ORIGIN OF MODERN HUMANS

Paleoanthropology: How Old Is the Oldest Human?
Jean-Jacques Hublin

Figure 1. This 3.3 mya specimen is known as “Oldoway Harmand.”
Harmand et al 2015 Nature 521:310-315

A 3.3 Ma old mandible unearthed in Ethiopia fills the gap between ape-like australopithecines and representatives of the genus Homo. It pushes the origin of large-brained hominins further back in time and highlights the complexity of the human evolutionary tree.

Current Biology 25, R444-R448, June 1, 2015

Cut marks in bone 3.4 mya

Stone tools 3.3 mya

Oldowan tools 2.6 - 1.5 mya

**APPEARANCE OF THE GENUS HOMO**

- Increasingly large brain
  - *habilis* - 650-800 cc
  - *erectus* - 900-1200 cc
  - *sapiens* - 1200-1400 cc

- Stone tool use
- Number of biological species???

*Homo habilis* 1.9 mya

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Molecular insights into human brain evolution

Robert Sean Hill and Christopher A. Walsh

Differences in cerebral cortical size are associated with differences in the cerebral cortex circuit diagram.
Intra- and Interspecific Variation in Primate Gene Expression Patterns

Enard et al. used an AFFIMETRIX gene chip with 12,000 human genes to analyze differences in expression patterns among 3 primates.

No significant differences were noted in blood or liver assays but large differences were observed in brain expression patterns.

Rapid evolution of gene expression patterns in the human brain.

Table 1. Brain protein pattern differences between humans and chimpanzees as analyzed by 2D gel electrophoresis. Differences between human and chimpanzee pairs were scored if confirmed in three individual human-chimpanzee pairs and were analyzed in the same way as in a larger mouse study comparing M. musculus and M. apenetus. Qualitative differences represent changes in electrophoretic mobility of spots, which likely result from amino acid substitutions, whereas quantitative differences reflect changes in the amount of protein.

<table>
<thead>
<tr>
<th>Comparison</th>
<th>Analyzed spots</th>
<th>Qualitative</th>
<th>Quantitative</th>
</tr>
</thead>
<tbody>
<tr>
<td>Human-chimpanzee</td>
<td>538</td>
<td>41 (7.6%)</td>
<td>169 (31.4%)</td>
</tr>
<tr>
<td>M. musculus-M. apenetus</td>
<td>8767</td>
<td>668 (7.6%)</td>
<td>658 (7.9%)</td>
</tr>
</tbody>
</table>

Caceres et al. (PNAS 2003) identified 169 genes that exhibited gene expression differences between human and chimpanzee lineages. 91 of these genes differentiated in the human lineage. Most differences involved up-regulation with higher expression levels humans.
Expression profiling in primates reveals a rapid evolution of human transcription factors

The neocortex expanded spectacularly during human evolution, giving rise to distinctively human anatomical and cognitive capabilities. Yet to date, just a handful of genetic loci have been associated with human-specific brain traits and none have been shown to functionally impact corticogenesis in an evolutionarily divergent fashion. In this study, we report the discovery of the first human-accelerated enhancer that functions in brain development... Our study suggests the intriguing hypothesis that evolutionary changes in HARE5 sequence and activity contributed to the origin of unique features of the human brain.

Human-Chimpanzee Differences in a FZD8 Enhancer Alter Cell-Cycle Dynamics in the Developing Neocortex
(Boyd et al. 2015)
Myosin gene mutation correlates with anatomical changes in the human lineage

STEDMAN et al.

Homo (Kenyanthropus) rudolfensis
2.5 – 1.9 mya

Homo ergaster
1.8 – 1.4 mya

“Turkana boy”
- 10-12 years old
- 1.6 million years ago
- Kenya
- called early African H. erectus or H. ergaster
"Turkana boy"
10-12 years old
1.6 million years ago

Starting ~1.6 mya a more sophisticated style of tool emerged

Eastern Africa - Lake Turkana region
coeexistence of different hominids

Richard Leakey
DMANISI, REPUBLIC OF GEORGIA

1.7 mya

Shortly after the origin of *Homo*, there is a migration out of Africa.

DMANISI STONE TOOLS

clearly not close to anatomically modern *H. sapiens*- like Turkana boy
1.77-million-year-old complete adult skull (braincase volume: 546 cubic centimeters) of early Homo from the site of Dmanisi, Georgia. Together with the fossilized bones of four additional individuals discovered in close proximity, the skull indicates that populations of early Homo comprised a wider range of morphological variation than traditionally assumed, which supports a single evolving lineage with continuity across continents.

“By now, the fossils have made it clear that these pioneers were startlingly primitive, with small bodies about 1.5 meters tall, simple tools, and brains one-third to one-half the size of modern humans.”
First migration out of Africa did not include the modern human lineage.

Asian *Homo erectus*

- "Java Man" 0.75 mya
- Peking Man China 0.77 mya

RECONSTRUCTION of Indonesian *H. erectus*
Brain Size:

- A. afarensis
  - 500cc
- H. erectus
  - 900 – 1200 cc
- H. sapiens
  - 1100 – 1400 cc

‘Daka’ Ethiopian Skull

- 1 million-year-old remains from Ethiopia show clear link between Asian Homo erectus and African H. erectus (widespread paleospecies)
- intermediate between earlier and later African fossils

There is a growing realization that there may have been a high degree of sexual dimorphism in this archaic lineage compared to more modern groups.

- Comparison of two H. erectus skulls found in Kenya showing the huge variation in size
Petralona Skull – 250,000 years old
• H. erectus or archaic H. sapiens?

JEBEL IRHOUD 1
MOROCCO
100,000 – 200,000 YEARS OLD

H. erectus or H. sapiens?

COMPETING HYPOTHESES FOR THE DEVELOPMENT OF MODERN HUMANS
**COMPETING HYPOTHESES FOR THE DEVELOPMENT OF MODERN HUMANS**

- **Multiregional model:** archaic *H. sapiens* (or *H. erectus*) dispersed throughout the Old World and simultaneously evolved to modern form with abundant gene flow.

- **Replacement Model (Out-of-Africa):** single group that relatively recently dispersed from Africa, evolved into modern form and replaced all archaic forms, including Neanderthals.

- **Hybridization & Assimilation Model:** This model suggests that archaic lineages spread out from Africa early, followed later by a second wave of dispersal from a more derived lineage. Some amount of hybridization accompanied the replacement of the archaic lineages.

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**Statistical evaluation of alternative models of human evolution**

Fangundes et al. 2007 PNAS 17614-17619

Using DNA data from 50 nuclear loci sequenced in African, Asian and Native American samples... a simple African replacement model with exponential growth has a higher probability (78%) as compared with alternative multiregional evolution or assimilation scenarios. A Bayesian analysis of the data under this best supported model points to an origin of our species 141 thousand years ago (Kya), an exit out-of-Africa 51 Kya, and a recent colonization of the Americas 10.5 Kya. We also find that the African replacement model explains not only the shallow ancestry of mtDNA or Y-chromosomes but also the occurrence of deep lineages at some autosomal loci, which has been formerly interpreted as a sign of interbreeding with *Homo erectus*.

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**Recent comparisons of ENTIRE mtDNA genome indicate a recent (0.2 mya), African origin of anatomically modern humans.**
Y CHROMOSOME STUDIES

Dorit et al. 1995  173,000
Hammer et al. 1995  188,000

- mtDNA and Y chromosome studies yield similar dates for the MRCA of modern humans.
- Taken together, these studies strongly suggest that the *multiregional model* cannot be correct.

And maybe the origin of modern humans goes back even a bit farther......
WHAT ABOUT NEANDERTHALS???

H. sapiens neanderthalensis
50,000 years ago
France

H. sapiens sapiens
Present

* Neanderthal fossils are found from 400,000 to about 30,000 years ago in Europe and western Asia.

50,000 year old skull from La Ferassie, France

Homo erectus
Homo neanderthalensis
Homo sapiens
RECONSTRUCTION BASED ON SKULLS FROM SHANIDAR CAVE IN IRAQ

- Prominent brow ridge
- Large bulbous nose
- Powerful build
- Possible advanced culture: sophisticated tools, burials, language?

HYOID BONES CLOSELY RESEMBLE MODERN HUMANS

Chimpanzee  Neanderthal

Sympatry H. neanderthalensis and H. sapiens
Ancient DNA samples from *H. neanderthalensis*


**mtDNA SEQUENCE ANALYSIS FROM NEANDERTHAL BONE (>30,000 YEARS OLD)**

- Neanderthal sequences are 3 times as divergent from modern human sequences than are the most divergent modern humans.
- It is unlikely that Neanderthals were assimilated into human populations.

1% to 4% of the DNA of Europeans and Asians, but not of Africans, was shared with Neandertals and concluded that modern humans interbred with Neandertals at low levels (*Science*, 7 May 2010, pp. 680, 710)
This view of the Altai Mountains is from just above Denisova Cave, where the fragment of bone analysed by Krause et al. was discovered. The excavation field camp is visible in the valley below.

**Stranger from Siberia**
Terence A. Brown

The sequencing of ancient DNA is generating dramatic results. The sequence from a bone fragment has revealed the existence of an unknown type of extinct human ancestor that lived in Asia 40,000 years ago.

- Pairwise nucleotide differences from all pairs of complete mtDNAs from 54 present-day and one Pleistocene modern human, six Neanderthals and the Denisova hominin are shown.

![Diagram showing pairwise nucleotide differences](image)

The complete mitochondrial DNA genome of an unknown hominin from southern Siberia

![Mitochondrial DNA phylogeny](image)
Location of the Middle Pleistocene site of Sima de los Huesos (yellow) as well as Late Pleistocene sites that have yielded Neanderthal DNA (red) and Denisovan DNA (blue).
Bayesian phylogenetic tree of hominin mitochondrial relationships based on the Sima de los Huesos mtDNA sequence determined using the inclusive filtering criteria.

A Revised Timescale for Human Evolution Based on Ancient Mitochondrial Genomes

Denisova  657 – 973 kya
Neanderthal  326 – 482 kya
Modern Humans  134 – 188 kya
Out-of-Africa  62 – 95 kya

Learning about human population history from ancient and modern genomes
Mark Stoneking & Johannes Krause
Palaeoanthropology: Human evolution writ small

We are the only living species of the genus Homo. Given the startling results of a cave excavation in Southeast Asia, it seems that we coexisted with another species until much more recently than had been thought.
A small, but significant, portion of modern human genomes has roots in archaic forms.

- Europeans and Asians have 2-4% Neanderthal DNA.
- Melanesians and Aboriginal Australians have up to 5% Denisovan DNA.

A number of phenotypic traits are associated with these archaic genes including an increased or decreased risk of:

- Osteoporosis
- Blood-coagulation disorders
- Nicotine addiction
- Depression
- Obesity
- Skin disorders

Specific genes include:

- Toll-like receptors involved in immunity (Neanderthals & Denisovans)
- EPAS1 involved in high altitude adaptation (Denisovans)
Cave Paintings ~35,000 years ago

35,000 year-old statues

More....
RAPID INCREASE IN TECHNOLOGY, ART, AND MUSIC DEMONSTRATES CULTURAL EVOLUTION IN MODERN HUMANS.

HUMANS ARE UNIQUE IN THEIR EXTREMELY HIGH RATE OF CULTURAL EVOLUTION

- To what extent is an interaction between cultural evolution and phenotypic evolution possible?

- H. floresiensis ~18,000
- Denisova ~40,000
- H. neanderthalensis ~30,000

Possibly 4 separate lineages as recently as 40,000 years ago

Co-occurrence Late