S2-01

Y-chromosomal DNA haplogroups and their implications for the dual origins of the Koreans

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We have analyzed eight Y-chromosomal binary markers (YAP, RPS4Y711, M9, M175, LINE1, SRY+465, 47z, and M95) and three Y-STR markers (DYS390, DYS391, and DYS393) in 738 males from 11 ethnic groups in east Asia in order to study the male lineage history of Korea. Haplogroup DE-YAP was found at a high frequency only in Japan but was also present at low frequencies in northeast Asia, including 2.5% in Korea, suggesting a northern origin for these chromosomes. Haplogroup C-RPS4Y711 was present in Korea and Manchuria at moderate frequencies. The major Y-chromosomal expansions in east Asia were those of haplogroup O-M175 (and its sublineages). This haplogroup is likely to have originated in southern east Asia and subsequently expanded to all of east Asia. The moderate frequency of one sublineage in the Koreans, haplogroup O-LINE1 (12.5%), could be a result of interaction with Chinese populations. The age of another sublineage, haplogroup O-SRY+465, and Y-STR haplotype diversity, provide evidence for relatively recent male migration, originally from China, through Korea into Japan. In conclusion, the distribution pattern of Y-chromosomal haplogroups reveals the complex origin of the Koreans, resulting from genetic contributions involving the northern Asian settlement and range expansions mostly from southern-to-northern China.

S2-02

Genetic Analyses of Mitochondrial DNA from Northeast Asian Peoples

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The characteristics of mitochondrial DNA (mtDNA) - which include strictly maternal mode of inheritance, high copy number, lack of recombination, and high substitution rate - provide an excellent tool for evolutionary studies, such as reconstructing human origins, tracing population history, and analyzing ancient DNA. Recent reports have shown that the D-loop region, which have been used in most of the sequence analyses, had extreme variation in substitution rate between sites, and that the whole mtDNA genome, excluding the D-loop, can be used for more accurate measurement. Here, we report that we have sequenced the entire mitochondrial genome from Koreans and Khalkha Mongolians. The sequences were analyzed together with hypervariable regions of three different ethnic groups in Mongila, and 186 whole mitochondrial genome sequences from diverse populations, which is available in the public database. In addition, we have also sequenced the entire mitochondrial genome from 80 Korean patients with muscular dystrophies, in search for disease-associated mtDNA polymorphisms. The preliminary results will be discussed in this presentation.