

Population Database and Mutation Study for Short Tandem Repeat Loci on Y-Chromosome (Y-STRs) in Japanese Populations

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ABSTRACT: A database for 14 short tandem repeat loci on Y-chromosome (Y-STRs) (DYS393, DYS19, DYS391, DYS437, DYS435, DYS439, DYS389II, DYS438, DYS436, DYS390, Y-GATA-H4, DYS385, DYS460, and DYS392) was constructed from 207 Honshu-Japanese (main island of Japan, Nagoya City) and 87 Okinawa-Japanese (southernmost islands of Japan) male DNA specimens. The samples were analyzed for 14 Y-STRs using two multiplex PCR typing systems, a newly devised 10-plex amplification system, and a commercially available Y-PLEX™ 6. The allele frequencies at 14 loci were calculated in each population and the distributions between both populations were significantly different at four of the 14 loci. A total of 244 unique haplotypes were observed in both Japanese populations, and 17 haplotypes were observed more than once. The two populations shared only seven haplotypes. The haplotype diversities for tested loci were 0.9987 and 0.9976 in Honshu- and Okinawa-Japanese, respectively. In the 10-plex, however, an extremely uninformative trinucleotide-repeat DYS436 was replaced with a more informative tetranucleotide-repeat locus, DYS389I, with a newly designed primer set to evolve modified 10-plex. With the modified 10-plex, since the number of unique haplotypes increased each one in both populations, the haplotype diversity values increased to 0.9988 and 0.9979 in Honshu- and Okinawa-Japanese, respectively. At present, in terms of the numbers of loci and individuals profiled, this database is the most informative in Japan. Only one instance of mutation was observed among the 51 Japanese father-son samples tested for the modified 14 loci. Haplotype analysis at 14 Y-STR loci, especially at the modified 14 Y-STR loci, would be more useful in forensic fields and in population genetics.

KEY WORDS: Y-Chromosome, database, Japanese, mutation, short tandem repeat.
