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Comparisons between Japanese and Han Chinese populations for 261 autosomal STR loci

T. Yamamoto *, K. Doi, R. Fukami, T. Yoshimoto, Y. Natori, A. Ishii

Department of Legal Medicine and Bioethics, Nagoya University, Japan

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ABSTRACT

In this study, Japanese and Han Chinese individuals (n = 32, each) were genotyped for 261 autosomal STRs, and allele frequencies were calculated for each locus in each population. The average number of alleles for all loci in Japanese and Han Chinese populations was 6.65 and 6.56, respectively. The tests for deviations from HWE performed using an exact test showed that the number of STRs ($P > 0.05$) in Japanese and Han Chinese populations was 236 and 241, respectively. Calculation of forensic parameters showed heterozygosity, and the exclusion means in the Japanese population were 0.7185 and 4813 and those in the Han Chinese population were 0.7308 and 0.5008. In addition, population genetic analyses, such as principal component analysis and factorial correspondence analysis, were performed and a differential formula with likelihood ratios was applied for various number of STR loci based on the effectiveness of differentiation between the two populations. Accordingly, this study suggests that statistical differentiation between genetically close populations, such as the Japanese and Han Chinese populations, is possible if approximately 40–50 effective STR loci are analyzed.

1. Introduction

Currently, ancestry informative markers, such as SNPs and microhaplotypes, are employed to trace the biogeographical ancestors of individuals. However, before determining these markers, we considered the possibility of simultaneously differentiating between individuals from genetically close populations using STR markers [1]. To do so, we must at least know the types of STRs that are effective and the number of STRs that are necessary. Therefore, over the past decade, we have collected basic information using more than one-fourth thousand autosomal STRs from 32 Japanese and Han Chinese individuals each. In this study, we provide preliminary information analyzed using genotype data.

2. Materials and methods

2.1. STR data

Japanese (Nagoya) and Chinese (Fujian) individuals (n = 32, each) were genotyped as sizing data for 269 autosomal STR loci, using multiplex amplification and multi-loading with a Genetic Analyzer 310. These data were rechecked from each electropherogram, and accurate data for 261 STRs from both populations were selected for this study.

2.2. Forensic statistics

Allele frequencies at each locus in both populations were calculated using GenAlEx 6.503. Two tests were performed to confirm deviations from the Hardy–Weinberg equilibrium (HWE) using GenAlEx 6.503 (χ^2 test) and Arlequin 3.5.2.2. (exact test) software. Forensic statistics, such as gene diversity, heterozygosity (Hobs), and power of exclusion, were calculated using STR Analysis for Forensics (STRAF) 1.0.5 software (http://cmpg.unibe.ch/shiny/STRAF/).

2.3. Population genetic analyses

Statistical differences in the allele frequency distributions at each locus between the Japanese and Chinese populations were confirmed using Arlequin 3.5.2.2. Genetic relative distributions for 64 individuals were constructed using principal component analysis (PCA) with STRAF 1.0.5 and factorial correspondence analysis (FCA) using Genetix 4.0.5.

* Corresponding author at: Department of Legal Medicine and Bioethics, Graduate School of Medicine, Nagoya University, 65 Tsurumai-cho, Showa-ku, Nagoya 466-8550 Japan.
E-mail address: yamachan@med.nagoya-u.ac.jp (T. Yamamoto).

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Table 1
Relationship between the numbers of deviations from HWE and P-values in the Japanese and Han Chinese populations and individually and both populations.

GenAlEx 6.503 software (χ^2 test)			Arlequin 3.5.2.2 software (Exact test)		
	Population (Pop)		Population (Pop)		
P-value	Japanese	Chinese	P-value	Japanese	Chinese
$P > 0.05$	236	241	$P > 0.05$	210	216
$P < 0.05$	16	14	$P < 0.05$	15	11
$P < 0.01$	7	4	$P < 0.01$	11	11
$P < 0.001$	2	2	$P < 0.001$	11	8
Total	261	261	Total	261	261
Both pops: $P > 0.05$	201		Both pops: $P > 0.05$		218
Both tests $P > 0.05$	185				

2.4. Differentiation using likelihood ratios

A trial to differentiate between the Japanese and Chinese populations using likelihood ratios was performed with various numbers of STRs.

3. Results and discussion

Allele frequencies at 261 autosomal STR loci were calculated for Japanese and Han Chinese individuals (32 each), and their average number of alleles for all loci was found to be 6.65 and 6.56, respectively. The χ^2 and exact tests were performed to confirm the deviation from HWE, and the results are summarized in Table 1. The number of STRs without significant deviation ($P > 0.05$) was 236 and 241 in the Japanese and Han Chinese populations, respectively. Forensic parameters were calculated using the STRAF 1.0.5 website, and the observed heterozygosity and the mean of exclusion were 0.7185 and 4813 in the Japanese population and 0.7308 and 0.5008 in the Chinese populations.

PCA performed for a population genetic analysis with the data for 261 STR loci in the Japanese and Chinese individuals showed mixed distributions at all 2D plots (PC1 vs PC2, PC1 vs PC3, and PC2 vs PC3). Therefore, we constructed seven triangle plots using PCA coordinates (PC1, PC2, and PC3) with various number of STR loci (261, 218, 201, 185, 44, 23, and 14) based on statistical tests in both populations. However, obtaining the results of differentiate between the Japanese and Chinese populations from these plots was difficult. Alternatively, FCA, another population genetic analysis method, was performed with the data at various numbers of STR loci (261, 218, 201, 185, 44, 23, and 14) in the same manner as PCA. The planar plots by FCA for more than 44 loci showed differentiation between the Japanese and Han Chinese individuals.

Finally, we calculated the likelihood ratios (LRs), which are more likely to be Japanese than Han Chinese, using the different allele frequencies between the Japanese and Chinese for each individual at all STRs. The LRs of all individuals at all loci were calculated, and then, after all LRs of each individual were multiplied, each multiplied value was transformed to a logarithmic value (called accumulated LRs). Fig. 1 shows the accumulated LR distribution by various number of STR loci based on statistical tests with normal distribution curves in both populations. Therefore, these distributions could possibly differentiate between the two populations.

Accordingly, this study suggests that statistical differentiation between genetically close populations, such as Japanese and Han Chinese, is possible if approximately 40–50 effective STR loci are analyzed.

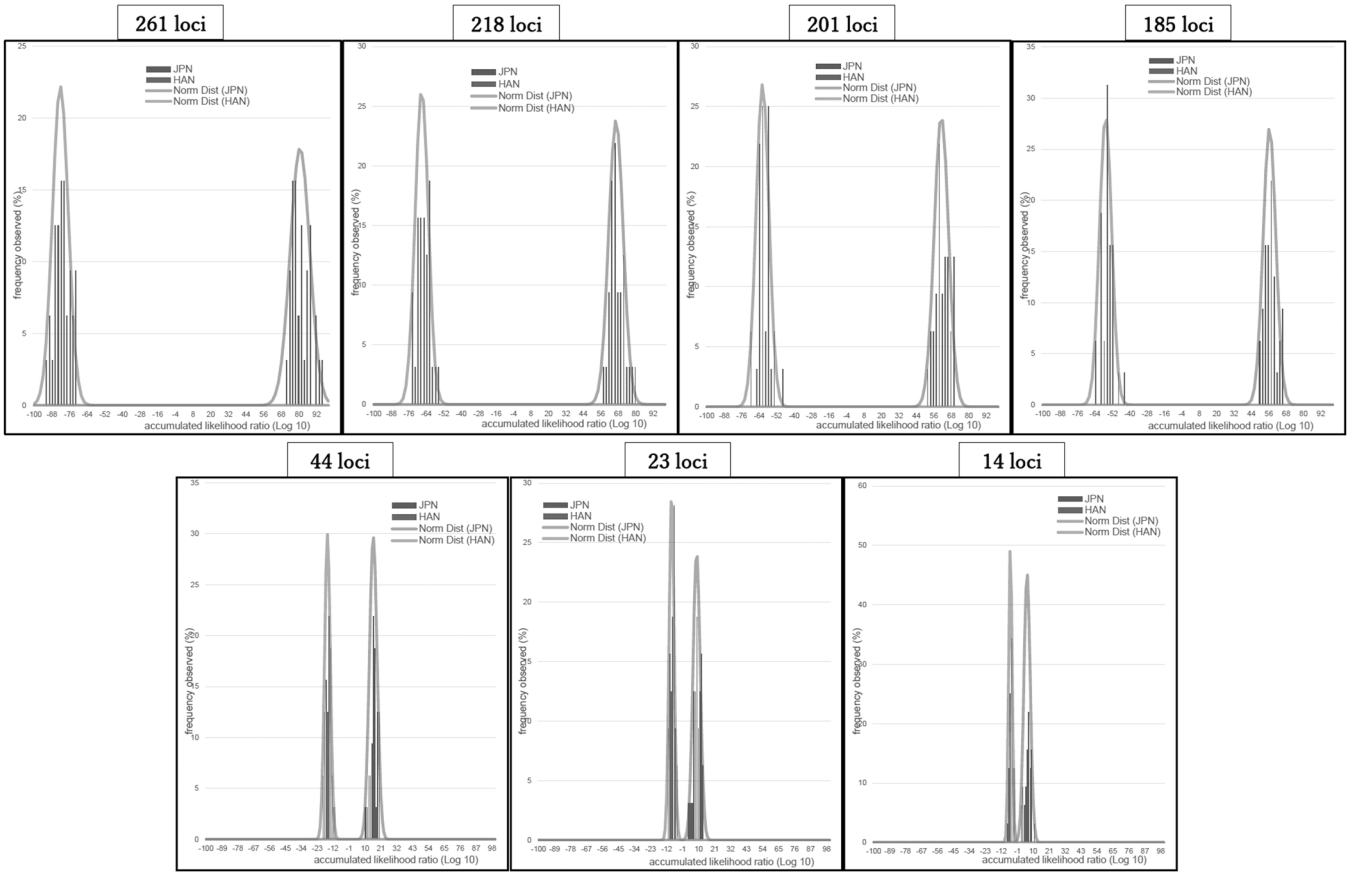

Fig. 1. Accumulated likelihood ratio distributions by various number of STR loci based on statistical tests with normal distribution curves in both populations (right peak, Japanese; left peak, Han Chinese).

Conflicts of interest

None.

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Reference

[1] T. Yamamoto, Y. Hiroshige, H. Ogawa, T. Yoshimoto, S. Mano, Potential statistical differentiation between Japanese and Korean populations with about 100 STRs, Forensic Sci. Int. Genet. Suppl. 5 (2015) e348–349.