

## 42-43. Human evolution

- The oldest fossils in the lineage leading to humans are dated to ~6 mya. These fossils were from individuals who were at least partially bipedal and had brains similar than those of modern chimpanzees.
- Fossils classified in the genus *Homo* first appeared 2.3 mya. By 1.8 mya, *H. erectus* was present in Africa. By 1.5 mya *H. erectus* fossils are found in Europe and Asia, indicating dispersal from Africa.
- Fossils similar to modern humans, *H. sapiens*, dated to 160,000-190,000 years ago, are found in eastern Africa and the Middle east. Fossils classified as a separate species, *H. neanderthalensis*, are found in Europe and western Asia beginning 100,000 years ago. Both *H. sapiens* and *H. neanderthalensis* lived in Europe for at least 70,000 years. There is no archaeological evidence that there was contact between the two species. The youngest Neanderthal fossils are ~32,000 years old.
- DNA sequences have helped resolve several questions about the evolution of humans. One question is about the time of human-chimpanzee speciation. Humans and chimpanzees differ at about 1.2% of the nucleotide positions, with slightly less divergence on the X and somewhat more on the Y. The best estimate of the divergence time between the ancestors of chimps and modern humans is between 6-8 my, suggesting that the oldest hominid fossils represent the hominid lineage shortly after the two species separated.
- Another question is whether modern Europeans and Asians descended from *H. erectus* in those regions (the local continuity theory) or whether *H. erectus* in Europe and Asia went extinct and were replaced later by *H. sapiens* dispersing from Africa (the out-of-Africa theory). The times of the MRCA of both mtDNA and Y-chromosomes trees much too recent to be consistent with the local continuity theory. Furthermore, the fact that African sequences are found at the deepest branches of both the mtDNA and Y-chromosome gene trees indicates that modern humans dispersed from Africa to the rest of the world.
- Nuclear genes generally have longer times of MRCAs, as old as 4 mya. This does not refute the out-of-Africa theory. It just illustrates how variable coalescence times can be.
- Another question is whether there was any genetic exchange between humans and Neanderthals while they coexisted in Europe. Thousands of complete mtDNA sequences from modern humans are available and they show no evidence of gene flow from Neanderthals to modern humans during the period they coexisted.
- Another question is about the effective population size of humans. In humans,  $\pi \approx 0.001$  per site. Using  $N = \pi / (4\mu L)$ ,  $N_{\pi} \approx 10^{-3} / (4 \times 2 \times 10^{-9}) = 11,250$ . This tells us that the effective population size of humans is much smaller than the current census size, which is about  $6 \times 10^9$ . The current  $\pi$  reflects a much smaller population in the distant past. In chimps and gorillas,  $\pi$  is at least twice as large as in humans. Together these observations suggest that there was a reduction in population size, a bottleneck, after the separation of the hominid lineage from the chimp lineage.
- The observation that Tajima's D tends to be negative in non-Africans but not in Africans suggests that there was also a bottleneck in the population that left Africa to colonize the rest of the world.

Underhill PA, Kivisild T (2007) Use of Y chromosome and mitochondrial DNA population structure in tracing human migrations. *Annual Review of Genetics* 41:539-564.  
<http://arjournals.annualreviews.org/doi/abs/10.1146/annurev.genet.41.110306.130407>

Garrigan D, Hammer MF (2006) Reconstructing human origins in the genomic era. *Nature Reviews Genetics* 7:669-680  
<http://www.nature.com/nrg/journal/v7/n9/abs/nrg1941.html>

Chimpanzee Sequencing Consortium (2005) Initial sequence of the chimpanzee genome and comparison with the human genome. *Nature* 437:69-87  
<http://www.nature.com/nature/journal/v437/n7055/full/nature04072.html>

42.1 What properties of the gene tree of mtDNA would have been found in the local continuity theory were correct?

Ans. (i) The time of the MRCA would be at least 1.5 million years, the time when *H. erectus* dispersed from Africa. (ii) The gene trees for mtDNAs from Europe, Africa and Asia would probably form monophyletic group.

42.2 What properties of the gene tree of mtDNA from humans and the Neanderthal would indicate that there was gene flow from the Neanderthal to modern humans?

Ans. The human gene tree would not be monophyletic. Some humans would be more closely related to the Neanderthal mtDNA than they would be to other modern humans.

42.3 If it is later established that there was gene flow between humans and Neanderthals, what difference would it make?

Ans. They would not be classified as separate species.