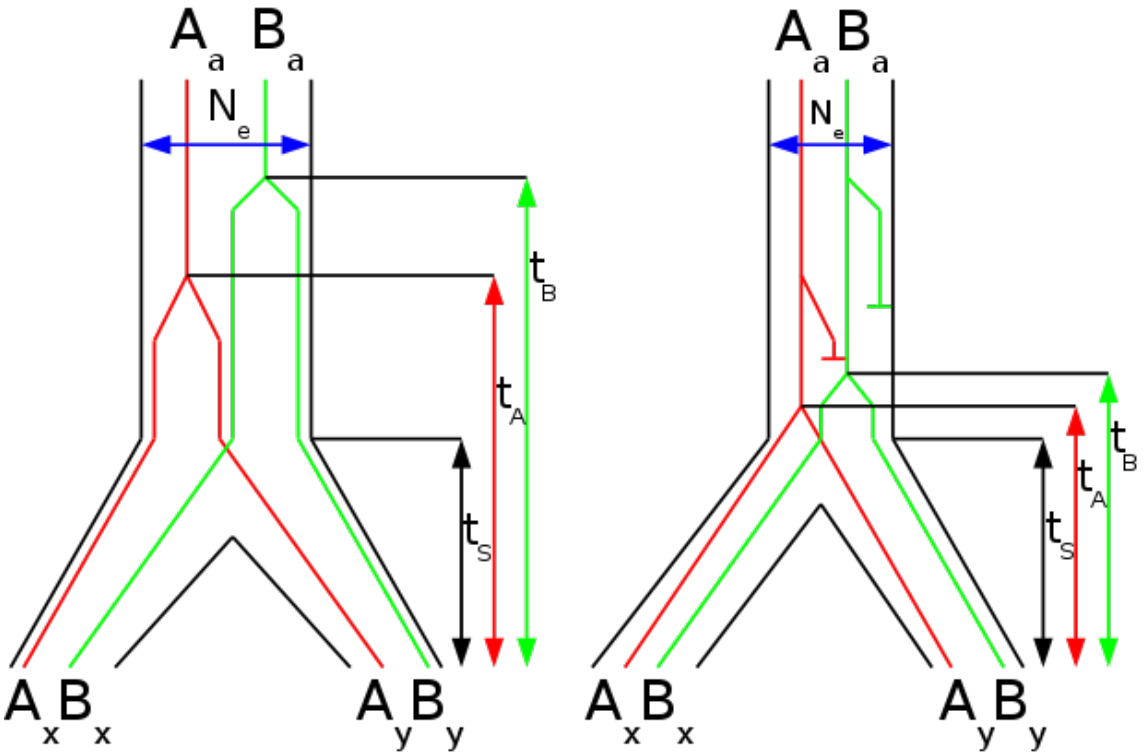
As discussed before, different parts of the genome show different sequence divergence between different hominoids. It has also been shown that the sequence divergence between DNA from humans and chimpanzees varies greatly. For example the sequence divergence varies between 0% to 2.66% between non-coding, non-repetitive genomic regions of humans and chimpanzees. Additionally gene trees, generated by comparative analysis of DNA segments, do not always fit the species tree. Summing up:

- The sequence divergence varies significantly between humans, chimpanzees and gorillas.
- For most DNA sequences, humans and chimpanzees appear to be most closely related, but some point to a human-gorilla or chimpanzee-gorilla clade.
- The human genome has been sequenced, as well as the chimpanzee genome. Humans have 23 pairs of chromosomes, while chimpanzees, gorillas and orangutans have 24. Human chromosome 2 is a fusion between two chromosomes that remained separate in the other primates.

Divergence times

The divergence time of humans from other apes is of great interest. One of the first molecular studies, published in 1967 measured immunological distances (IDs) between different primates. Basically the study measured the strength of immunological response that an antigen from one species (human albumin) induces in the immune system of another species (human, chimpanzee, gorilla and Old World monkeys). Closely related species should have similar antigens and therefore weaker immunological response to each other's antigens. The immunological response of a species to its own antigens (e.g. human to human) was set to be 1. The ID between humans and gorillas was determined to be 1.09, that between humans and chimpanzees was determined as 1.14. However the distance to six different Old World monkeys was on average 2.46 indicating that the African apes are far closer related to humans than to monkeys. The authors consider the divergence time between Old World monkeys and hominoids to be 30 million years ago (MYA), based on fossil data and the immunological distance was considered to grow at a constant rate. They concluded that divergence time of humans and the African apes to be roughly ~5 MYA. That was a surprising result. Most scientists at that time thought that humans and great apes diverged much earlier (>15 MYA). The gorilla was, in ID terms, closer to human than to chimpanzees, however the difference was so slight that the trichotomy could not be resolved with certainty. Later studies based on molecular genetics were able to resolve the trichotomy: chimpanzees are phylogenetically closer to humans than to gorillas. However, the divergence times estimated later (using much more sophisticated methods in molecular genetics) do not substantially differ from the very first estimate in 1967.

Divergence times and ancestral effective population size

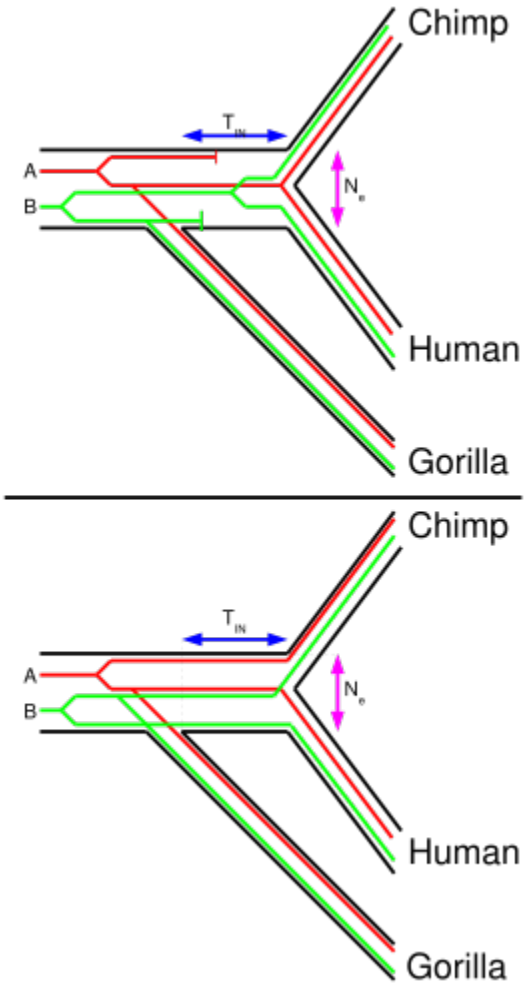


The sequences of the DNA segments diverge earlier than the species. A large effective population size in the ancestral population (left) preserves different variants of the DNA segments (=alleles) for a longer period of time. Therefore, on average, the gene divergence times (t_A for DNA segment A; t_B for DNA segment B) will deviate more from the time the species diverge (t_S) compared to a small ancestral effective population size (right).

Current methods to determine divergence times use DNA sequence alignments and molecular clocks. Usually the molecular clock is calibrated assuming that the orangutan split from the African apes (including humans) 12-16 MYA. Some studies also include some old world monkeys and set the divergence time of them from hominoids to 25-30 MYA. Both calibration points are based on very little fossil data and have been criticized. If these dates are revised, the divergence times estimated from molecular data will change as well. However, the relative divergence times are unlikely to change. Even if we can't tell absolute divergence times exactly, we can be pretty sure that the divergence time between chimpanzees and humans is about sixfold shorter than between chimpanzees (or humans) and monkeys.

One study (Takahata *et al.*, 1995) used 15 DNA sequence from different regions of the genome from human and chimpanzee and 7 DNA sequences from human, chimpanzee and gorilla. They determined that chimpanzees are more closely related to humans than gorillas. Using various statistical methods, they estimated the divergence time human-chimp to be 4.7 MYA and the divergence time between gorillas and humans (and chimps) to be 7.2 MYA. Additionally they estimated the effective population size of the

common ancestor of humans and chimpanzees to be ~100,000. This was somewhat surprising since the present day effective population size of humans is estimate to be only ~10,000. If true that means that the human lineage would have experienced an immense decrease of its effective population size (and thus genetic diversity) in its evolution.



A and B are two different loci. In the upper figure they fit to the species tree. The DNA that is present in today's gorillas diverged earlier from the DNA that is present in today's humans and chimps. Thus both loci should be more similar between human and chimp than between gorilla and chimp or gorilla and human. In the lower graph, locus A has a more recent common ancestor in human and gorilla compared to the chimp sequence. Whereas chimp and gorilla have a more recent common ancestor for locus B. Here the gene trees are incongruent to the species tree.

Another study (Chen & Li, 2001) sequenced 53 non-repetitive, intergenic DNA segments from a human, a chimpanzee, a gorilla and orangutan. When the DNA sequences were concatenated to a single long sequence, the generated neighbor-joining tree supported the *Homo-Pan* clade with 100% bootstrap (that is that humans and chimpanzees are the closest related species of the four). When three species are fairly closely related to each

other (like human, chimpanzee and gorilla), the trees obtained from DNA sequence data may not be congruent with the tree that represents the speciation (species tree). The shorter internodal time span (T_{IN}) the more common are incongruent gene trees. The effective population size (N_e) of the internodal population determines how long genetic lineages are preserved in the population. A higher effective population size causes more incongruent gene trees. Therefore, if the internodal time span is known, the ancestral effective population size of the common ancestor of humans and chimpanzees can be calculated.

When each segment was analyzed individually, 31 supported the *Homo-Pan* clade, 10 supported the *Homo-Gorilla* clade and 12 supported the *Pan-Gorilla* clade. Using the molecular clock the authors estimated that gorillas split up first 6.2-8.4 MYA and chimpanzees and humans split up 1.6-2.2 million years later (internodal time span) 4.6-6.2 MYA. The internodal time span is useful to estimate the ancestral effective population size of the common ancestor of humans and chimpanzees.

A parsimonious analysis revealed that 24 loci supported the *Homo-Pan* clade, 7 supported the *Homo-Gorilla* clade, 2 supported the *Pan-Gorilla* clade and 20 gave no resolution. Additionally they took 35 protein coding loci from databases. Of these 12 supported the *Homo-Pan* clade, 3 the *Homo-Gorilla* clade, 4 the *Pan-Gorilla* clade and 16 gave no resolution. Therefore only ~70% of the 52 loci that gave a resolution (33 intergenic, 19 protein coding) support the 'correct' species tree. From the fraction of loci which did not support the species tree and the internodal time span they estimated previously, the effective population of the common ancestor of humans and chimpanzees was estimated to be ~52 000 to 96 000. This value is not as high as that from the first study (Takahata), but still much higher than present day effective population size of humans.

A third study (Yang, 2002) used the same dataset that Chen and Li used but estimated the ancestral effective population of 'only' ~12,000 to 21,000, using a different statistical method.

Genetic differences between humans and other great apes

The alignable sequences within genomes of humans and chimpanzees differ by about 35 million single nucleotide substitutions. Additionally about 3% of the complete genomes differ by deletions, insertions and duplications.

Since mutation rate is relatively constant, roughly one half of these changes occurred in the human lineage. Only a very tiny fraction of those fixed differences gave rise to the different phenotypes of humans and chimpanzees and finding those is a great challenge. The vast majority of the differences are neutral and do not affect the phenotype.

Molecular evolution may act in different ways, through protein evolution, gene loss, differential gene regulation and RNA evolution. All are thought to have played some part in human evolution.

Gene loss

Many different mutations can inactivate a gene, but few will change its function in a specific way. Inactivation mutations will therefore be readily available for selection to act on. Gene loss could thus be a common mechanism of evolutionary adaptation (the "less-is-more" hypothesis).

80 genes were lost in the human lineage after separation from the last common ancestor with the chimpanzee. 36 of those were for olfactory receptors. Genes involved in chemoreception and immune response are overrepresented. Another study estimated that 86 genes had been lost.

Hair keratin gene KRTHAP1

A gene for type I hair keratin was lost in the human lineage. Keratins are a major component of hairs. Humans still have nine functional type I hair keratin genes but the loss of that particular gene may have caused the thinning of human body hair. The gene loss occurred relatively recently in human evolution—less than 240,000 years ago.

Myosin gene MYH16

Stedman *et al.* (2004) stated that the loss of the sarcomeric myosin gene MYH16 in the human lineage led to smaller masticatory muscles. They estimated that the mutation that led to the inactivation (a two base pair deletion) occurred 2.4 million years ago, predating the appearance of *Homo ergaster/erectus* in Africa. The period that followed was marked by a strong increase in cranial capacity, promoting speculation that the loss of the gene may have removed an evolutionary constraint on brain size in the genus *Homo*.

Another estimate for the loss of the MYH16 gene is 5.3 million years ago, long before *Homo* appeared.

Other

- CASPASE12, a cysteinyl aspartate proteinase

Gene addition

Segmental duplications (SDs or LCRs) have had roles in creating new primate genes and shaping human genetic variation.

Selection pressures

Human accelerated regions are areas of the genome that differ between humans and chimpanzees to a greater extent than can be explained by genetic drift over the time since the two species shared a common ancestor. These regions show signs of being subject to natural selection, leading to the evolution of distinctly human traits. Two examples are

HAR1F, which is believed to be related to brain development and HAR2 (a.k.a HACNS1) that may have played a role in the development of the opposable thumb.

Genetic differences between humans and Neanderthals

An international group of scientists has completed a draft of 63 percent of the Neanderthal genome, consisting of 3.7 billion base pairs. Some parts of the Neanderthal genome have more in common with chimps than humans, while other parts are shared by both Neanderthals and humans. Neanderthals and most humans share a lactose-intolerant variant of the lactase gene that encodes an enzyme that is unable to break down lactose in milk after weaning. Humans and Neanderthals also share the FOXP2 gene variant associated with brain development and with speech in humans, indicating that Neanderthals may have been able to speak. Chimps have two amino acid differences in FOXP2 compared with human FOXP2. In May 2010 the entire Neanderthal genome was completed and the results were published in the journal Nature. The results indicate some breeding between humans and Neanderthals as the genomes of non-African humans are 1-4% Neanderthal.

Sequence divergence between humans and apes

The draft sequence of the common chimpanzee genome published in the summer 2005 showed the regions that are similar enough to be aligned with one another account for 2400 million of the human genome's 3164.7 million bases – that is, 75.8% of the genome. This 75.8% of the human genome is 1.23% different from the chimpanzee genome in single nucleotide polymorphisms (changes of single DNA "letters" in the genome). Another type of difference, called indels (insertions/deletions) account for another ~3 % difference between the alignable sequences. In addition, variation in copy number of large segments (> 20 kb) of similar DNA sequence provides a further 2.7% difference between the two species. Hence the total similarity of the genomes could be as low as about 70%.

The figures above do not take into account differences in the organization of the alignable sequences within the genomes of humans and chimps. Short stretches of alignable sequence may be in very different orders and locations within the two genomes. At present we cannot fully assess the difference in structure of the two genomes, because the human genome was used as a scaffold when the chimpanzee draft genome was assembled. When genomes are sequenced, relatively short sequences of DNA are produced and these sequences have to be fitted together like a jigsaw puzzle. This requires multiple overlapping reads to accurately assemble the overall sequence. The human genome sequence is relatively accurate, with 8 to 9-fold coverage, but the chimpanzee draft genome only has 3.6-fold coverage. The human genome was sequenced using a hierarchical shotgun method which can deal with duplications and difficult-to-assemble sequences better than the whole genome shotgun method that was used for the chimpanzee draft genome. The human genome was used as a template for the assembly of the draft chimpanzee genome, on the assumption that the two genomes would be similar.

Almost half of that 1.23% SNP change belongs to the human at 0.53%, whose genetic variance is lower than a chimp and just over half to the chimp at 0.7%. If we also take into account that random "genetic drift" takes up the bulk of the 0.54% difference, then that percentage difference where SNPs have a potential positive impact on human abilities, is between 0.01% and 0.02%. The bonobo is a sibling species of common chimpanzee and is genetically about as different from humans as are common chimps.

Percentage sequence divergence between humans and other hominids

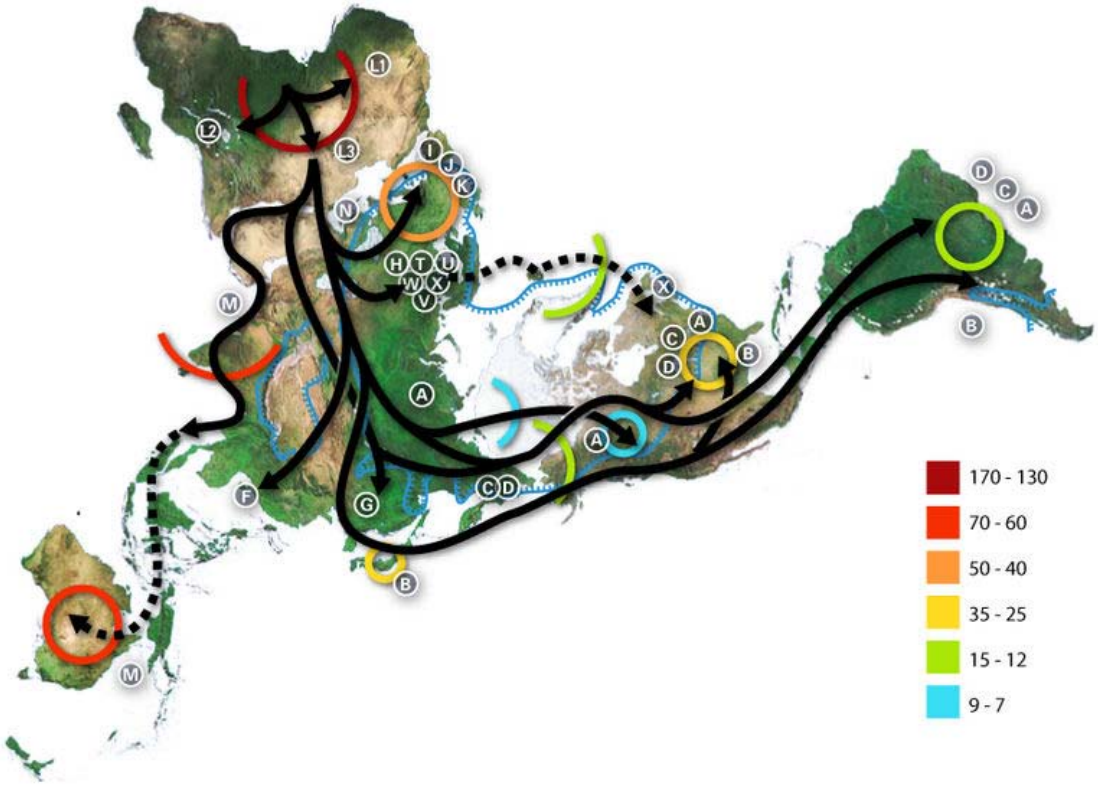
Locus	Human-Chimp	Human-Gorilla	Human-Orangutan
Alu elements	2	-	-
Non-coding (Chr. Y)	1.68 ± 0.19	2.33 ± 0.2	5.63 ± 0.35
Pseudogenes (autosomal)	1.64 ± 0.10	1.87 ± 0.11	-
Pseudogenes (Chr. X)	1.47 ± 0.17	-	-
Noncoding (autosomal)	1.24 ± 0.07	1.62 ± 0.08	3.08 ± 0.11
Genes (K_s)	1.11	1.48	2.98
Introns	0.93 ± 0.08	1.23 ± 0.09	-
Xq13.3	0.92 ± 0.10	1.42 ± 0.12	3.00 ± 0.18
Subtotal for X chromosome	1.16 ± 0.07	1.47 ± 0.08	-
Genes (K_a)	0.8	0.93	1.96

The sequence divergence has generally the following pattern: Human-Chimp < Human-Gorilla << Human-Orangutan, highlighting the close kinship between humans and the African apes. Alu elements diverge quickly due to their high frequency of CpG dinucleotides which mutate roughly 10 times more often than the average nucleotide in the genome. The mutation rate is higher in the male germ line, therefore the divergence in the Y chromosome—which is inherited solely from the father—is higher than in autosomes. The X chromosome is inherited twice as often through the female germ line as through the male germ line and therefore shows slightly lower sequence divergence. The sequence divergence of the Xq13.3 region is surprisingly low between humans and chimpanzees.

Mutations altering the amino acid sequence of proteins (K_a) are the least common. In fact ~29% of all orthologous proteins are identical between human and chimpanzee. The typical protein differs by only two amino acids.

The measures of sequence divergence shown in the table only take the substitutional differences, for example from an A (adenine) to a G (guanine), into account. DNA sequences may however also differ by insertions and deletions (indels) of bases. These are usually stripped from the alignments before the calculation of sequence divergence is performed. The overall sequence divergence between humans and chimpanzees for example is close to 5% if indels would be included.

Modern humans



Map of the migration of modern humans out of Africa, based on mitochondrial DNA. Coloured rings indicate years before present, in thousands.

Molecular biologists starting with Wesley Brown on mtDNA and Allan Wilson on mtDNA have produced observations relevant to human evolution.

Age of the common ancestor

By estimating the rate at which mutations occur in mtDNA, the age of the common ancestral mtDNA type can be estimated: "the common ancestral mtDNA (type a) links mtDNA types that have diverged by an average of nearly 0.57%. Assuming a rate of 2%-4% per million years, this implies that the common ancestor of all surviving mtDNA types existed 140,000-290,000 years ago." This observation is robust and this common direct female line ancestor (or mitochondrial most recent common ancestor (mtMRCA)) of all extant humans has become known as Mitochondrial Eve. The observation that the mtMRCA is the direct matrilineal ancestor of all living humans does not mean either that she was the first anatomically modern human, nor that no other female humans lived concurrently with her. Other women would have lived at the same time and passed nuclear genes down to living humans, but their mitochondrial lineages were lost over time. This could be due to random events such as producing only male children.

African origin for modern humans

There is evidence that modern human mtDNA has an African origin: "We infer from the tree of minimum length... that Africa is a likely source of the human mitochondrial gene pool. This inference comes from the observation that one of the two primary branches leads exclusively to African mtDNAs... while the second primary branch also leads to African mtDNAs... By postulating that the common ancestral mtDNA... was African, we minimize the number of intercontinental migrations needed to account for the geographic distribution of mtDNA types."

The broad study of African genetic diversity headed by Sarah Tishkoff found the San people to express the greatest genetic diversity among the 113 distinct populations sampled, making them one of 14 "ancestral population clusters". The research also located the origin of modern human migration in south-western Africa, near the coastal border of Namibia and Angola.

Y chromosome findings

The Y chromosome is much larger than mtDNA and is relatively homogeneous; therefore it has taken much longer to find distinct lineages and to analyse them. Conversely, because the Y chromosome is so large by comparison, it holds more genetic information. Y chromosome studies show similar findings to those made with mtDNA. The estimate for the age of the ancestral Y chromosome for all extant Y chromosomes is given at about 70,000 years ago and is also placed in Africa; the individual who contributed this Y chromosomal heritage is sometimes referred to as Y chromosome Adam. The difference in dates between Y chromosome Adam and mitochondrial Eve is usually attributed to a higher extinction rate for Y chromosomes due to greater differential reproductive success between individual men, which means that a small number of very successful men may produce many children, while a larger number of less successful men will produce far fewer children.

Chapter- 5

Human Genetic Variation

Human genetic variation is the genetic diversity of humans and represents the total amount of genetic characteristics observed within the human species. Genetic differences are observed between humans at both the individual and the population level. There may be multiple variants of any given gene in the human population (alleles), leading to polymorphism. Many genes are not polymorphic, meaning that only a single allele is present in the population: that allele is then said to be fixed.

No two humans are genetically identical. Even monozygotic twins, who develop from one zygote, have infrequent genetic differences due to mutations occurring during development and gene copy number variation has been observed. Differences between individuals, even closely related individuals, are the key to techniques such as genetic fingerprinting. Alleles occur at different frequencies in different human populations, with populations that are more geographically and ancestrally remote tending to differ more.

Causes of differences between individuals include the exchange of genes during meiosis and various mutational events. There are at least two reasons why genetic variation exists between populations. Natural selection may confer an adaptive advantage to individuals in a specific environment if an allele provides a competitive advantage. Alleles under selection are likely to occur only in those geographic regions where they confer an advantage. The second main cause of genetic variation is due to the high degree of neutrality of most mutations. Most mutations do not appear to have any selective effect one way or the other on the organism. The main cause is genetic drift, this is the effect of random changes in the gene pool. In humans, founder effect and past small population size (increasing the likelihood of genetic drift) may have had an important influence in neutral differences between populations.

The theory that humans recently migrated out of Africa is sometimes given as an example of this. It has been theorized that the population which migrated out of Africa only represented a small fraction of the genetic variation in Africa and that this is a contributing cause of the observed lower levels of diversity in all indigenous humans outside of Africa. Generally, more recent neutral polymorphisms caused by mutation are likely to be relatively geographically localized and rare, while older polymorphisms are more likely to be shared by a wider range of human groups. The large majority of observed genetic variation occurs within a population in any geographic region and not

between populations in different regions, although it is still usually possible to accurately identify the geographic origins of any individual's ancestors by genetic means.

The study of human genetic variation has both evolutionary significance and medical applications. The study can help scientists understand ancient human population migrations as well as how different human groups are biologically related to one another. From a medical perspective the study of human genetic variation may be important because some disease causing alleles occur at a greater frequency in people from specific geographic regions.

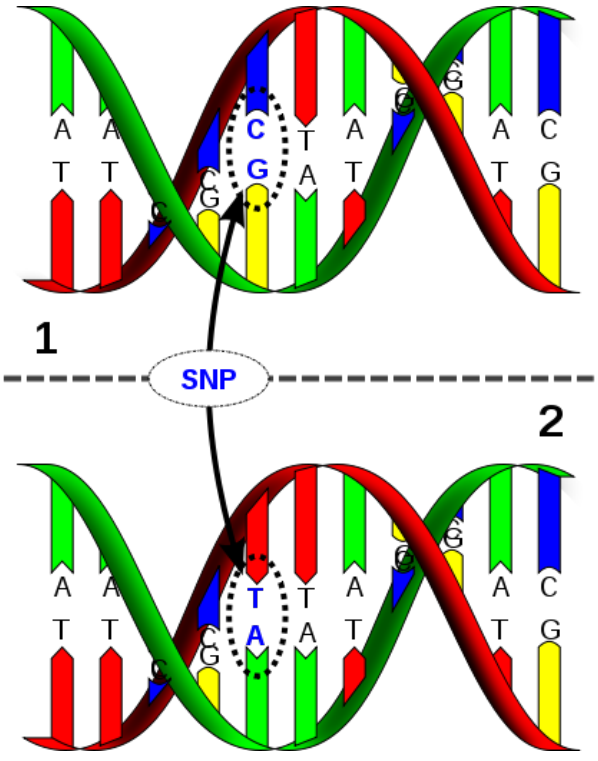
Genetic variation

Genetic variation, variation in alleles of genes, occurs both within and among populations. Genetic variation is important because it provides the “raw material” for natural selection.

Measures of variation

"Genetic variation among individual humans occurs on many different scales, ranging from gross alterations in the human karyotype to single nucleotide changes."

Single nucleotide polymorphisms



DNA molecule 1 differs from DNA molecule 2 at a single base-pair location (a C/T polymorphism).

Nucleotide diversity is based on single mutations called single nucleotide polymorphisms (SNPs). The nucleotide diversity between humans is about 0.1%, which is 1 difference per 1,000 base pairs. A difference of 1 in 1,000 nucleotides between two humans chosen at random amounts to approximately 3 million nucleotide differences since the human genome has about 3 billion nucleotides. Most of these SNPs are neutral but some are functional and influence phenotypic differences between humans through alleles. It is estimated that a total of 10 million SNPs exist in the human population of which at least 1% are functional.

Copy number variation

More recently a better understanding of the structure of the genome has been gained with the publication of two examples of full sequences of an individual's genome. This represents a new development because the Human Genome Project and a parallel project by Celera Genomics produced two haploid sequences, both of which were an amalgamation of sequences from many individuals. Recently the diploid sequences of both Craig Venter and James Watson have been published. Analysis of diploid sequences has shown that non-SNP variation accounts for much more human genetic variation than single nucleotide diversity. This non-SNP variation includes copy number variation and results from deletions, inversions, insertions and duplications. It is estimated that approximately 0.4% of the genomes of unrelated people typically differ with respect to copy number. When copy number variation is included, human to human genetic variation is estimated to be at least 0.5% (99.5% similarity). Copy number variations are inherited but can also arise during development.

Epigenetics

Epigenetics is another type of genetic variation. "This type of variation arises from chemical tags that attach to DNA and affect how it gets read. The chemical tags, called epigenetic markings, act as switches that control how genes can be read." At some alleles, the epigenetic state of the DNA and associated phenotype, can be inherited transgenerationally.

Genetic variability

Genetic variability is a measure of the tendency of individual genotypes in a population to vary (become different) from one another. Variability is different from genetic diversity, which is the amount of variation seen in a particular population. The variability of a trait describes how much that trait tends to vary in response to environmental and genetic influences.

Clines

In biology, a cline is a term used to describe a continuum of species, populations, races, varieties, or forms of organisms that exhibit gradual phenotypic and/or genetic differences over a geographical area, typically as a result of environmental heterogeneity.

In the scientific study of human genetic variation, a gene cline can be rigorously defined and subjected to quantitative metrics.

Haplogroups

In the study of molecular evolution, a haplogroup is a group of similar haplotypes that share a common ancestor with a single nucleotide polymorphism (SNP) mutation. Haplogroups pertain to deep ancestral origins dating back thousands of years.

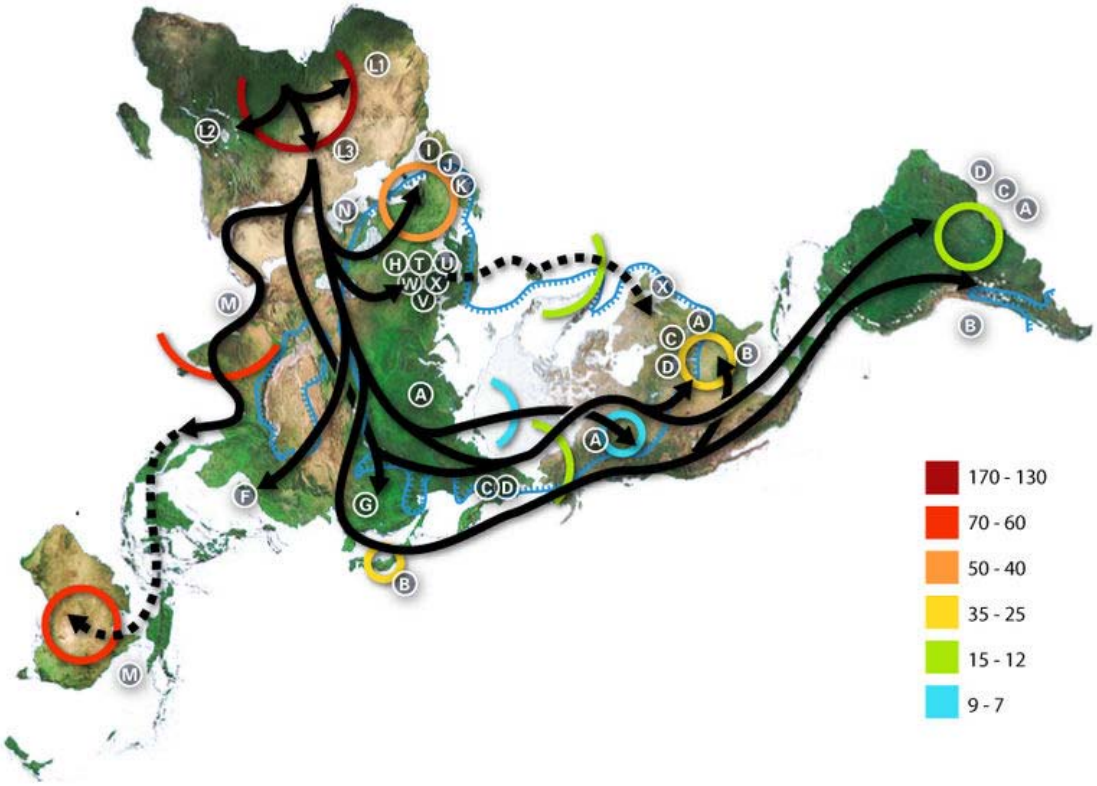
In human genetics, the haplogroups most commonly studied are Y-chromosome (Y-DNA) haplogroups and mitochondrial DNA (mtDNA) haplogroups, both of which can be used to define genetic populations. Y-DNA is passed solely along the patrilineal line, from father to son, while mtDNA is passed down the matrilineal line, from mother to both daughter and son. The Y-DNA and mtDNA may change by chance mutation at each generation.

Variable number tandem repeats

A variable number tandem repeat (VNTR) is a location in a genome where a short nucleotide sequence is organized as a tandem repeat. These can be found on many chromosomes and often show variations in length between individuals. Each variant acts as an inherited allele, allowing them to be used for personal or parental identification. Their analysis is useful in genetics and biology research, forensics and DNA fingerprinting.

There are two principal families of VNTRs: microsatellites and minisatellites. The former are repeats of sequences less than about 5 base pairs in length, while the latter involve longer blocks.

History and geographic distribution



Map of the migration of modern humans out of Africa, based on mitochondrial DNA. Colored rings indicate thousand years before present.

A 10-year study published in April 2009 analyzed the patterns of variation at 1,327 DNA markers of 121 African populations, 4 African American populations and 60 non-African populations. The research showed that there is more human genetic diversity in Africa than anywhere else on Earth. The genetic structure of Africans was traced to 14 ancestral population clusters and the ancestral origin of humans was determined to probably be located in southern Africa, near the border of Namibia and South Africa.

Human genetic diversity decreases in native populations with migratory distance from Africa and this is thought to be the result of bottlenecks during human migration, which are events that temporarily reduce population size. It has been shown that variations in skull measurements decrease with distance from Africa at the same rate as the decrease in genetic diversity. These data support the Out of Africa theory over the multiregional origin of modern humans hypothesis. The aforementioned April 2009 study identifies the likely origin of modern human migration as being in southwestern Africa, near the coastal border of Namibia and Angola and the exit point out of Africa as being in East Africa.

The *recent African origin of modern humans* is the mainstream model describing the origin and early dispersal of anatomically modern humans, *Homo sapiens sapiens*. The

theory is known popularly as the *(Recent) Out-of-Africa* model. The hypothesis originated in the 19th century, with Darwin's *Descent of Man*, but remained speculative until the 1980s when it was corroborated based on a study of present-day mitochondrial DNA, combined with evidence based on physical anthropology of archaic specimens.

According to both genetic and fossil evidence, archaic Homo sapiens evolved to anatomically modern humans solely in Africa, between 200,000 and 100,000 years ago, with members of one branch leaving Africa by 60,000 years ago and over time replacing earlier human populations such as Neanderthals and *Homo erectus*. According to this theory, around the above time frame, one of the African subpopulations went through a process of speciation prohibiting gene flow between African and Eurasian Human populations.

Population genetics

In the field of population genetics, it is believed that the distribution of neutral polymorphisms among contemporary humans reflects human demographic history. It is believed that humans passed through a population bottleneck before a rapid expansion coinciding with migrations out of Africa leading to an African-Eurasian divergence around 100,000 years ago (ca. 5,000 generations), followed by a European-Asian divergence about 40,000 years ago (ca. 2,000 generations). Richard G. Klein, Nicholas Wade and Spencer Wells, among others, have postulated that modern humans did not leave Africa and successfully colonize the rest of the world until as recently as 60,000 - 50,000 years B.P., pushing back the dates for subsequent population splits as well.

The rapid expansion of a previously small population has two important effects on the distribution of genetic variation. First, the so-called founder effect occurs when founder populations bring only a subset of the genetic variation from their ancestral population. Second, as founders become more geographically separated, the probability that two individuals from different founder populations will mate becomes smaller. The effect of this assortative mating is to reduce gene flow between geographical groups and to increase the genetic distance between groups. The expansion of humans from Africa affected the distribution of genetic variation in two other ways. First, smaller (founder) populations experience greater genetic drift because of increased fluctuations in neutral polymorphisms. Second, new polymorphisms that arose in one group were less likely to be transmitted to other groups as gene flow was restricted.

Our history as a species also has left genetic signals in regional populations. For example, in addition to having higher levels of genetic diversity, populations in Africa tend to have lower amounts of linkage disequilibrium than do populations outside Africa, partly because of the larger size of human populations in Africa over the course of human history and partly because the number of modern humans who left Africa to colonize the rest of the world appears to have been relatively low (Gabriel *et al.* 2002). In contrast, populations that have undergone dramatic size reductions or rapid expansions in the past and populations formed by the mixture of previously separate ancestral groups can have unusually high levels of linkage disequilibrium (Nordborg and Tavare 2002).

Many other geographic, climatic and historical factors have contributed to the patterns of human genetic variation seen in the world today. For example, population processes associated with colonization, periods of geographic isolation, socially reinforced endogamy and natural selection all have affected allele frequencies in certain populations (Jorde *et al.* 2000b; Bamshad and Wooding 2003). In general, however, the recency of our common ancestry and continual gene flow among human groups have limited genetic differentiation in our species.

Distribution of variation

The distribution of genetic variants within and among human populations are impossible to describe succinctly because of the difficulty of defining a "population," the clinal nature of variation and heterogeneity across the genome (Long and Kittles 2003). In general, however, an average of 85% of genetic variation exists within local populations, ~7% is between local populations within the same continent and ~8% of variation occurs between large groups living on different continents,. (Lewontin 1972; Jorde *et al.* 2000a; Hinds *et al.* 2005). The recent African origin theory for humans would predict that in Africa there exists a great deal more diversity than elsewhere and that diversity should decrease the further from Africa a population is sampled. Long and Kittles show that indeed, African populations contain about 100% of human genetic diversity, whereas in populations outside of Africa diversity is much reduced, for example in their population from New Guinea only about 70% of human variation is captured.

Phenotypic variation

Sub-Saharan Africa has the most human genetic diversity and the same has been shown to hold true for phenotypic diversity. Genetic diversity decreases smoothly with migratory distance from that region, which many scientists believe to be the origin of modern humans and that decrease is mirrored by a decrease in phenotypic variation. Skull measurements are an example of a physical attribute whose within-population variation decreases with distance from Africa.

The distribution of many physical traits resembles the distribution of genetic variation within and between human populations (American Association of Physical Anthropologists 1996; Keita and Kittles 1997). For example, ~90% of the variation in human head shapes occurs within continental groups and ~10% separates groups, with a greater variability of head shape among individuals with recent African ancestors (Relethford 2002).

A prominent exception to the common distribution of physical characteristics within and among groups is skin color. Approximately 10% of the variance in skin color occurs within groups and ~90% occurs between groups (Relethford 2002). This distribution of skin color and its geographic patterning — with people whose ancestors lived predominantly near the equator having darker skin than those with ancestors who lived predominantly in higher latitudes — indicate that this attribute has been under strong selective pressure. Darker skin appears to be strongly selected for in equatorial regions to

prevent sunburn, skin cancer, the photolysis of folate and damage to sweat glands (Sturm *et al.* 2001; Rees 2003).

Neanderthal admixture

Interbreeding of Neanderthals and anatomically modern humans during the Middle Paleolithic is a hypothesis. In May 2010, the Neanderthal Genome Project presented genetic evidence that interbreeding did likely take place and that a small but significant portion of Neanderthal admixture is present in the DNA of modern non-African populations.

Categorization of the world population

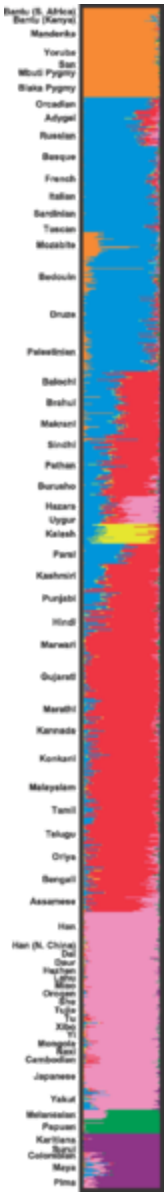


Chart showing human genetic clustering

New data on human genetic variation has reignited the debate surrounding race. Most of the controversy surrounds the question of how to interpret this new data and whether conclusions based on existing data are sound. A large majority of researchers endorse the view that continental groups do not constitute different subspecies. However, other researchers still debate whether evolutionary lineages should rightly be called "races". These questions are particularly pressing for ancestry related health issues, where self-identified race is often used as an indicator of ancestry.

Although the genetic differences among human groups are relatively small, these differences in certain genes such as duffy, ABCC11, SLC24A5, called ancestry-informative markers (AIMs) nevertheless can be used to reliably situate many individuals within broad, geographically based groupings or self-identified race. For example, computer analyses of hundreds of polymorphic loci sampled in globally distributed populations have revealed the existence of genetic clustering that roughly is associated with groups that historically have occupied large continental and subcontinental regions (Rosenberg *et al.* 2002; Bamshad *et al.* 2003).

Some commentators have argued that these patterns of variation provide a biological justification for the use of traditional racial categories. They argue that the continental clusterings correspond roughly with the division of human beings into sub-Saharan Africans; Europeans, Western Asians, Central Asians, Southern Asians and Northern Africans; Eastern Asians, Southeast Asians, Polynesians and Native Americans; and other inhabitants of Oceania (Melanesians, Micronesians & Australian Aborigines) (Risch *et al.* 2002). Other observers disagree, saying that the same data undercut traditional notions of racial groups (King and Motulsky 2002; Calafell 2003; Tishkoff and Kidd 2004). They point out, for example, that major populations considered races or subgroups within races do not necessarily form their own clusters.

Furthermore, because human genetic variation is clinal, many individuals affiliate with two or more continental groups. Thus, the genetically based "biogeographical ancestry" assigned to any given person generally will be broadly distributed and will be accompanied by sizable uncertainties (Pfaff *et al.* 2004).

In many parts of the world, groups have mixed in such a way that many individuals have relatively recent ancestors from widely separated regions. Although genetic analyses of large numbers of loci can produce estimates of the percentage of a person's ancestors coming from various continental populations (Shriver *et al.* 2003; Bamshad *et al.* 2004), these estimates may assume a false distinctiveness of the parental populations, since human groups have exchanged mates from local to continental scales throughout history (Cavalli-Sforza *et al.* 1994; Hoerder 2002). Even with large numbers of markers, information for estimating admixture proportions of individuals or groups is limited and estimates typically will have wide confidence intervals (Pfaff *et al.* 2004).

Lewontin's Fallacy

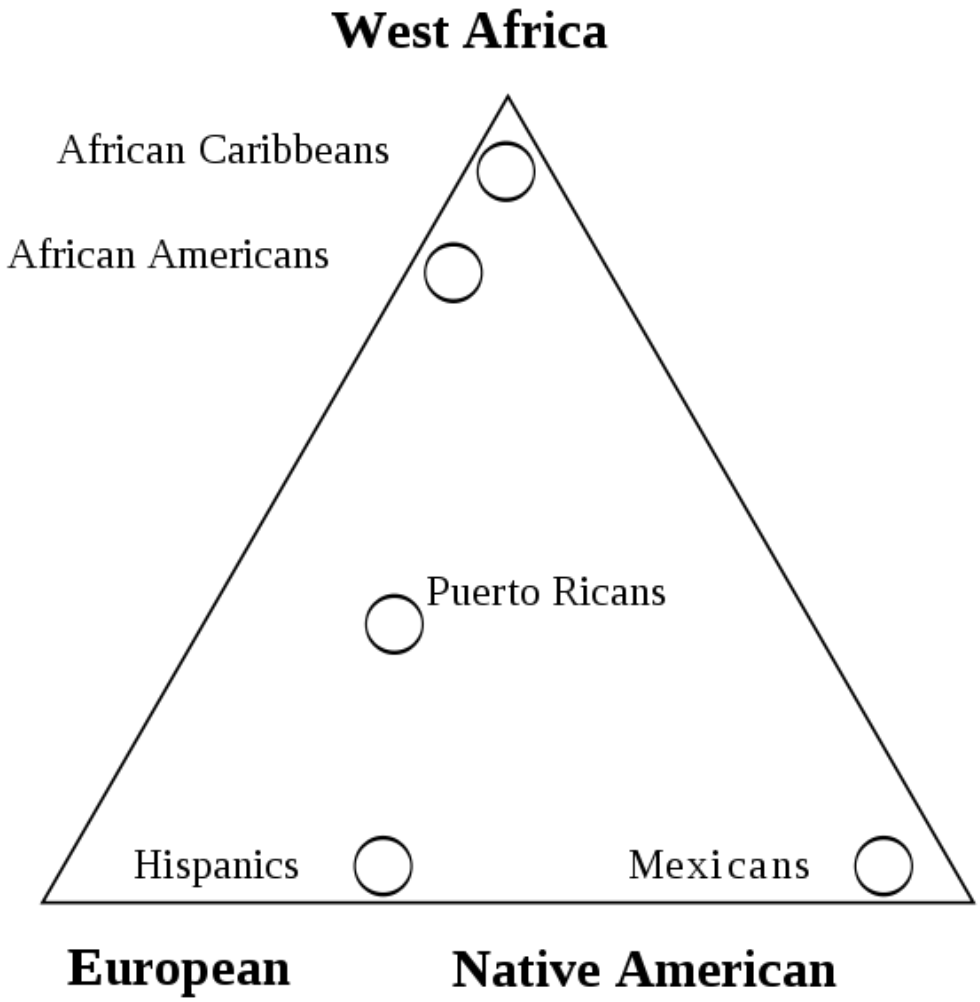
In 2003 A. W. F. Edwards wrote a paper called Lewontin's Fallacy, rebutting the argument that because most genetic variation is within-group, classification of humans is not possible. He claimed that this conclusion ignores the fact that most of the information that distinguishes populations is hidden in the correlation structure of the data and not simply in the variation of the individual factors. Edwards concludes that "It is not true that 'racial classification is ... of virtually no genetic or taxonomic significance' or that 'you can't predict someone's race by their genes'." Undeterred, in an article titled "Confusions About Human Races" published in 2006, Lewontin maintains that race is no more than a social construct.

Genetic clustering

Genetic data can be used to infer population structure and assign individuals to groups that often correspond with their self-identified geographical ancestry. Recently, Lynn Jorde and Steven Wooding argued that "Analysis of many loci now yields reasonably accurate estimates of genetic similarity among individuals, rather than populations. Clustering of individuals is correlated with geographic origin or ancestry."

Forensic anthropology

Forensic anthropologists can determine race (e.g. Asian, African, or European ancestry) from skeletal remains with a high degree of accuracy by conducting bone analysis. Studies have shown that individual test methods such as midfacial measurements and femur traits can be over 80 percent accurate and in combination can achieve very high levels of accuracy. The skeletons of mixed-race individuals can, however, exhibit characteristics of more than one racial group. Despite the success of this method with the remains of individuals with ancestry predominantly from a single race, anthropologists, including George W. Gill and C. Loring Brace, disagree on whether race is a valid biological concept.



Triangle plot shows average admixture of five North American ethnic groups. Individuals that self-identify with each group can be found at many locations on the map, but on average groups tend to cluster differently.

Admixture

Miscegenation between two populations reduces the average genetic distance between the populations. During the Age of Discovery which began in the early 15th century, European explorers sailed all around the globe, reaching all the major continents. In the process they came into contact with many populations that had been isolated for thousands of years. It is generally accepted that the Tasmanian aboriginals were the most isolated group on the planet. They were driven to extinction by European explorers, however a number of their descendants survive today as a result of admixture with Europeans. This is an example of how modern migrations have begun to reduce the genetic divergence of the human race.

The demographic composition of the old world has not changed significantly since the age of discovery. However new world demographics were radically changed within a short time following the voyage of Columbus. The colonization of the Americas brought Native Americans into contact with the distant populations of Europe, Africa and Asia. As a result many countries in the Americas have significant and complex multiracial populations. Furthermore many who identify themselves by only one race still have multiracial ancestry.

Health

Differences in allele frequencies contribute to group differences in the incidence of some monogenic diseases and they may contribute to differences in the incidence of some common diseases (Risch *et al.* 2002; Burchard *et al.* 2003; Tate and Goldstein 2004). For the monogenic diseases, the frequency of causative alleles usually correlates best with ancestry, whether familial (for example, Ellis-van Creveld syndrome among the Pennsylvania Amish), ethnic (Tay-Sachs disease among Ashkenazi Jewish populations), or geographical (hemoglobinopathies among people with ancestors who lived in malarial regions). To the extent that ancestry corresponds with racial or ethnic groups or subgroups, the incidence of monogenic diseases can differ between groups categorized by race or ethnicity and health-care professionals typically take these patterns into account in making diagnoses.

Even with common diseases involving numerous genetic variants and environmental factors, investigators point to evidence suggesting the involvement of differentially distributed alleles with small to moderate effects. Frequently cited examples include hypertension (Douglas *et al.* 1996), diabetes (Gower *et al.* 2003), obesity (Fernandez *et al.* 2003) and prostate cancer (Platz *et al.* 2000). However, in none of these cases has allelic variation in a susceptibility gene been shown to account for a significant fraction of the difference in disease prevalence among groups and the role of genetic factors in generating these differences remains uncertain (Mountain and Risch 2004).

Neil Risch of Stanford University has proposed that self-identified race/ethnic group could be a valid means of categorization in the USA for public health and policy considerations. While a 2002 paper by Noah Rosenberg's group makes a similar claim "The structure of human populations is relevant in various epidemiological contexts. As a result of variation in frequencies of both genetic and nongenetic risk factors, rates of disease and of such phenotypes as adverse drug response vary across populations. Further, information about a patient’s population of origin might provide health care practitioners with information about risk when direct causes of disease are unknown."

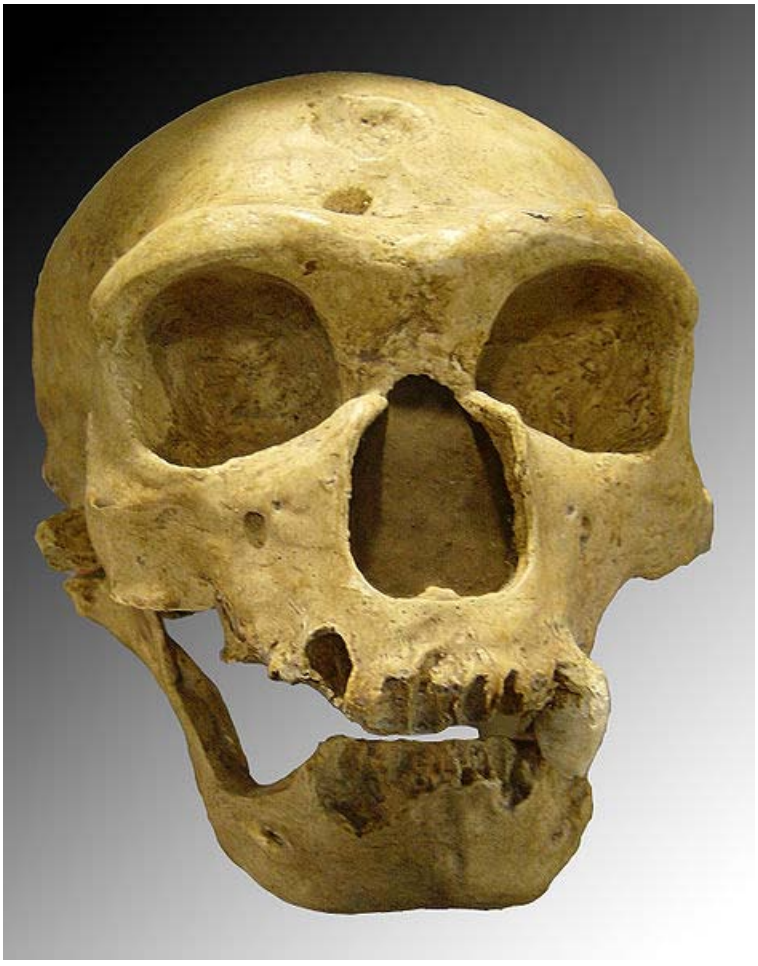
Genome projects

Human genome projects are scientific endeavors that determine or study the structure of the human genome. The Human Genome Project was a landmark genome project.

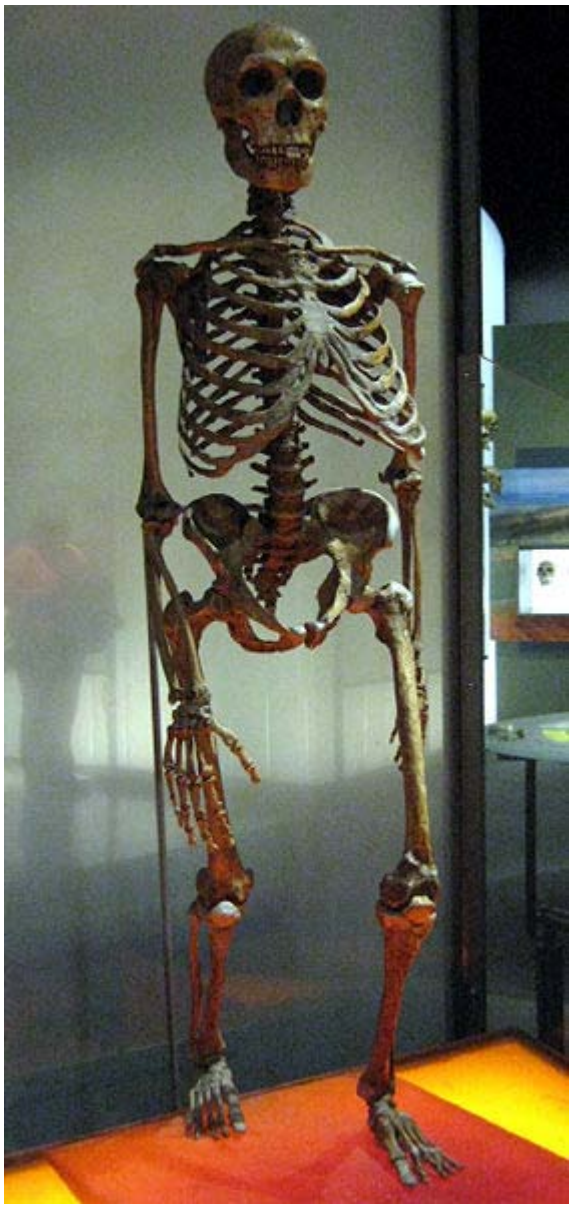
Chapter- 6

Neanderthal

Neanderthal
Fossil range: Middle to Late Pleistocene0.6–0.03 Ma



A Skull, La Chapelle-aux-Saints



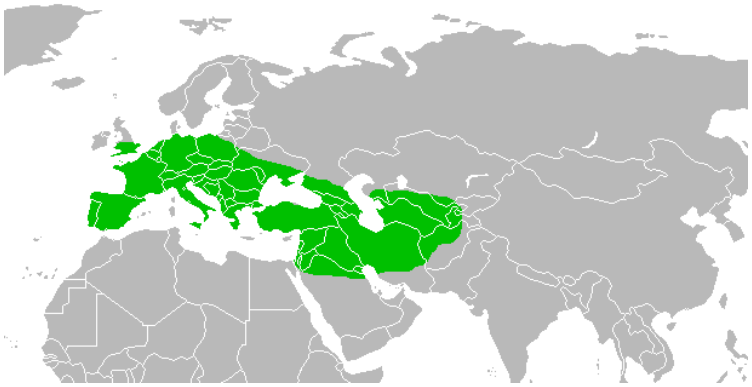
Mounted Neanderthal skeleton, American Museum of Natural History

Scientific classification	
Kingdom:	Animalia
Phylum:	Chordata
Class:	Mammalia
Order:	Primates
Family:	Hominidae
Genus:	*Homo*
Species:	***H. neanderthalensis***

Binomial name

Homo neanderthalensis

King, 1864



Range of *Homo neanderthalensis*. Eastern and northern ranges may be extended to include Okladnikov in Altai and Mamotnaia in Ural

Synonyms

Palaeoanthropus neanderthalensis
H. s. neanderthalensis

The **Neanderthal** (short for **Neanderthal Man**; in modern orthography **Neandertal**) is an extinct member of the *Homo* genus that is known from Pleistocene specimens found in Europe and parts of western and central Asia. Neanderthals are either classified as a subspecies (or race) of modern humans (***Homo sapiens neanderthalensis***) or as a separate human species (***Homo neanderthalensis***).

The first proto-Neanderthal traits appeared in Europe as early as 600,000–350,000 years ago. Proto-Neanderthal traits are occasionally grouped to another phenetic 'species', *Homo heidelbergensis*, or a migrant form, *Homo rhodesiensis*.

By 130,000 years ago, complete Neanderthal characteristics had appeared. These characteristics then disappeared in Asia by 50,000 years ago and in Europe by about 30,000 years ago, with no further individuals having enough Neanderthal morphological traits to be considered as part of *Homo neanderthalensis*.

Current (as of 2010) genetic evidence suggests interbreeding took place with *Homo sapiens* (anatomically modern humans) between roughly 80,000 and 50,000 years ago in the Middle East, resulting in 1–4% of the genome of people from Eurasia having been contributed by Neanderthals.

The youngest Neanderthal finds include Hyaena Den (UK), considered older than 30,000 years ago, while the Vindija (Croatia) Neanderthals have been re-dated to between 32,000 and 33,000 years ago. No definite specimens younger than 30,000 years ago have been found; however, evidence of fire by Neanderthals at Gibraltar indicate they may have survived there until 24,000 years ago. Cro-Magnon or early modern human skeletal remains with 'Neanderthal traits' were found in Lagar Velho (Portugal), dated to 24,500

years ago and controversially interpreted as indications of extensively admixed populations.

Neanderthal stone tools provide further evidence for their presence where skeletal remains have not been found. The last traces of Mousterian culture, a type of stone tools associated with Neanderthals, were found in Gorham's Cave on the remote south-facing coast of Gibraltar. Other tool cultures sometimes associated with Neanderthal include Châtelperronian, Aurignacian and Gravettian, with the latter extending to 22,000 years ago, the last indication of Neanderthal presence.

Neanderthal cranial capacity is thought to have been as large as that of *Homo sapiens*, perhaps larger, indicating their brain size may have been comparable, as well. In 2008, a group of scientists created a study using three-dimensional computer-assisted reconstructions of Neanderthal infants based on fossils found in Russia and Syria, showing that they had brains as large as modern humans' at birth and larger than modern humans' as adults. On average, the height of Neanderthals was comparable to contemporaneous *Homo sapiens*. Neanderthal males stood about 165–168 cm (65–66 in) and were heavily built with robust bone structure. They were much stronger than *Homo sapiens*, having particularly strong arms and hands. Females stood about 152–156 cm (60–61 in).

In 2010 a U.S. researcher reported finding cooked plant matter in the teeth of a Neanderthal skull, indicating the earlier belief they were exclusively (or almost exclusively) carnivorous and apex predators was incorrect.

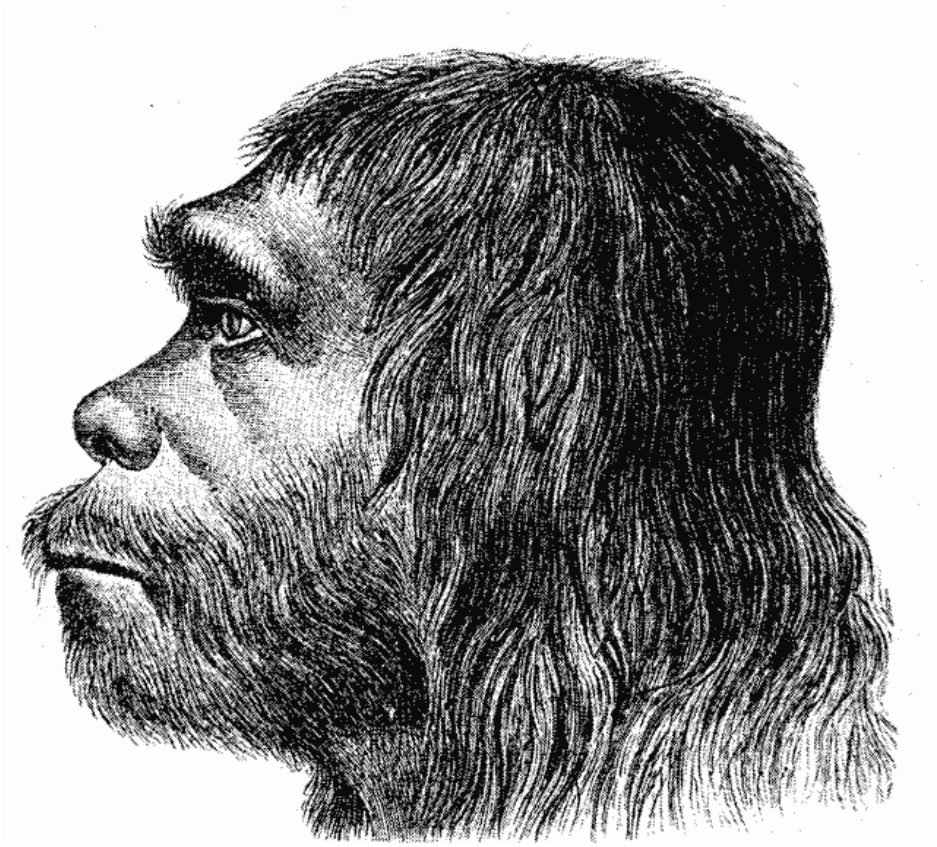
Etymology

The Neanderthal is named after the Neandertal valley, formerly spelled *Neanderthal*, which is located about 12 km (7.5 mi) east of Düsseldorf, Germany. The valley itself was named after the theologian Joachim Neander, who lived nearby in Düsseldorf in the late 17th century. "Neander" is a classicized form of the common German surname Neumann. "Tal" (spelled "Thal" until the German spelling reform of 1901) is the German word for valley. The fossil discovered in the Neandertal in 1856, Neanderthal 1, was known as the "Neanderthal skull" or "Neanderthal cranium" in anthropological literature and the individual reconstructed on the basis of the skull was occasionally called the "Neanderthal man". The binomial name *Homo neanderthalensis*, extending the name "Neanderthal man" from the individual type specimen to the entire species, is due to the Anglo-Irish geologist William King (1864). The practice of referring to the members of the species simply as "the Neanderthals", singular "a Neanderthal", emerges in popular literature of the 1920s.

The spelling of the German word *Thal* ("dale, valley"), was changed to *Tal* in 1901 and the spelling of the valley was also changed accordingly to *Neandertal*. The former spelling is, however, often retained in English for the hominid. The spelling with *th* is in addition always used in scientific names throughout the world. In German, however, the modern spelling with *t* is used in referring to both the hominid and the valley. *Neandertal*

is a widespread alternative spelling in English, becoming so common it is sometimes now listed first in dictionaries, for example MSN Encarta.

Classification



First reconstruction of a Neanderthal male

For some time, scientists have debated whether Neanderthals should be classified as *Homo neanderthalensis* or "*Homo sapiens neanderthalensis*", the latter placing Neanderthals as a subspecies of *Homo sapiens*. Some morphological studies support that *Homo neanderthalensis* is a separate species and not a subspecies. Others, for example University of Cambridge Professor Paul Mellars, say "no evidence has been found of cultural interaction" and evidence from mitochondrial DNA studies have been interpreted as evidence Neanderthals were not a subspecies of *H. sapiens*, though more recent genomic evidence showed otherwise.

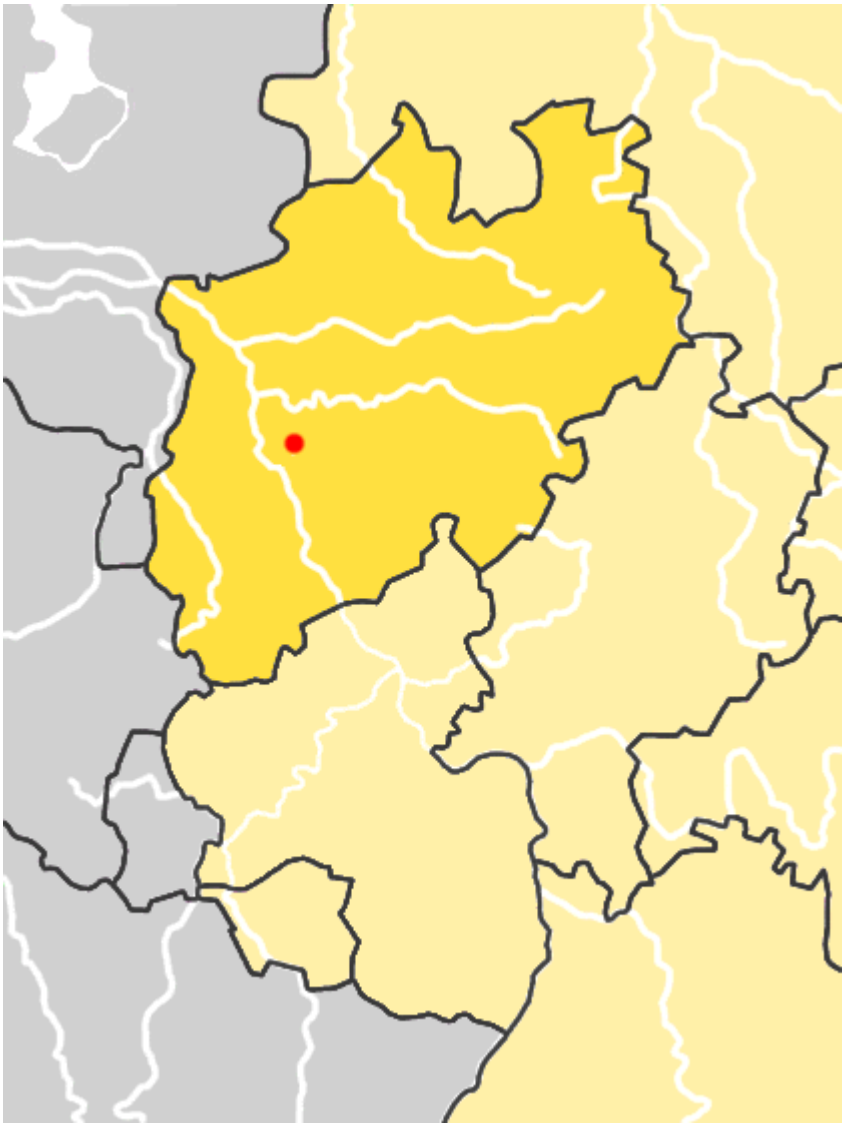
Neanderthals evolved from early *Homo* along a path similar to *Homo sapiens*, both deriving from a chimp-like ancestor between five and 10 million years ago. Like *H. sapiens*, Neanderthals are related to *Australopithecus*, *Homo habilis* and *Homo ergaster*; the exact descent remains uncertain. The last common ancestor between anatomically modern *Homo sapiens* and Neanderthals appears to be *Homo rhodesiensis*, named after an archaic *Homo sapiens* fossil, Broken hill 1 (Kabwe 1) discovered in the territory of Rhodesia in 1921.

Homo rhodesiensis arose in Africa an estimated 0.7 to 1 million years ago. The earliest estimates for *Homo rhodesiensis* reaching Europe are approximately 800 thousand years ago when a type of human referred to as *Homo antecessor* or *Homo cepranensis* already inhabited the region. These two human types may be forerunners to European *Homo heidelbergensis*; however, stone tools dating from 1.2 to 1.56 million years ago of an unknown creator have been discovered in south-western Europe. The evidence at the Sima de los Huesos (in the Atapuerca cave system on the Iberian Peninsula) suggests *Homo heidelbergensis* was already in Europe by 600,000 years ago.

Molecular phylogenetic analysis suggests *Homo rhodesiensis* and *Homo heidelbergensis* continued to intermix until 350,000 years ago, after which they were separate species and sometime within the last 200,000 years *Homo heidelbergensis* evolved into *Homo neanderthalensis*, the classic Neanderthal human. It appears the original Neanderthal population was, in fact, more distantly related to today's human than is *Homo heidelbergensis*. However, recent evidence of successful interbreeding between Neanderthals and modern humans has made that issue moot, at least insofar as some Neanderthal populations were concerned.

Discovery

The site of Kleine Feldhofer Grotte where the first Neanderthal was unearthed by miners in the 19th century



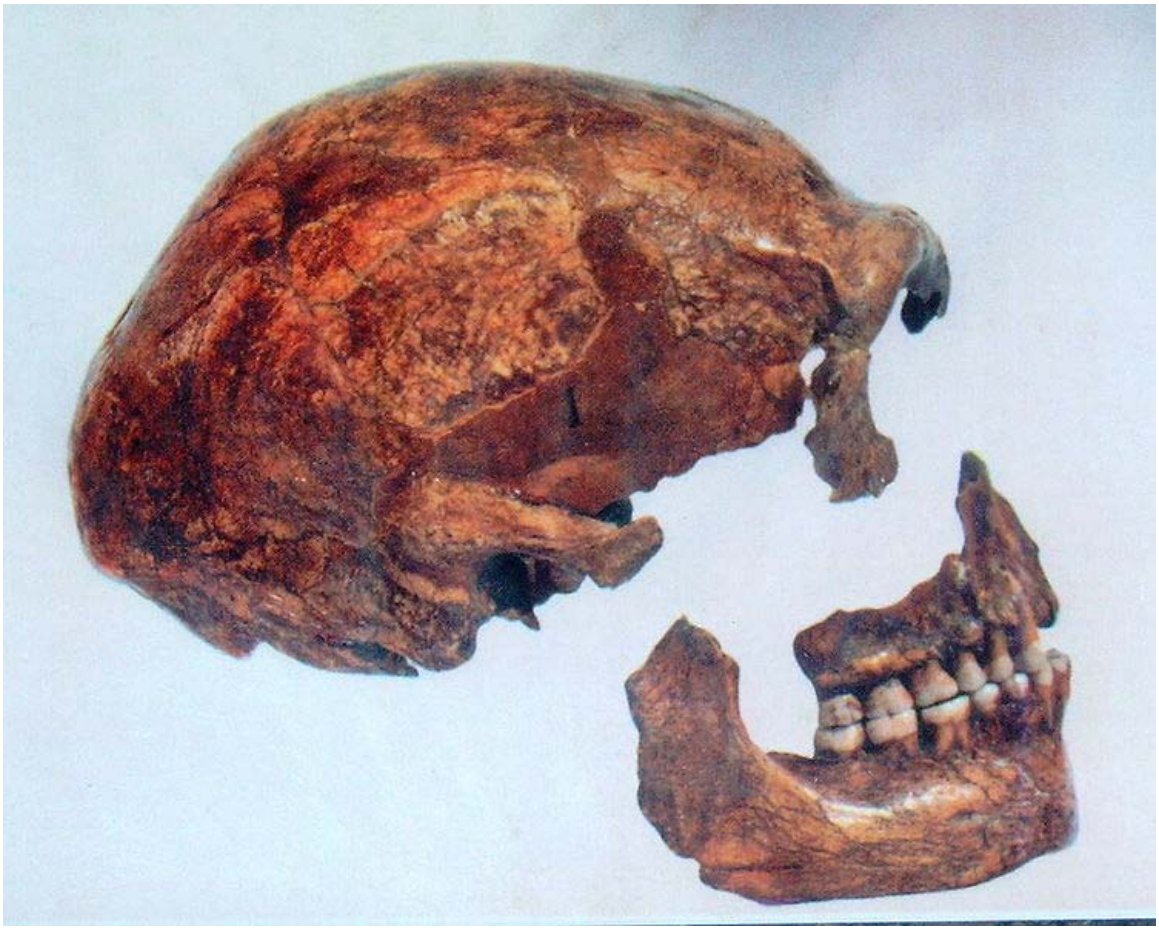
Location of Neander Valley, Germany. (The highlighted area is the modern federal state of North Rhine-Westphalia.)

Neanderthal skulls were first discovered in Engis, Belgium (1829) by Philippe-Charles Schmerling and in Forbes' Quarry, Gibraltar (1848), both prior to the type specimen discovery in a limestone quarry of the Neander Valley in Erkrath near Düsseldorf in August 1856, three years before Charles Darwin's *On the Origin of Species* was published.

The type specimen, dubbed Neanderthal 1, consisted of a skull cap, two femora, three bones from the right arm, two from the left arm, part of the left ilium, fragments of a scapula and ribs. The workers who recovered this material originally thought it to be the remains of a bear. They gave the material to amateur naturalist Johann Carl Fuhlrott, who turned the fossils over to anatomist Hermann Schaaffhausen. The discovery was jointly announced in 1857.

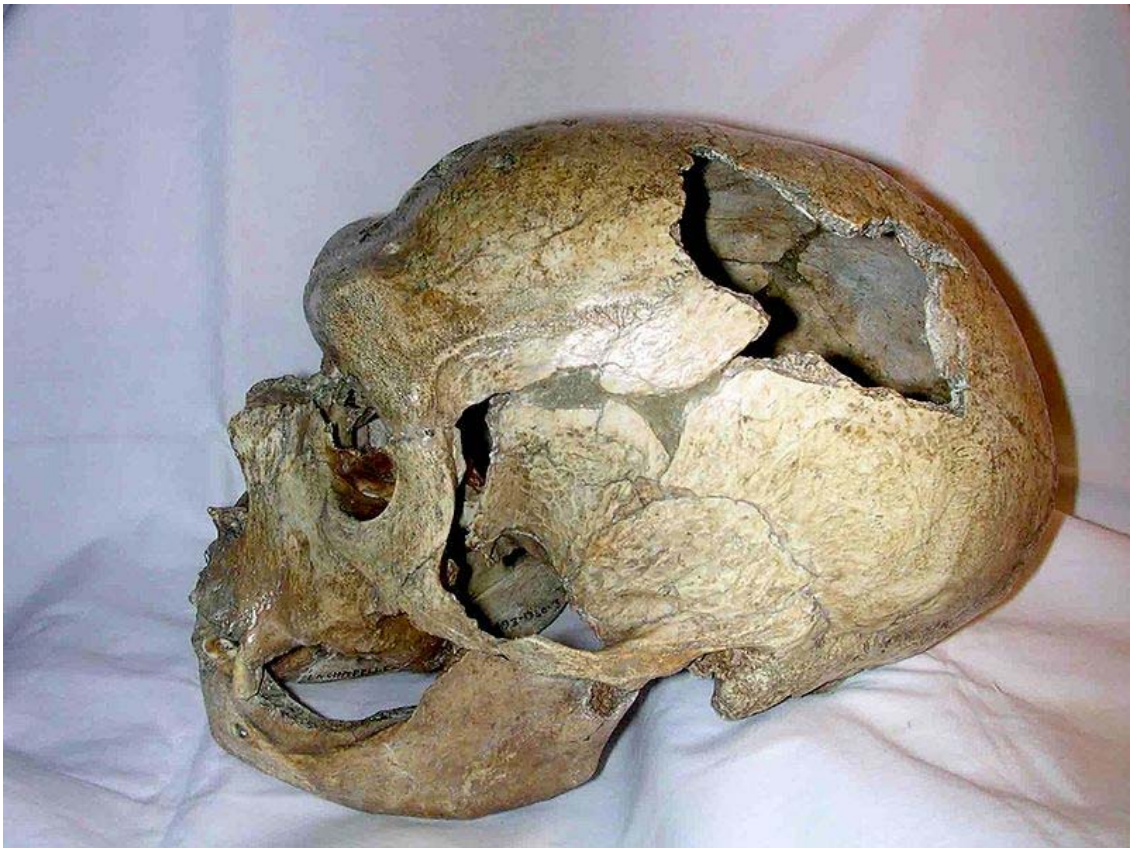
The original Neanderthal discovery is now considered the beginning of paleoanthropology. These and other discoveries led to the idea these remains were from ancient Europeans who had played an important role in modern human origins. The bones of over 400 Neanderthals have been found since.

Timeline



Skull, found in 1886 in Spy, Belgium

Frontal bone of a neanderthal child from the cave of La Garigüela



Skull from La Chapelle aux Saints

- 1829: Neanderthal skulls were discovered in Engis, Belgium.
- 1848: Neanderthal skull found in Forbes' Quarry, Gibraltar. Called "an ancient human" at the time.
- 1856: Johann Karl Fuhlrott first recognized the fossil called "Neanderthal man", discovered in Neanderthal, a valley near Mettmann in what is now North Rhine-Westphalia, Germany.
- 1880: The mandible of a Neanderthal child was found in a secure context and associated with cultural debris, including hearths, Mousterian tools and bones of extinct animals.
- 1886: Two nearly perfect skeletons of a man and woman were found at Spy, Belgium at the depth of 16 ft with numerous Mousterian-type implements.
- 1899: Hundreds of Neanderthal bones were described in stratigraphic position in association with cultural remains and extinct animal bones.
- 1908: A nearly complete Neanderthal skeleton was discovered in association with Mousterian tools and bones of extinct animals.
- 1925: Francis Turville-Petre finds the 'Galilee Man' or 'Galilee Skull' in the Zuttiyeh Cave in Wadi Amud in Palestine (now Israel).
- 1953–1957: Ralph Solecki uncovered nine Neanderthal skeletons in Shanidar Cave in northern Iraq.
- 1975: Erik Trinkaus's study of Neanderthal feet confirmed they walked like modern humans.
- 1987: Thermoluminescence results from Israeli fossils date Neanderthals at Kebara to 60,000 BP and humans at Qafzeh to 90,000 BP. These dates were confirmed by electron spin resonance (ESR) dates for Qafzeh (90,000 BP) and Es Skhul (80,000 BP).
- 1991: ESR dates showed the Tabun Neanderthal was contemporaneous with modern humans from Skhul and Qafzeh.
- 1997: Matthias Krings *et al.* are the first to amplify Neanderthal mitochondrial DNA (mtDNA) using a specimen from Feldhofer grotto in the Neander valley.
- 2000: Igor Ovchinnikov, Kirsten Liden, William Goodman *et al.* retrieved DNA from a Late Neanderthal (29,000 BP) infant from Mezmaikaya Cave in the Caucasus.
- 2005: The Max Planck Institute for Evolutionary Anthropology launched a project to reconstruct the Neanderthal genome.
- 2006: The Max Planck Institute for Evolutionary Anthropology announced it planned to work with Connecticut-based 454 Life Sciences to reconstruct the Neanderthal genome.
- 2009: The Max Planck Institute for Evolutionary Anthropology announced the "first draft" of a complete Neanderthal genome is completed.
- 2010: Comparison of Neanderthal genome with modern humans from Africa and Eurasia shows 1–4% of modern non-African human genetic material is identical with Neanderthal DNA.
- 2010: Discovery of Neanderthal tools far away from the influence of homo sapiens indicate that the species might have been able to create and evolve tools on its own and therefore be more intelligent than previously thought. Furthermore, it was proposed that the Neanderthals might be more closely related

to homo sapiens that previously thought and that may in fact be a sub species of it.

Habitat and range

Sites where typical Neanderthal fossils have been found

Early Neanderthals lived in the Last Glacial age for a span of about 100,000 years. Because of the damaging effects the glacial period had on the Neanderthal sites, not much is known about the early species. Countries where their remains are known include most of Europe south of the line of glaciation, roughly along the 50th parallel north, including most of Western Europe, including the south coast of Great Britain, Central Europe and the Balkans, some sites in the Ukraine and in western Russia and outside of Europe in the Zagros Mountains and in the Levant.

Neanderthal fossils have not been found to date in Africa, but there have been finds rather close to Africa, both at Gibraltar and in the Levant. At some Levantine sites, Neanderthal remains, in fact, date after the same sites were vacated by *Homo sapiens*. Mammal fossils of the same time period show cold-adapted animals were present alongside these Neanderthals in this region of the Eastern Mediterranean. This implies Neanderthals were better adapted biologically to cold weather than *H. sapiens* and at times displaced *H. sapiens* in parts of the Middle East when the climate got cold enough. *Homo sapiens* appears to have been the only human type in the Nile River Valley during these periods and Neanderthals are not known to have ever lived south-west of modern Israel. When further climate change caused warmer temperatures, the Neanderthal range likewise retreated to the north along with the cold-adapted species of mammals. Apparently these

weather-induced population shifts took place before "modern" people secured competitive advantages over the Neanderthal, as these shifts in range took place well over ten thousand years before "moderns" totally replaced the Neanderthal, despite the recent evidence of some successful interbreeding.

There were separate developments in the human line, in other regions such as Southern Africa, that somewhat resembled the European and Western/Central Asian Neanderthals, but these people were not actually Neanderthals. One such example is Rhodesian Man (*Homo rhodesiensis*) who existed long before any classic European Neanderthals, but had a more modern set of teeth and arguably some *H. rhodesiensis* populations were on the road to modern *Homo sapiens sapiens*.

To date, no intimate connection has been found between these similar people and the Western/Central Eurasian Neanderthals, at least during the same time as classic Eurasian Neanderthals and *H. rhodesiensis* seems to have evolved separately and earlier than classic Neanderthals in a case of convergent evolution.

It appears incorrect, based on present research and known fossil finds, to refer to any fossil outside Europe or Western and Central Asia as a true Neanderthal. True Neanderthals had a known range that possibly extended as far east as the Altai Mountains, but not farther to the east or south and apparently not into Africa. At any rate, in Africa the land immediately south of the Neanderthal range was possessed by "modern" *H. sap.*, since at least 160,000 years before the present.

Classic Neanderthal fossils have been found over a large area, from northern Germany to Israel and Mediterranean countries like Spain and Italy in the south and from England and Portugal in the west to Uzbekistan in the east. This area probably was not occupied all at the same time. The northern border of their range, in particular, would have contracted frequently with the onset of cold periods. On the other hand, the northern border of their range as represented by fossils may not be the real northern border of the area they occupied, since Middle Palaeolithic-looking artifacts have been found even further north, up to 60° N, on the Russian plain. Recent evidence has extended the Neanderthal range by about 1,250 miles (2,010 km) east into southern Siberia's Altai Mountains.

Anatomy

Neanderthal anatomy was more robust than anatomically modern humans.

Behavior

Neanderthals were almost exclusively carnivorous and apex predators, however new studies do indicate that they had cooked vegetables in their diet. They made advanced tools, had a language (the nature of which is debated) and lived in complex social groups.

Genome

Early investigations concentrated on mitochondrial DNA (mtDNA), which, owing to strictly matrilineal inheritance and subsequent vulnerability to genetic drift, is of limited value in evaluating the possibility of interbreeding of Neanderthals with Cro-Magnon people.

In 1997, geneticists were able to extract a short sequence of DNA from Neanderthal bones from 30,000 years ago.

In July 2006, the Max Planck Institute for Evolutionary Anthropology and 454 Life Sciences announced that they would sequence the Neanderthal genome over the next two years. This genome is expected to be roughly the size of the human genome, three-billion base pairs and share most of its genes. It was hoped the comparison would expand understanding of Neanderthals, as well as the evolution of humans and human brains.

Svante Pääbo has tested more than 70 Neanderthal specimens. Preliminary DNA sequencing from a 38,000-year-old bone fragment of a femur found at Vindija Cave, Croatia, in 1980 showed *Homo neanderthalensis* and *Homo sapiens* share about 99.5% of their DNA. From mtDNA analysis estimates, the two species shared a common ancestor about 500,000 years ago. An article appearing in the journal *Nature* has calculated the species diverged about 516,000 years ago, whereas fossil records show a time of about 400,000 years ago. A 2007 study pushes the point of divergence back to around 800,000 years ago.

Edward Rubin of the Lawrence Berkeley National Laboratory in Berkeley, California, states recent genome testing of Neanderthals suggests human and Neanderthal DNA are some 99.5% to nearly 99.9% identical.

On 16 November 2006, Lawrence Berkeley National Laboratory issued a press release suggesting Neanderthals and ancient humans probably did not interbreed. Edward M. Rubin, director of the U.S. Department of Energy's Lawrence Berkeley National Laboratory and the Joint Genome Institute (JGI), sequenced a fraction (0.00002) of genomic nuclear DNA (nDNA) from a 38,000-year-old Vindia Neanderthal femur. They calculated the common ancestor to be about 353,000 years ago and a complete separation of the ancestors of the species about 188,000 years ago. Their results show the genomes of modern humans and Neanderthals are at least 99.5% identical, but despite this genetic similarity and despite the two species having coexisted in the same geographic region for thousands of years, Rubin and his team did not find any evidence of any significant crossbreeding between the two. Rubin said, "While unable to definitively conclude that interbreeding between the two species of humans did not occur, analysis of the nuclear DNA from the Neanderthal suggests the low likelihood of it having occurred at any appreciable level."

In 2008 Richard E. Green et al. from Max Planck Institute for Evolutionary Anthropology in Munich, Germany published the full sequence of Neanderthal

mitochondrial DNA (mtDNA) and suggested "Neanderthals had a long-term effective population size smaller than that of modern humans." Writing in *Nature* about Green et al.'s findings, James Morgan asserted the mtDNA sequence contained clues that Neanderthals lived in "small and isolated populations and probably did not interbreed with their human neighbours."

In the same publication, it was disclosed by Svante Pääbo that in the previous work at the Max Planck Institute that "Contamination was indeed an issue," and they eventually realized that 11% of their sample was modern human DNA. Since then, more of the preparation work has been done in clean areas and 4-base pair 'tags' have been added to the DNA as soon as it is extracted so the Neanderthal DNA can be identified.

With 3 billion nucleotides sequenced, analysis of about 1/3rd showed no sign of admixture between modern humans and Neanderthals, according to Pääbo. This concurred with the work of Noonan from two years earlier. The variant of microcephalin common outside Africa, which was suggested to be of Neanderthal origin and responsible for rapid brain growth in humans, was not found in Neanderthals. Nor was the MAPT variant, a very old variant found primarily in Europeans.

However, an analysis of a first draft of the Neanderthal genome by the same team released in May 2010 indicates interbreeding may have occurred. "Those of us who live outside Africa carry a little Neanderthal DNA in us," said Pääbo, who led the study. "The proportion of Neanderthal-inherited genetic material is about 1 to 4 percent. It is a small but very real proportion of ancestry in non-Africans today," says Dr. David Reich of Harvard Medical School in Boston, who worked on the study. This research compared the genome of the Neanderthals to five modern humans from China, France, sub-Saharan Africa and Papua New Guinea. The finding is that about 1 to 4 percent of the genes of the non-Africans came from Neanderthals, compared to the baseline defined by the two Africans. This indicates a gene flow from Neanderthals to modern humans, i.e., interbreeding between the two populations. Since the three non-African genomes show a similar proportion of Neanderthal sequences, the interbreeding must have occurred early in the migration of modern humans out of Africa, perhaps in the Middle East. No evidence for gene flow in the direction from modern humans to Neanderthals was found. The latter result would not be unexpected if contact occurred between a small colonizing population of modern humans and a much larger resident population of Neanderthals. A very limited amount of interbreeding could explain the findings, if it occurred early enough in the colonization process.

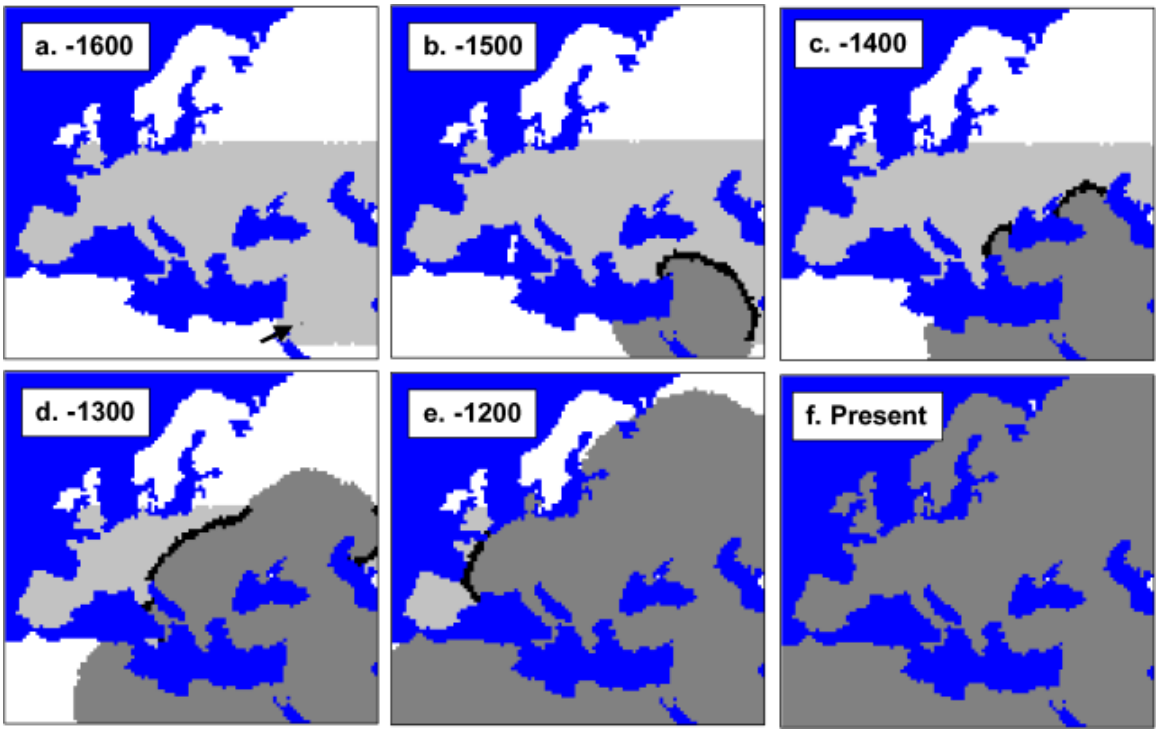
While interbreeding is viewed as the most parsimonious interpretation of the genetic discoveries, the authors point out they cannot conclusively rule out an alternative scenario, in which the source population of non-African modern humans was already more closely related to Neanderthals than other Africans were, due to ancient genetic divisions within Africa.

Among the genes shown to differ between present-day humans and Neanderthals were *RPTN*, *SPAG17*, *CAN15*, *TTF1* and *PCD16*.

Extinction hypotheses

The Neanderthals disappear from the fossil record after about 25,000 years ago. The last traces of Mousterian culture (without human specimens) have been found in Gorham's Cave on the remote south-facing coast of Gibraltar, dated 30,000 to 24,500 years ago. Possible scenarios are:

1. Neanderthals were a separate species from modern humans and became extinct (due to climate change or interaction with humans) and were replaced by *H. sapiens* moving into its habitat beginning around 80 kya. Competition from *H. sapiens* probably contributed to Neanderthal extinction. Jared Diamond has suggested a scenario of violent conflict and displacement.
2. Neanderthals were a contemporary subspecies that bred with *Homo sapiens* and disappeared through absorption (interbreeding hypothesis).



mtDNA-based simulation of modern human expansion in Europe starting 1600 generations ago. Neanderthal range in light grey.

As Jordan notes: "A natural sympathy for the underdog and the disadvantaged lends a sad poignancy to the fate of the Neanderthal folk, however it came about." Jordan, though, does say that there was perhaps interbreeding to some extent, but that populations that remained totally Neanderthal were likely out-competed and marginalized to extinction by the Aurignacians.

Climate change

About 55,000 years ago, the weather began to fluctuate wildly from extreme cold conditions to mild cold and back in a matter of a few decades. Neanderthal bodies were well suited for survival in a cold climate—their barrel chests and stocky limbs stored body heat better than the Cro-Magnons. However, the rapid fluctuations of weather caused ecological changes to which the Neanderthals could not adapt. The weather changes were so rapid that within a lifetime, plants and animals someone grew up with would be replaced by completely different plants and animals. Neanderthal's ambush techniques would have failed as grasslands replaced trees. A large number of Neanderthals would have died during these fluctuations, which peaked about 30,000 years ago.

Studies on Neanderthal body structures have shown that they needed more energy to survive than any other species. Their energy needs were up to 100-350 calories more per day comparing to projected anatomically modern human males weighing 68.5 kg and females 59.2 kg. When food became scarce, this difference may have played a major role in the Neanderthals' extinction.

Coexistence with H. sapiens

There is no longer certainty regarding the identity of the humans who produced the Aurignacian culture, even though the presumed westward spread of anatomically modern humans (AMHs) across Europe is still based on the controversial first dates of the Aurignacian. Currently, the oldest European anatomically modern *Homo sapiens* is represented by a robust modern-human mandible discovered at Peştera cu Oase (southwest Romania), dated to 34–36 thousand years ago. Human skeletal remains from the German site of Vogelherd, so far regarded as the best association between anatomically modern *Homo sapiens* and Aurignacian culture, were revealed to represent intrusive Neolithic burials into the Aurignacian levels and subsequently all the key Vogelherd fossils are now dated to 3.9–5.0 thousand years ago instead. As for now, the expansion of the first anatomically modern humans into Europe cannot be located by diagnostic and well-dated AMH fossils "west of the Iron Gates of the Danube" before 32 thousand years ago.

Consequently, the exact nature of biological and cultural interaction between Neanderthals and other human groups between 50 and 30 thousand years ago is currently hotly contested. A new proposal strives to resolve the issue by proposing the Gravettians rather than the Aurignacians as the anatomically modern humans who contributed to the Eurasian genetic pool after 30 thousand years ago. Correspondingly, the human skull fragment found at the Elbe River bank at Hahnöfersand near Hamburg was once radiocarbon-dated to 36,000 years ago and seen as possible evidence for the intermixing of Neanderthals and anatomically modern humans. It is now dated to the more recent Mesolithic.

Interbreeding hypotheses

An alternative to extinction is that Neanderthals were absorbed into the Cro-Magnon population by interbreeding. This would be counter to strict versions of the Recent African Origin, since it would imply that at least part of the genome of Europeans would descend from Neanderthals.

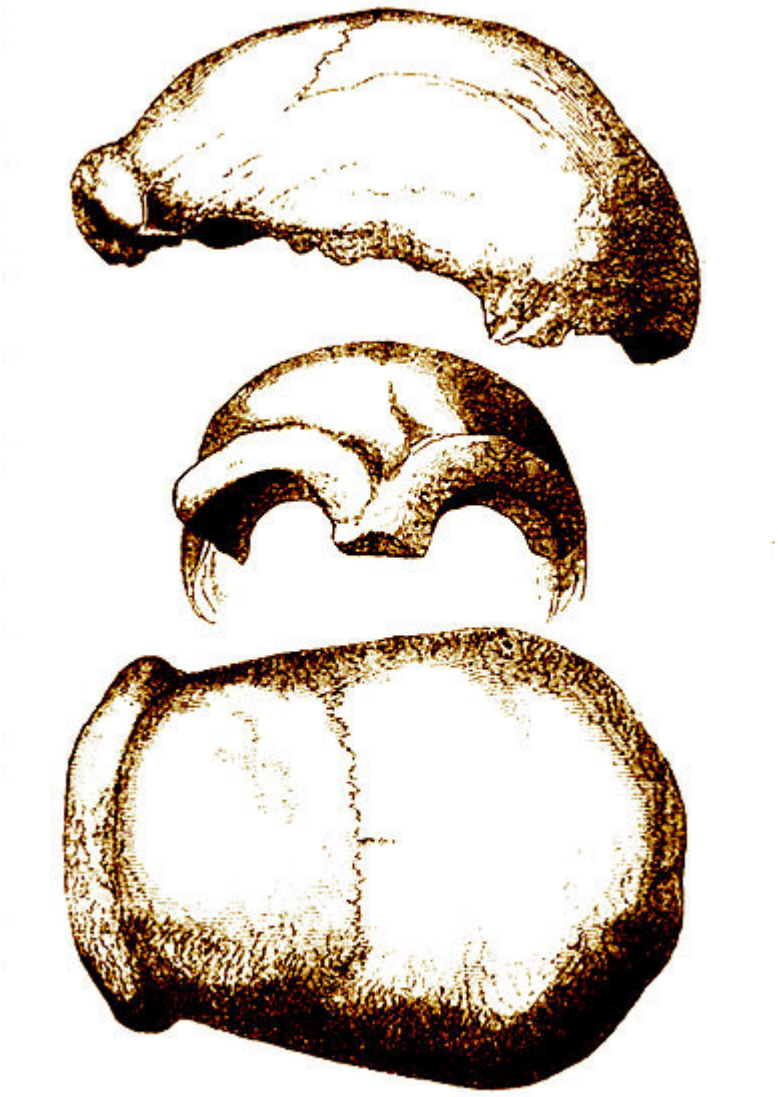
The most vocal proponent of the hybridization hypothesis is Erik Trinkaus of Washington University. Trinkaus claims various fossils as hybrid individuals, including the "child of Lagar Velho", a skeleton found at Lagar Velho in Portugal dated to about 24,000 years ago. In a 2006 publication co-authored by Trinkaus, the fossils found in 1952 in the cave of Peștera Muierii, Romania, are likewise claimed as hybrids.

An estimated 1 to 4 percent of the DNA in Europeans and Asians (i.e. French, Chinese and Papua probands) is non-modern and shared with ancient Neanderthal DNA rather than with Sub-Saharan Africans (i.e. Yoruba and San probands). The cause of this is unclear. It has been suggested it is due to interbreeding between Neanderthals and the ancestors of non-Africans after they left Africa, but this is not certain.

Specimens

The Ferrassie skull

- Neanderthal 1: Initial Neanderthal specimen found during an archaeological dig in August 1856. Discovered in a limestone quarry at the Feldhofer grotto in Neanderthal, Germany. The find consisted of a skull cap, two femora, the three right arm bones, two of the left arm bones, ilium and fragments of a scapula and ribs.
- La Chapelle-aux-Saints 1: Called the Old Man, a fossilized skull discovered in La Chapelle-aux-Saints, France, by A. and J. Bouyssonie and L. Bardon in 1908. Characteristics include a low vaulted cranium and large browridge typical of Neanderthals. Estimated to be about 60,000 years old, the specimen was severely arthritic and had lost all his teeth, with evidence of healing. For him to have lived on would have required that someone process his food for him, one of the earliest examples of Neanderthal altruism (similar to Shanidar I.)
- La Ferrassie 1: A fossilized skull discovered in La Ferrassie, France, by R. Capitan in 1909. It is estimated to be 70,000 years old. Its characteristics include a large occipital bun, low-vaulted cranium and heavily worn teeth.
- Le Moustier: A fossilized skull, discovered in 1909, at the archaeological site in Peyzac-le-Moustier, Dordogne, France. The Mousterian tool culture is named after Le Moustier. The skull, estimated to be less than 45,000 years old, includes a large nasal cavity and a somewhat less developed brow ridge and occipital bun as might be expected in a juvenile.



Type Specimen, Neanderthal 1

- Shanidar 1: Found in the Zagros Mountains in northern Iraq; a total of nine skeletons found believed to have lived in the Middle Paleolithic. One of the nine remains was missing part of its right arm; theorized to have been broken off or amputated. The find is also significant because it shows that stone tools were present among this tribe's culture. One was buried with flowers, showing that some type of burial ceremony may have occurred.

Chapter- 7

Homo Floresiensis

Homo floresiensis
Fossil range: Late Pleistocene

A cast of a *Homo floresiensis* skull, American Museum of Natural History

Scientific classification (disputed)	
Kingdom:	Animalia
Phylum:	Chordata
Class:	Mammalia
Order:	Primates
Family:	Hominidae
Tribe:	Hominini

Genus:	*Homo*
Species:	***H. floresiensis***

Binomial name

†***Homo floresiensis***
Brown et al., 2004

Homo floresiensis ("**Flores Man**", nicknamed "hobbit") is a possible species, now extinct, in the genus *Homo*. The remains were discovered in 2004 on the island of Flores in Indonesia. Partial skeletons of nine individuals have been recovered, including one complete cranium (skull). These remains have been the subject of intense research to determine whether they represent a species distinct from modern humans and the progress of this scientific controversy has been closely followed by the news media at large. This hominin is remarkable for its small body and brain and for its survival until relatively recent times (possibly as recently as 12,000 years ago). Recovered alongside the skeletal remains were stone tools from archaeological horizons ranging from 94,000 to 13,000 years ago.

The discoverers (archaeologist Mike Morwood and colleagues) proposed that a variety of features, both primitive and derived, identify these individuals as belonging to a new species, *H. floresiensis*, within the taxonomic tribe of Hominini. Hominini currently comprises the extant species human (the only living member of the genus *Homo*), bonobo (genus *Pan*) and chimpanzee (genus *Pan*); their ancestors; and the extinct lineages of their common ancestor. The discoverers also proposed that *H. floresiensis* lived contemporaneously with modern humans (*Homo sapiens*) on Flores.

Doubts that the remains constitute a new species were soon voiced by the Indonesian anthropologist Teuku Jacob, who suggested that the skull of LB1 was a microcephalic modern human. Two studies by paleoneurologist Dean Falk and her colleagues (2005, 2007) rejected this possibility. Falk *et al.* (2005) has been rejected by Martin *et al.* (2006) and Jacob *et al.* (2006) and defended by Morwood (2005) and Argue, Donlon *et al.* (2006).

Two orthopedic researches published in 2007 both reported evidence to support species status for *H. floresiensis*. A study of three tokens of carpal (wrist) bones concluded there were similarities to the carpal bones of a chimpanzee or an early hominin such as *Australopithecus* and also differences from the bones of modern humans. A study of the bones and joints of the arm, shoulder and lower limbs also concluded that *H. floresiensis* was more similar to early humans and apes than modern humans. In 2009, the publication of a cladistic analysis and a study of comparative body measurements provided further support for the hypothesis that *H. floresiensis* and *Homo sapiens* are separate species.

Critics of the claim for species status continue to believe that these individuals are *Homo sapiens* possessing pathologies of anatomy and physiology. A second hypothesis in this category is that the individuals were born without a functioning thyroid, resulting in a type of endemic cretinism (myxoedematous, ME).

Discovery

Cave on Flores Island where the specimens were discovered

Flores Island in Indonesia, shown highlighted in red

The specimens were discovered on the Indonesian island of Flores by a joint Australian-Indonesian team of archaeologists in 2003 looking for evidence of the original human migration of *H. sapiens* from Asia to Australia. They were not expecting to find a new species and were surprised at the recovery of a nearly complete skeleton of a hominin they dubbed LB1 because it was unearthed inside the Liang Bua Cave. Subsequent excavations recovered seven additional skeletons, dating from 38,000 to 13,000 years ago. An arm bone provisionally assigned to *H. floresiensis* is about 74,000 years old. The specimens are not fossilized and have been described as having "the consistency of wet blotting paper"; once exposed, the bones had to be left to dry before they could be dug up.[:86]

Researchers hope to find preserved mitochondrial DNA to compare with samples from similarly unfossilised specimens of *Homo neanderthalensis* and *H. sapiens*.

Sophisticated stone implements of a size considered appropriate to the 1-meter-tall human are also widely present in the cave. The implements are at horizons from 95,000 to 13,000 years ago and are associated with (found in the same stratigraphic layer as) an elephant of the extinct genus *Stegodon* (which was widespread throughout Asia during the Quaternary), presumably the prey of LB1. They also shared the island with giant rats, Komodo dragons and even larger species of lizards. *Homo sapiens* reached the region by around 45,000 years ago.

Anatomy

The most important and obvious identifying features of *H. floresiensis* are its small body and small cranial capacity. Brown and Morwood also identified a number of additional, less obvious features that might distinguish LB1 from modern *H. sapiens*, including the form of the teeth, the absence of a chin and the lesser angle in the head of the humerus (upper arm bone). Each of these putative distinguishing features has been heavily scrutinized by the scientific community, with different research groups reaching differing conclusions as to whether these features support the original designation of a new species, or whether they identify LB1 as a severely pathological *H. sapiens*. The discovery of additional partial skeletons has verified the existence of some features found in LB1, such as the lack of a chin, but Jacob and other research teams argue that these features do not distinguish LB1 from local *H. sapiens* morphology. Lyras *et al.* have asserted, based on 3D-morphometrics, that the skull of LB1 differs significantly from all *H. sapiens* skulls, including those of small-bodied individuals and microcephalics and is similar to the skull of *Homo erectus* alone.

Small bodies

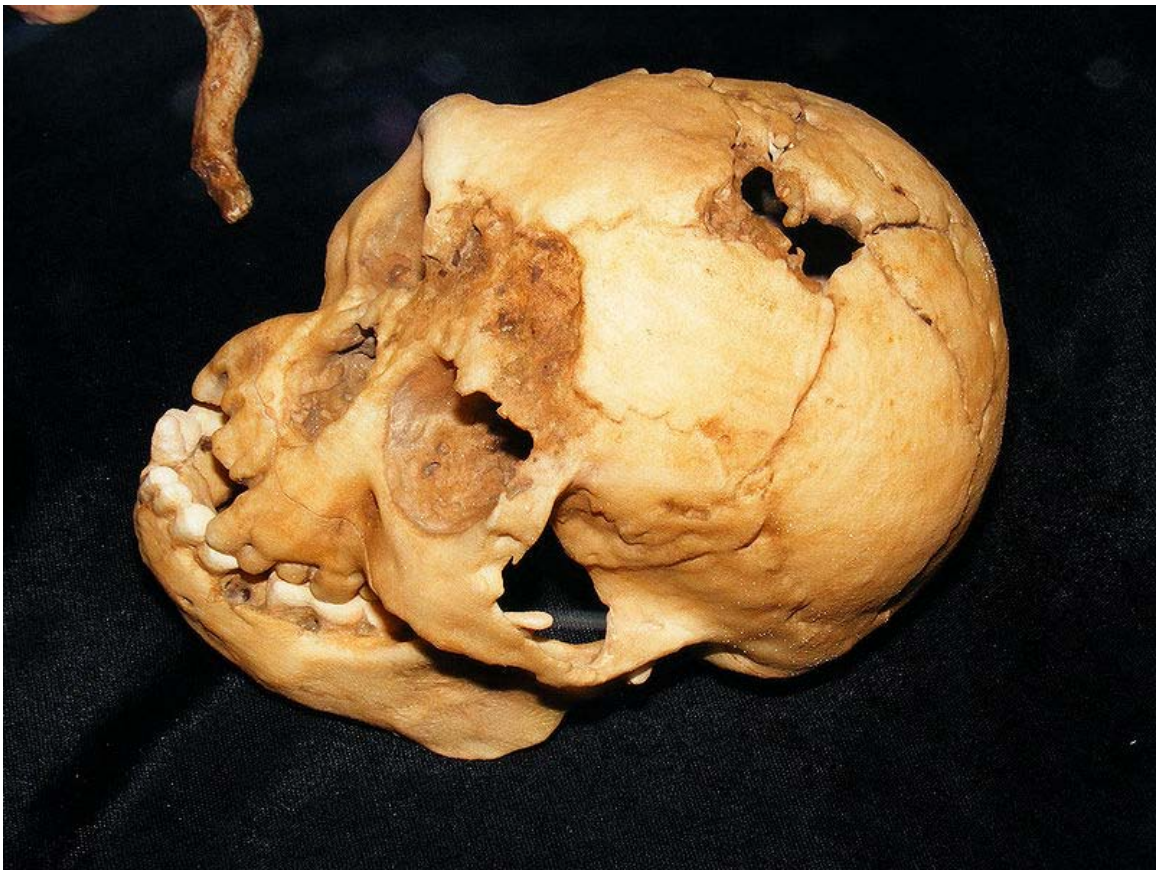
The first set of remains to have been found, LB1, was chosen as the type specimen for the proposed species. LB1 is a fairly complete skeleton, including a nearly complete cranium (skull), determined to be that of a 30-year-old female. LB1 has been nicknamed the *Little Lady of Flores* or "Flo".

LB1's height has been estimated at about 1.06 m (3 ft 6 in). The height of a second skeleton, LB8, has been estimated at 1.09 m (3 ft 7 in) based on measurements of its tibia. These estimates are outside the range of normal modern human height and considerably shorter than the average adult height of even the smallest modern humans, such as the Pygmies (< 1.5 m (4 ft 11 in)), Twa, Semang (1.37 m (4 ft 6 in) for adult women) of Africa, or the Andamanese (1.37 m (4 ft 6 in) for adult women). Body mass is generally considered more biophysically significant than length and by that measure, differences between modern pygmies and *Homo floresiensis* are even greater. LB1's body mass has been estimated at 25 kg (55 lb). This is smaller than that of not only modern *H. sapiens*, but also *H. erectus*, which Brown and colleagues have suggested is the immediate ancestor of *H. floresiensis*. LB1 and LB8 are also somewhat smaller than the australopithecines from three million years ago, not previously thought to have expanded beyond Africa. Thus, LB1 and LB8 may be the shortest and smallest members of the extended human family discovered thus far.

Aside from smaller body size, the specimens seem otherwise to resemble *H. erectus*, a species known to have been living in Southeast Asia at times coincident with earlier finds purported to be of *H. floresiensis*. These observed similarities form the basis for the suggested phylogenetic relationship. Controversially, the same team has reported finding material evidence on Flores (stone tools) of a *H. erectus* occupation dating back 840,000 years ago, but not remains of *H. erectus* itself, much less transitional forms.

To explain the small stature of *H. floresiensis*, Brown *et al.* have suggested that in the limited food environment on Flores, *H. erectus* underwent strong insular dwarfism, a form of speciation which has been observed in other species on Flores also – including a *Stegodon* elephant species on Flores. (This elephant, of normal size, emerged on the island by 750,000 years ago, replacing a dwarf *Stegodon* species that went extinct by 840,000 years ago.) This hypothesis has been criticized by Teuku Jacob and colleagues who argue that LB1 is similar to the midget humans who populate a Flores village, Rampasasa, – and who point out that size can vary substantially in pygmy populations. Contradictory evidence has emerged.

Small brains



Top view of a cast of the LB1 skull

In addition to a small body size, *H. floresiensis* had a remarkably small brain. The brain of the holotype LB1 is estimated to have had a volume of 380 cm^3 (23 cu in)), placing it at the lower range of chimpanzees or the extinct australopithecines. LB1's brain size is half that of its presumed immediate ancestor, *H. erectus* (980 cm^3 (60 cu in)). The brain to body mass ratio of LB1 lies between that of *H. erectus* and the great apes. Insular dwarfism has been posited to explain the brain size reduction.

An indicator of intelligence is the size of Brodmann's area 10, the dorsomedial prefrontal cortex, an area of the brain associated with self-awareness. LB1's region 10 is about the same size as that of modern humans, despite the much smaller overall size of the brain.

Notwithstanding the small brain of *H. floresiensis*, the discoverers have associated it with advanced behaviors. Their cave shows evidence of the use of fire for cooking and *Stegodon* bones associated with the hominins have cut marks. The hominin specimens have also been associated with stone tools of the sophisticated Upper Paleolithic tradition typically associated with modern humans, who have nearly quadruple the brain volume (1,310–1,475 cm^3 (80–90.0 cu in)) and 2.6 times greater body mass. Some of these tools were apparently used in the necessarily cooperative hunting of *Stegodon* by these hominins.

Additional features

Additional features used to argue that the finds come from a population of previously unidentified hominids include the absence of a chin, the relatively low twist of the arm bones and the thickness of the leg bones. The presence of each of these features has been confirmed by independent investigators but their significance has been disputed.

The forearm and pectoral girdle of *H. floresiensis* have been examined by Larson *et al.* (2007). Modern humans have the top of the bone twisted between 145 to 165 degrees to the plane of the elbow joint. For LB1, the twist was initially reported to be 110 degrees. Larson later revised this measurement to 120 degrees. This could be an advantage when arm-swinging, but it complicates activities associated with modern people, such as tool-making. As for the pectoral girdle of *H. floresiensis*, they studied a broken clavicle of LB1 and a shoulder blade of LB6. The clavicle was relatively short, which in combination with the shape of the shoulder blade and the low twist of the arm bone resulted in the shoulder being moved slightly forward, as if it was shrugged. Thus *H. floresiensis* could bend the elbow in the way modern people do and Larson concluded that it may have been able to make tools.

Tocheri *et al.* (2007) examined three carpal bones believed to belong to LB1. The shapes of these bones were claimed to differ significantly from the bones of the modern human wrist and to resemble the wrist of great African apes or *Australopithecus*.

The feet of *H. floresiensis* were unusually flat and unusually long in relation with the rest of the body. As a result, when walking, it would have to bend its knees further back than modern people do. This forced the gait to be high stepped and the creature was not able to walk very fast. The toes had an unusual shape and the big toe was very short.

Recent survival

The species is thought to have survived on Flores at least until 12,000 years before present, making it the longest lasting non-modern human, surviving long past the Neanderthals (*H. neanderthalensis*), which became extinct about 24,000 years ago.

Because of a deep neighboring strait, Flores remained isolated during the Wisconsin glaciation (the most recent glacial period), despite the low sea levels that united Sundaland. This has led the discoverers of *H. floresiensis* to conclude that the species, or its ancestors, could only have reached the isolated island by water transport, perhaps arriving in bamboo rafts around 100,000 years ago (or, if they are *H. erectus*, then about 1 million years ago). This idea of *H. floresiensis* using advanced technology and cooperation on a modern human level has prompted the discoverers to hypothesize that *H. floresiensis* almost certainly had language. This suggestion has been one of the most controversial of the discoverers' findings.

Local geology suggests that a volcanic eruption on Flores approximately 12,000 years ago was responsible for the demise of *H. floresiensis*, along with other local fauna,

including the elephant *Stegodon*. Gregory Forth hypothesized that *H. floresiensis* may have survived longer in other parts of Flores to become the source of the *Ebu Gogo* stories told among the local people. The *Ebu Gogo* are said to have been small, hairy, language-poor cave dwellers on the scale of this species. Believed to be present at the time of the arrival of the first Portuguese ships during the 16th century, these creatures are claimed to have existed as recently as the late 19th century. Gerd van den Bergh, a paleontologist working with the fossils, reported hearing of the Ebu Gogo a decade before the fossil discovery. On the island of Sumatra, there are reports of a 1-1.5m tall humanoid, the Orang Pendek which might be related to *H. floresiensis*. Henry Gee, senior editor at *Nature* magazine, speculates that species like H.floresiensis might still exist in the unexplored tropical forest of Indonesia.

Some anthropologists do not believe the specimens represent a different species. Teuku Jacob, formerly chief paleontologist of the Indonesian Gadjah Mada University argued that they were members of "... a sub-species of *Homo sapiens* classified under the Austrolomelanesid race". He contended that the LB1 find is from a 25–30 year-old omnivorous subspecies of *H. sapiens*, probably a pygmy and that the small skull is due to the genetic disorder microcephaly, which produces a small brain and skull.

Scandal over specimen damage

In early December 2004, Teuku Jacob removed most of the remains from their repository, Jakarta's National Research Centre of Archaeology, with the permission of only one of the project team's directors and kept them for three months. Some scientists expressed the fear that important scientific evidence would be sequestered by a small group of scientists who neither allowed access by other scientists nor published their own research. Jacob returned the remains on February 23, 2005 with portions severely damaged and missing two leg bones to the worldwide consternation of his peers. Reports noted the condition of the returned remains; "[including] long, deep cuts marking the lower edge of the Hobbit's jaw on both sides, said to be caused by a knife used to cut away the rubber mould"; "the chin of a second Hobbit jaw was snapped off and glued back together. Whoever was responsible misaligned the pieces and put them at an incorrect angle"; and, "The pelvis was smashed, destroying details that reveal body shape, gait and evolutionary history" and causing the discovery team leader Morwood to remark "It's sickening, Jacob was greedy and acted totally irresponsibly". Jacob, however, denied any wrongdoing. He stated that the damages occurred during transport from Yogyakarta back to Jakarta despite the physical evidence to the contrary that the jawbone had been broken while making a mold of bones.

In 2005 Indonesian officials forbade access to the cave. Some news media, such as the BBC, expressed the opinion that the reason for the restriction was to protect Jacob, who was considered "Indonesia's king of palaeoanthropology", from being proven to be wrong. Scientists were allowed to return to the cave in 2007 shortly after the death of Jacob.

Microcephaly hypothesis

Prior to Jacob's removal of the fossils, a CT scan was taken of the skull and a virtual endocast of the skull (i.e., a computer-generated model of the skull's interior) of *H. floresiensis* was produced and analysed by Dean Falk *et al.* This team concluded that the brainpan was not that of a pygmy nor an individual with a malformed skull and brain.

In response, Weber *et al.* conducted a survey the same year comparing the computer model of LB1's skull with a sample of microcephalic human skulls, concluding that the skull size of LB1 falls in the middle of the size range of the human samples and is not inconsistent with microcephaly. Next to dispute the finding of Falk *et al.* (2005) were Martin *et al.* (2006), who objected to the failure to compare the model of LB1's skull with a typical example of adult microcephaly. Martin and his coauthors concluded that the skull was probably microcephalic, arguing that the brain is far too small to be a separate dwarf species; if it were, the 400-cubic-centimeter brain would indicate a creature only one foot in height, one-third the size of the discovered skeleton. Shortly thereafter, a group of scientists from Indonesia, Australia and the United States came to the same conclusion by examining bone and skull structure (Jacob (2006)).

A cast of LB1 (left) was compared to several microcephalic skulls, amongst which is that of the microcephalic (right) used by Henneman in his attempt to present LB1 as a microcephalic. Argue (2006) and Lyras (2008) contend the opposite.

Brown and Morwood countered by claiming that the skeptics had drawn incorrect conclusions about bone and skull structure and mistakenly attributed the height of *H. floresiensis* to microcephaly. Falk's team replied to the critics of their study (Falk *et al.* (2006)). Morphologist Jungers examined the skull and concluded that the skeleton displays "no trace of disease". Argue, Donlon, *et al.* (2006) rejects microcephaly and concludes that the finds are indeed a new species.

Falk *et al.* (2007) offered further evidence that the claims of a microcephalic *H. sapiens* were not credible. Virtual endocasts of an additional nine microcephalic brains and ten normal human brains were examined and it was found that the *floresiensis* skulls are similar in shape to normal human brains, yet have unique features which are consistent with what one would expect in a new species. The frontal and temporal lobes of the *floresiensis* brain were found to be highly developed, in strong contrast to the microcephalic brain and advanced in ways different from modern human brains. This finding also answered past criticisms that the *floresiensis* brain was simply too small to be capable of the intelligence required for the members of *H. floresiensis* to create the tools found in their proximity. Falk *et al.* (2007) conclude that the onus is now upon the critics that continue to claim microcephaly to produce a brain of a microcephalic that bears resemblance to the *floresiensis* brain.

Falk's argument was supported by Lyras et al. (2008) in that 3D-morphometric features of the skulls of microcephalic *H. sapiens* indeed fall within the range of normal *H. sapiens* and that the LB1 skull falls well outside this range. This was interpreted as proving that LB1 cannot, on the basis of either brain or skull morphology, be classified as a microcephalic *H. sapiens*.

In 2009, a study by Jungers et al. presented a statistical analysis of skull shapes of healthy modern humans, microcephalic humans and several ancient human species, as well as *H. floresiensis*. They showed that the three grouped separately, with *H. floresiensis* among the ancient humans, providing evidence that *H. floresiensis* is a separate species instead of a diseased modern human.

Laron syndrome hypothesis

The anatomist Gary D. Richards introduced a new skeptical hypothesis in June 2006: that the skeletons from Flores might be the remains of people who suffered from Laron syndrome, a genetic disorder first reported in 1966. The next year, a team including Laron himself published a paper arguing that the morphological features of *H. floresiensis* are essentially indistinguishable from those of Laron syndrome. The team said that to determine whether the *H. floresiensis* individuals had Laron syndrome would require testing their DNA for the presence of the defective genes, if samples of that DNA ever become available. Critics of the hypothesis have however pointed out that despite the low stature, people suffering from Laron syndrome look nothing like the *Homo floresiensis* remains, particularly in the anatomy of the cranial vault.

Endemic cretinism hypothesis

In 2008 Australian researchers Peter J. Obendorf, Charles E. Oxnard and Ben J. Kefford suggested that LB1 and LB6 suffered from myxoedematous (ME) endemic cretinism resulting from congenital hypothyroidism and that they were part of an affected population of *H. sapiens* on the island. This disease, caused by various environmental factors including iodine deficiency, is a form of dwarfism which can still be found among the local Indonesian population. Affected people, who were born without a functioning thyroid, have both small bodies and reduced brain size but their mental retardation and motor disability is not as severe as with neurological endemic cretins. According to the authors of the study, the critical environment could have been present on Flores approximately 18,000 years ago, the period to which the LB fossils are dated. They wrote that various features found on the fossils, such as enlarged pituitary fossa, unusually straight and untwisted tops of the upper arm bone and relatively thick limbs, are a sign of this diagnosis. The double rooted lower premolar and primitive wrist morphology can be explained in this way as well. The oral stories about strange human-like creatures may also be a record of cretinism.

Falk challenged the premise of Oberndorf *et al.* Studying computer tomography scans of LB1's pituitary fossa, she came to the conclusion that it is not larger than usual. Peter Brown declared that the remains of the pituitary fossa were very poorly preserved and no meaningful measurement was possible.

In 2010 Colin Groves compared the Flores bones with those of ten people who had had cretinism, focusing on anatomical features which are typical of the disease. He found no overlap and stated that he had put the claim to rest. Unfortunately this article remains unpublished and the published abstract is insufficient to judge the merits or demerits of this work. However, a more recent article by Oxnard, Obendorf and Kefford rejects Groves' argument and revives the cretinism hypothesis. Oxnard *et al* also criticise the cladistic analysis of Argue *et al.* (2009), stating that it is not logically possible for the analysis to conclude that the Liang Bua remains represent a separate species and not a pathology because the cladistics analysis assumes that they do not represent a pathology.

Bone structure

Cast of the entire LB1 specimen

The bone structure of *H. floresiensis*' shoulders, arms and wrists have been described as very different from modern humans, much closer to the bone structure of chimpanzees or an early hominin. This adds support to the idea that *H. floresiensis* is a separate species of early human rather than a modern human with a physical disorder.

Susan G. Larson *et al.* analyzed the upper limb of LB1. They found that in LB1 the angle of humeral torsion is much less than in modern humans. This had been previously studied by Richards et al., who declared that it is a sign of modern pygmy populations and T. Jacob et al., who pointed out that muscle attachments on the bone suggest LB1 had weak muscles which resulted in little development of humeral torsion. Larson *et al.* rejected Richards' conclusion, arguing that the humeral torsion of pygmy populations is usually similar to that of peoples of average stature. They argued that Richards *et al.* cited a 1972 paper which had studied a sample of six female Eastern Central African pygmies and this sample was too small to represent the whole population. Larson *et al.* also failed to find signs of microcephaly on the studied bones.

Larson *et al.* also studied the relatively short clavicle and the unusual formation of the pectoral girdle. They compared their finding with the skeleton of Nariokotome Boy

(variously classified as *H. ergaster* or *H. erectus*) and suggested that the pectoral girdle of *H. floresiensis* was a transitional stage in human shoulder evolution.

While some specialists, including paleoanthropologist Russell Ciochon of the University of Iowa, supported the conclusion, others, including Eric Delson of Lehman College, City University of New York, pointed out that the recent sample of *H. floresiensis* individuals is too small and that Larson's research was based just on one shoulder bone.

Tocheri *et al.* (2007) (including Morwood, Larson and Jungers), compared three carpal bones believed to belong to LB1 with carpal bones of modern humans, some earlier hominids and African apes. They concluded that the carpals from the Liang Bua cave resembled ape carpal bones and were significantly different from the bones of *H. sapiens*, *Homo neanderthalensis* or even *Homo antecessor* and that they were comparable to carpal bones of *Australopithecus*. The carpal bones of *H. floresiensis* were found to lack features that evolved with ancestors of modern humans at least about 800,000 years ago. These features are already formed during embryogenesis and therefore Tocheri et al. argue that it is improbable that the shape of *H. floresiensis* wrist bones could be a result of a developmental disease.

This conclusion was challenged by Robert Martin, since Jacob's death the leading proponent of the microcephaly hypothesis and Alan Thorne. Martin noted that no research has been done on wrists of microcephalic people. Thorne maintained that the differences were small and that similar variation could occur with living modern humans. He also pointed out that the carpal bones had been found scattered in the cave and it was not certain that they all belonged to the same individual. Project leader Morwood countered that there were also other features, such as the stature, body proportions, brain size, shoulder, pelvis, jaw and teeth which suggested that *H. floresiensis* is a separate species that evolved in isolation on the island.