

MITOCHONDRIAL DNA DIVERSITY IN ETHIOPIA

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East African populations present the highest genetic diversity levels, namely for mitochondrial DNA (mtDNA). mtDNA databases must have a considerable size in order to be informative because the vast majority of haplotypes in a population are unique. Here we report the mtDNA diversity (for hypervariable regions I and II) in 78 individuals from Ethiopia. The quality criteria adopted in the forensic field were followed, namely performing sequencing in both directions (by using the kit Big-Dye Terminator Cycle Sequencing Ready Reaction and the automatic sequencer ABI 3100; AB Applied Biosystems), and carrying phylogenetic evaluation to check sequence edition. Sequence classification into haplogroup was done according to recent nomenclature recommendations. Molecular diversity indexes were calculated in Arlequin 3.0 and compared with published data on neighbour populations.

As expected, levels of diversity were high, attaining a value of 0.994 ± 0.004 for haplotype diversity in HVI, comparable with values in neighbouring populations: 0.993 ± 0.005 in Egypt (n=68); 0.977 ± 0.008 in Nubia (n=80); 0.989 ± 0.004 in Sudan; and 0.993 ± 0.001 in another Ethiopian sample (n=270). The random haplotype match probability in HVI was of 2.19% compared to 2.16% in Egypt, 3.54% in Nubia and 2.04% in Sudan.

The haplogroup distribution in Ethiopia was: 21.8% of Eurasian ancestry; 9.4% of the East African M1 lineage; and 64.1% of sub-Saharan affiliation. In the sub-Saharan pool, a proportion of 48.8% is represented by the haplogroup L3, the ancestor of the worldwide mtDNA diversity outside Africa. Given the recent interest in the alternative routes for Out-of-Africa migration(s) (Levant versus Southern), these L3 Ethiopian haplotypes will contribute information to shed light on this issue.