Forensic Genetic Analysis of Mitochondrial DNA Hypervariable Region I/II Sequences: An Expanded Korean Population Database

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We have analyzed variation of the mitochondrial DNA (mtDNA) hypervariable segments I and II (HVS-I and HVS-II) in 185 randomly-chosen individuals from Korea to provide an expanded and reliable Korean database. Combined sequence comparison of HVS-I and HVS-II led to the identification of 167 different haplotypes characterized by 154 variable sites. One hundred and fiftyone of the haplotypes were individual-specific, 14 were found in two individuals and two were found in three individuals. A pairwise comparison of the 185 HVS-I/II sequences found an average of 10.11 ± 4.63 differences between individuals. The random match probability and gene diversity for the combined hypervariable regions were estimated at 0.66% and 0.9988, respectively. Analyzing the expanded database including three previously reported data sets and the present data using haplogroup-based comparisons and comparison with closely related sequences allowed errors to be detected and eliminated, thus considerably improving data quality. Sample division comparisons based on Φ_{ST} genetic distance measures revealed no significant population differentiation in the distribution of mtDNA sequence variations between the present data set and a database in The Scientific Working Group on DNA Analysis Methods (SWGDAM), but did indicate differences from other sets of data. Based on the results of mtDNA profiles, almost all of the mtDNA types studied here could be classified into subsets of haplogroups common in east Asia, and show that the Koreans possess lineages from both the southern and the northern haplogroup complexes of east Asian populations. The new data, combined with other mtDNA sequences, demonstrate how useful comparison with closely related mtDNA sequences can be for improving database quality, as well as providing haplotype information for forensic and population genetic analyses in the Korean population.

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