

P606

High-resolution mtDNA Haplogroups and their Implications for the Peopling of Korea

Wook Kim

Department of Biological Sciences, Dankook University, Cheonan 330-714

Studies of the early peopling of east Asia have long been a subject of interest in the field of human evolutionary history. Current hypotheses can be classified into two major models of the early migration routes into east Asia. The first model postulates that a southeast Asian origin is most likely, followed by a northward migration. Recent genetic surveys using autosomal microsatellite markers and Y-chromosomal binary markers supported this model. In contrast, the second model suggests a bi- and/or multidirectional route: one migration through central Asia and one through southeast Asia. Since Korea and Japan lie between the southeast and northeast Asian gene pools, their population genetic data can give us valuable information about the prehistoric migration route(s) and population expansions in east Asia. The Koreans are generally considered a northeast Asian group, since the Korean peninsula is bounded to the north by China, to the northeast by Russia, and to the south by the Korean Strait and Japan. The Korean peninsula and Japanese Archipelago were contiguous land before the rise in sea level between 10,000 and 7,000 years ago. Based on the results of archaeological data, the earliest modern human lithic cultures are found from 25,000 to 45,000 years ago in the Altai Mountains and southeastern Siberia as well as the Korean peninsula. Anthropological and/or archaeological evidence suggests that the early Korean population was related to Mongolian ethnic groups who inhabited the general area of the Altai Mountains in central Asia. Therefore, the ancient Koreans (proto-Koreans) may have shared a common origin with the northeast Asian groups who inhabited the general area of Altai Mountains and Lake Baikal regions of southeastern Siberia. There is also evidence for recent migration and range expansions via north China to Korea. According to Korea's founding myths, the Ancient Chosun (the first state-level society) was established around 2,333 BC in the region of southern Manchuria, but later moved into the Pyongyang area of northwest Korea. Recent northeastward migration from China, beginning in the 3rd century BC, led to the decline of the Ancient Chosun, with various Ancient States in the regions of southern Manchuria and the Korean peninsula emerging. In addition, archaeological evidence implies that rice cultivation had spread to all parts of the Korean peninsula by around 1,000 BC, introduced from the Yangtze River basin in southern China. Studies of classical genetic markers (protein and nuclear DNA) show that Koreans tend to have a close genetic affinity with Mongolians among northeast Asians. In contrast, genetic surveys of mitochondrial DNA (mtDNA) variation indicate that Koreans are more closely related to the Chinese and Japanese among east Asian populations. Recent studies of Y-chromosomal DNA markers show that the Koreans possess lineages from both northeast and southeast Asia. These results lead us to consider that the peopling of Korea is likely to have involved multiple events, and that different aspects revealed can be by different molecular genetic markers and DNA samples. Thus, further markers and DNAs from diverse regions of east Asia are required to explore the origin and genetic history of modern Koreans. We have analyzed mitochondrial DNA (mtDNA) sequence variation of the hypervariable segments I and II (HVS-I and HVS-II), and specific coding region sites in 185 unrelated Korean individuals to study the maternal lineage history and forensic application in the Korean population. Combined sequence comparison of the HVS-I (342 bp) and HVS-II (268 bp) represented the identification of 166 different haplotypes characterized by 156 variable sites, among which 149 were individual-specific, 15 were found in 2 individuals and 2 was found in 3 individuals. A pairwise comparison of the 185 sequences at the HVS-I/II region was found to be an average number of 10.26 ± 4.70 bp differences between sequences. The random match probability and the genetic diversity for the combined hypervariable regions were estimated at 0.66% and 0.9988, respectively. We also determined phylogenetic haplogroups by using the coding region and the HVS-I/II sequences from the same sample studied here. The major mtDNA expansion in Korea were haplogroup D4 and its sistergroups (D and its sublineages), although a large number of East Asian-prevalent haplogroups except for haplogroup E were widely but not evenly distributed in this population. The present survey also confirmed the occurrence of the population specificity of mtDNA lineages of haplogroups A5 and M7a1, which are found exclusively in Korea and Japan. Phylogenetic and Principal components analyses based on mtDNA haplogroup frequencies indicate that the Koreans have close affinities with southern/northern Han Chinese and Japanese populations, illustrating the southern and northern genetic contribution to the Koreans as well as the complexity of the population history for early settlement processes in East Asia.