HUMAN ORIGINS AND CONTINUING EVOLUTION IN AFRICA – THE PLACE OF THE KHOISAN

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There is overwhelming support for the origin of our species, *Homo sapien sapiens* in Africa. Both mitochondrial DNA (mtDNA) and Y chromosome DNA lineages are rooted in Africa with subsequent differentiation taking place within and outside the African continent.

MtDNA lineages found in African populations have been defined as haplogroup L which is further differentiated into three major sub-haplogroups: L1, L2 and L3. The frequencies and distribution of the mtDNA sub-haplogroups differ among the various sub-Saharan African populations[1].

Y chromosome studies making use of both biallelic and short tandem repeat markers have contributed in understanding how males have shaped the gene pool of African populations[2,3]. Haplogroups (HGs) E-M2 and E-M191 are the commonest HGs found among sub-Saharan African populations. The frequencies and distribution of haplotypes within these HGs supports a recent geographic expansion of both HGs, probably via the “Bantu Expansion”.

In the present study, we examine how evolution within Africa has contributed in shaping the gene pool of present-day Khoisan-speaking groups. It is generally accepted that southern Africa was occupied exclusively by the Khoisan prior to the arrival of Bantu-speaking people, approximately 2 000 years ago. Prehistoric and recent contact between groups speaking Khoisan and Bantu-languages have resulted in several cultural, linguistic and genetic exchanges among them. We make use of both mtDNA and Y chromosome DNA markers to examine the genetic affinities of Khoisan-speaking groups from southern Africa with other African populations.

[1] Salas *et al*. Am.J.Hum.Genet, 71:1082-1111, 2002.
[2] Cruciani *et al*. Am.J.Hum.Genet, 20:1197-1214, 2002
[3] Pereira *et al*. Ann. Hum. Genet, 66: 369-378, 2002