Human Mitochondrial DNA Sequence Variations in Korean Population

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From 136 unrelated Koreans, we have analyzed a nucleotide sequence variation of a 715 base pair region of the human mitochondrial DNA encompassing the D-loop. (16021-16400 as HV1, 66-400 as HV2). As a result, 92 and 45 mutation spots were detected in HV1 and HV2 respectively when compared with the Anderson sequence. 129 unique sequence haplotypes were defined, 7 haplotypes of which were sequences overlapped by more than one sample. Especially in the HV1 region, there were two spots which showed significant differences from the previously reported Caucasian database. They were 16223 and 16362, in which spot high C to T(>70%) and T to C(>50%) transition rates were observed respectively, while it proved to be common in most oriental populations. The average number of mutations were 4.6 and 5.0 in HV1 and HV2, respectively. The most frequent mutation type was transition (76% among the mutations) except that C base addition in 309 and 315 region of HV2 was observed frequently. The pedigree analysis showed no significant deviations between mother and child.

In 1997, an old Korean woman who had been a war comfort woman during World War 2 and lived in Cambodia for over 50 years has retrieved her nationality as a Korean by the comparison of mtDNA sequences with her alleged sister who has lived in Korea. In conclusion, it is considered that mtDNA sequencing could be applicable to human identification combined with conventional STR analysis.