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COMMENTARY

A commentary on assignment of Y-chromosomal SNPs found in Japanese population to Y-chromosomal haplogroup tree

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The binary polymorphisms on the human Y chromosome are useful uniparental markers not only for understanding human evolutionary histories but also for individualizing forensic biological materials.[1,2] In 2008, a monophyletic phylogenetic tree of the Y chromosome was extensively revised, containing 311 distinct haplogroups based on approximately 600 binary markers.[3] The haplogroups (branches) are defined by at least one binary polymorphism within the nonrecombining portion of the Y chromosome. New binary markers have been discovered in different populations, and have been assigned or re-assigned to upgrade the Y-chromosome haplogroup tree, improving the phylogenetic resolving power of the tree.[4,5]

In this issue of the journal, Naitoh *et al.*[6] investigated 121 binary polymorphisms found in a Japanese population of the Japanese Single-Nucleotide Polymorphisms (JSNP) database and five new additional non-JSNP markers found through the analysis of PCR products. They determined the phylogenetic positions of 14 binary markers that define the new terminal branches and several markers, which are phylogenetically equivalent to previously reported mutations. They assigned five JSNP markers to the Y-chromosome tree in a previous study,[7] and these markers were incorporated into the revised tree.[3] The authors have provided us with the most detailed phylogenetic tree of Japanese Y chromosomes and frequency data on the Y haplogroups.

The three clades C, D and O comprise the majority of the Japanese. The C1-M105 and D2-M55 lineages, associated with Jomon Y-chromosome lineages, are almost entirely restricted to the Japanese. The O2b-SRY465 lineages, associated with Yayoi lineages, are abundant in the Koreans and Japanese.[8,9] Almost all binary markers were assigned to one of the three clades in their study.[6] The C3*-M217 paragroup was divided into three haplogroups by two mutations. The D2a*-M116a paragroup (11.4%) was also trifurcated, and the frequency of D2a*-M116a and the new lineage D2a-022456 was 5.3%. The D2a1b-022457 lineage was subdivided, but the new haplogroup D2a1b-006841 was predominant. D2a1b-006841 and O2b-SRY465 accounted for 19.8% and 33.5% of 263 investigated Japanese chromosomes, respectively. These are the reasons why the value of haplogroup diversity increased only slightly from 86.2% for 18 haplogroups[7] to 87.5% for 32.[6] Although D2a1b-006841 and O2b-SRY465 are relatively young,[8,9] further efforts are needed to subdivide them.

Further studies of East Asian populations are also needed to establish the value of the newly assigned markers. The two mutations defining haplogroups C3-64562 + 13 and C3-2613-27 would be useful for the investigation of populations with C3*-M217. The detailed distribution of D2a*-M116a and D2a-022456 across the Japanese archipelago is important to clarify the diversity of the Jomon lineages and regional differences in the Japanese. In their study,[6] the binary markers were investigated using single-strand conformation polymorphism technique, which is tedious, laborious and time-consuming for routine analysis. A simple multiplex genotyping tool, for example, based on single-base primer extension technology,[10] should be developed. The application of such new methods may remove some markers, because they are located at recurrently mutated loci, mononucleotide repeats or multicopy loci. Finally, the phylogenetic resolving power of the Y-chromosome haplogroup tree presented in their study[6] will be enhanced by the incorporation of recently upgraded trees.[4,5]

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