

DNA ANALYSIS OF THE JAPANESE Y CHROMOSOME USING 7 BINARY MARKERS

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A simple Y chromosome haplogroup typing system using multiplex allele-specific PCR and capillary electrophoresis has been developed. To increase success rates for obtaining typing results from degraded DNA samples, PCR primers were designed to make PCR targets as short as ~100 bp. By simultaneous analyses of 7 Y chromosome binary markers (M130, M131, M57, M125, M175, M122 and M134), 1346 Japanese male individuals were divided into 7 Y-haplogroups i.e., C-M130, C-M131, D-M57, D-M125, O-M175, O-M122 and O-M134, and a "no mutation detected" group. Frequencies of C-M130, C-M131, D-M57, D-M125, O-M175, O-M122, O-M134, and the "no mutation detected" group were 0.0617, 0.0565, 0.1441, 0.182, 0.3418, 0.11, 0.0847 and 0.0193, respectively. No non-specific PCR amplification was observed from 155 Japanese female DNA samples. Y-haplogroup typing was successful in analyzing DNA mixture samples containing 1ng male DNA and 500ng female DNA. Y-haplogroup typing was also successful in analyzing bloodstain samples stored at ambient temperature for 23 years, indicating the ability of our system to analyze degraded DNA samples.

Samples from which no mutation was detected by simultaneous analyses of the 7 binary markers were further analyzed for M174, M214 and P36 by direct sequencing. These were found to belong to the D-haplogroup (M174 mutation positive), NO-haplogroup (M214 mutation positive) and Q-haplogroup (P36 mutation positive). Their frequencies in the 1346 Japanese male subjects were 0.0015, 0.0141 and 0.0037, respectively.