Primates, population genetics and human evolution[1]

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Abstract: *We humans like to think of ourselves as special, set apart from the rest of the other primates by our ability to walk, talk, build complex structures, exhibit emotions, have special functions of structural genes and so on. But, recent discoveries of primatologists place other higher primates in most of these characteristic very close to humans.*

In contrast to creationists, evolutionists consider humans as products of evolutionary chains and launch convincing arguments to conclude that each creature stands at the end of its evolutionary chain and, therefore, is unique. On the basis of this conclusion, there is no reason for arguing that humans have an overwhelming special position within the animal kingdom. Yet, the question remains which of our traits make us human?

In terms of morphology, physiology, behaviour, and, in particular, of genetics, extant chimps and gorillas are our closest relatives, because humans and pongids share the same last common ancestor (LCA) of about 6–8 million years ago. And, therefore, there is more a continuum in quantitative traits than a profound distinction in qualitative characteristics between these forms. First convincing roots of Homo sapiens *are verified by 100,000 years old fossil remains of humans in Africa and Near East, that started emigrating into all continents. This "Out-of-Africa Hypothesis" finds its strong support by genetic data.*

Population genetics in catarrhine primates elucidates evolutionary processes (migration, genetic drift, differential reproduction) which lead to genetic differentiation within a relatively short time and which might have played a major role also in the population history of early humans.

Nonhuman primates, population genetics and early humans

Philosophers, morphologists, anthropologists and others of former times attributed to humans a special place in nature. Philosophical endeavours envisaged to define the essence of "man", such as only humans are rational creatures (F. Hegel), have an awareness of time and history (F. Nietzsche), of death (M. Heidegger). Others defined "man" as an *animal symbolicum* (Cassirer), as

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tool making animal (B. Franklin), as tool maker (K.P. Oakley). In contrast, there is recent good evidence that other catarrhine primates (such as macaques and pongids), explore their habitat rationally, fully aware of time during their daily routine walks through the woods. As for the discussion of tool use and consequently for the initial steps towards culture in nonhuman primates, it can be proven that orangs (*Pongo abelii*, *Pongo pygmaeus*; Schaik et al., 2003) chimps (Boesch and Boesch, 2000) and gorillas exhibit tool use and correlated phenomena of a "culture". Highly experienced anatomists (among others: Schultz, 1936; Hofer and Altner, 1972; Groves, 1986) argued that the pongids represent the closest living relatives to "man" on the one hand, but that humans exhibit specifications on the other, as far as the skeleton and the soft tissues are concerned. At the time being, the discussion runs over the statement that humans are different more in quantitative than in distinct qualitative traits (see **Tab. 1**). This argument can best be discussed with a closer look at the evolution of primate chromosomes where structural rearrangements lead to the species-specific differences between the species whereas the genetic material remained preserved. The banding patterns of chromosomes in pongids and humans show striking similarities, a strong hint of their phylogenetic relationships (Dutrillaux, 1975). Beyond that, due to the banding pattern, the human chromosome 2 is the product of a fusion of two pongid chromosomes which has occurred in an early ancestor of *Homo*, the reason why humans have 46 chromosomes in the diploid set, instead of 48 as in pongids (Marks, 2005) but preserved identical loci in both lineages. And when it comes to genes, Klein's example of the leucocyte antigens-complex (MHC) holds for the general picture where humans and chimps share a great deal of identical gene products, because they stem from the same ancestor and beyond that, they have their species-specific gene informations, due to occurred mutations after the splitting into the pongid and hominid lineage (for instance locus MHC-DQ). Molecular geneticists claim a small DNA difference of only 1.6% between humans and chimpanzees that must be responsible for the visible quantitative differences in morphology, physiology, behaviour etc. between these two species. The evidence of multiple genes in the studies of (Satta et al., 2000; Chen and Li, 2001; O'h Uigin et al., 2002) lead to the conclusions that 1) the chimpanzee rather than the gorilla is the nearest living relative of *Homo sapiens*, 2) the splitting of their lineages took place in a relatively short time interval. Furthermore, in cladistic terminology, the human and the chimpanzee species form a monophyletic group (clade), they are sister species. The issue is still in debate, which small but effective gene mutations made us uniquely human. At present, there is reliable evidence that even subtle differences in gene activity can result in big phenotypic distinctions.

Many studies of population genetics in nonhuman primates demonstrate how populations differentiate genetically within a short period, in particular due to fissions into smaller units (social groups) and to migration patterns of certain individuals. The combined data of field studies, behaviour and genetics in *Macaca sylvanus* give hints that after the fission event, the new filial groups have become genetically rather different, due to a kind of kin structured distribution into the new units which ends up in a higher portion of related individuals in

Tab. 1. Selected characteristics in humans and other primates (mainly based on Groves, 1986).

Characteristics	Humans	Pongids
Skull	Upright on the vertebral column	Angled on the vertebral column
Chin	Present	Absent
Canines	Small Almost no sexual dimorphism Canines erupt before premolars	Relatively big, often sexual dimorphism Canines erupt after premolars
Spine	S-shaped	Relatively straight
Os ilium	Broader than high	
Thumb	Elongated	
Heel bone	Elongated prominence	
Adult female breast	Permanently enlarged	
Penis	Boneless	Bone: os baculum
Sperma	Density low	Density higher
Gestation period	Lengthened	Very much shorter
Chromosomes	Chromosome 2 as product of chromosomal fusion	Other chromosomal rearrangements of identical loci

the filial groups (Scheffrahn et al., 1993). Field studies in *Macaca fascicularis* on Sumatra lead to the observation of differential reproduction, in the sense that higher ranking males reproduce more children over a period of 4–6 years when the alpha-male is in power. Due to this effect, neighboring social groups exhibit a genetically different composition of the gene pool (Scheffrahn et al., 1996).

Very similar processes of group/population differentiation might have occurred during the global expansion of *Homo sapiens*. This conclusion is also corroborated by the observation on migration of smaller human groups of hunter and gatherers (a striking example has been given by Ducros, 1976). New evidence for the so-called "Out-of-Africa Hypothesis" of modern humans suggests that our ancestors did evolve somewhere in East Africa about 200,000 years ago and started leaving the continent about 100,000 years ago. This period scenario is highly likely because the most confidential early *Homo sapiens* has been excavated in the Near East (Kafzeh IX; Vandermeersch, 1981), and has been dated around 95,000 years old. Early *Homo sapiens* populations have migrated into the other continents until they have reached the remotest corners of our globe about 30,000–10,000 years ago (see **Fig. 1**). In reference to that short time period, it seems to be highly likely that the observed distinguishing features between the extant human populations are less caused by new gene mutations than by the combined effects of other evolutionary processes, such as genetic drift, migration of small social units, differential reproduction ("leader effect") etc. Edwards and Cavalli-Sforza proposed one of the first gene trees of extant human populations (Edwards et al., 1964), drawn from gene frequencies of blood protein data (20 alleles of 5 blood group systems only), and came, with their

general grouping, close to the phylogenetic tree of Nei and Roychoudhury in 1982 which was based on 121 gene loci in some analyses (Nei and Roychoud-hury, 1982). They gained several far-reaching conclusions: 1) The tree reveals that all extant human populations are derived from an ancestral population that existed in Africa 110,000 years ago; 2) There are three main groups of populations covering the Africans, Asians and Europeans. In particular, the first conclusion met the interest of palaeoanthropologists who found a great deal of fossils to corroborate the hypothesis that *Homo sapiens* originated in Africa and then spread to all the other continents. At this point, an important observation has to be mentioned: The degree of variation of numerous genetic systems appears in African populations considerably higher than in Non-African groups. A very plausible explanation for this phenomenon is the loss of alleles per locus due to bottleneck effects and genetic drift during the exodus.
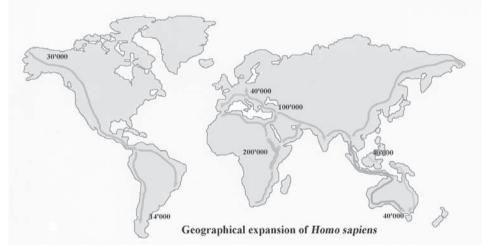

Fig. 1. Geographical dispersal of *Homo sapiens* starting in East Africa (200,000 BP) to the remotest continental corners of South America, South Africa and South Australia.

By the early 1980s, the palaeoanthropological debate over the origin of "anatomically modern humans" restarted with publications by P. Andrews, Ch. Singer, G. Bräuer and others (cf. Bräuer, 1984) by arguing that all extant humans have their origin in Africa. Pilbeam has tried to bring both branches of this dispute together, the genetic and fossil record of human evolution, and to set up a synthesis by concluding that their findings are compatible with the interpretation that the cradle of humans stood in Africa (Pilbeam, 1996).

The present palaeoanthropological record matches best the formal scenario of the "Out-of-Africa Hypothesis". It postulates that modern humans first arose as a new species *Homo sapiens* 150,000 to 200,000 years ago in Africa and had undertaken its initial dispersion out from Africa into other regions of the globe. According to this hypothesis, archaic humans, such as European Neanderthals, were replaced, due to the higher cultural standard of the newcomers that lead to their demographic expansion ("replacement model"). At the time being, this scenario has roughly been supported by genetic data over the last

Extant
Caucasians

– Lake Mungo

and

Asians

Extant
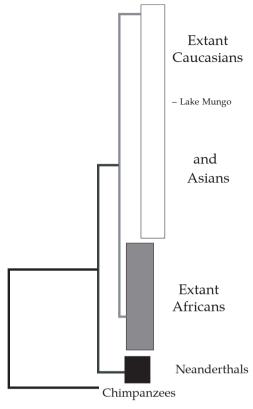Africans

Neanderthals

Chimpanzees

Fig. 2. Phylogenetic tree of the human mtDNA hypervariable segment 1, based on Y. Satta (in: Klein and Takahata, 2002)

20 years. Recent genetic evidence of DNA data of microsatellites, the Y-chromosome and of mitochondria make the acceptance of this hypothesis possible. The genetic debate over human origins restarted with mtDNA sequence analyses (Cann et al., 1987), revised and reinterpreted (Stoneking, 1993; Ruvolo, 1996). Nowadays, it is possible to analyze the entire mtDNA genome of many individuals from very diverse geographic and ethnic global areas. The mtDNA make-up of these sufficiently numerous individuals favours the hypothesis to trace our common ancestry back to Sub-Saharan Africa where somewhen existed a relatively small founder population of 40,000–60,000 individuals which gave rise to all modern humans. Detailed analysis of Y-chromosome sequences and microsatellites (Hammer et al., 1997) of different genetic loci delivered also strong evidence in favour of the "Out-of-Africa Hypothesis". Y. Satta's compilation of mtDNA sequence data (in: Klein and Takahata, 2002) from Africans, Asians, Caucasians, from three Neandertals and of the ancient *Homo sapiens* from Lake Mungo, Australia, illustrates (see **Fig. 2**) best how powerful the synthesis of fossil and genetic data is, to trace back the human ancestry. According to Satta's analysis, the three Neanderthals are genetically rather different from the ancient *Homo sapiens* from Lake Mungo which falls in the range of modern humans from Asia and Europe. Extant Africans are remarkably set apart, their mtDNA sequences are rich of variation and gave origin of all other humans. Asians and Europeans do not significantly differ, apparently due to repeated intercontinental gene flow.

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