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Differences in allele distribution at 15 STR loci among four Japanese regional populations

Toshimichi Yamamoto*, Naohiro Watanabe, Rieko Uchihi, Takashi Yoshimoto, Shi-Lin Li, Rina Kurihara, Yoshinao Katsumata

Department of Legal Medicine and Bioethics, Nagoya University Graduate School of Medicine, 65 Tsurumai-cho, Showa, Nagoya 466-8550, Japan

Abstract. We examined allele frequency distributions at 15 autosomal short tandem repeat (STR) loci including 13 Combined DNA Index System (CODIS) core STRs in four regional Japanese populations (Akita, Nagoya, Oita and Okinawa). Those distributions were compared pairwise and statistically among them. We also analyzed the distributions with published data on a Korean population by genetic distance D_A to construct a tree based on the neighbor-joining method. Consequently, we obtained one that coincides well with their geographical distributions.

Keywords: STR; Japanese; Genetic relationship

1. Introduction

Polymorphic tetranucleotide repeat short tandem repeat (STR) markers have been widely used for personal identification and paternity tests in forensic fields. It has been possible to make a large database for a large number of those loci conveniently and accurately using commercially released multiplex typing kits. Using more than 15 such markers, the allele frequency data in another Japanese population were published previously [1]. Since those STRs have higher mutation rates at meiosis than other DNA markers such as single nucleotide polymorphisms (SNPs), their allelic distributions have a potential to differentiate among geographically close populations. In the present study, the genotypes at 15 STR loci were analyzed using a commercially available multiplex typing kit among four regional populations (Okinawa, Oita, Nagoya and Akita) in Japan. Those allele frequencies at each locus were calculated, and their distributions were compared pairwise and statistically among their populations. Their genetic relationship was also examined by constructing a phylogenetic tree.

* Corresponding author. Tel.: +81-52-744-2119; fax: +81-52-744-2121.
E-mail address: yamachan@med.nagoya-u.ac.jp (T. Yamamoto).

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2. Materials and methods

2.1. DNA samples

Blood samples were collected from four regions (196 samples in Okinawa, 175 in Oita, 200 in Nagoya and 198 in Akita) in Japan. DNA were extracted from blood samples by a usual organic extraction method or with a QIAamp DNA Blood Mini kit (QIAGEN).

2.2. PCR amplification and typing

Those DNA samples were amplified at 15 STR loci using an AmpFlSTR Identifiler kit according to the manufacturer's instructions except a quarter volume of PCR reaction mixture. The PCR products were analyzed using a Genetic Analyzer 310 and genotyped.

2.3. Statistical analyses

Tests for Hardy–Weinberg equilibrium (HWE) were carried out using a homozygosity test, a likelihood ratio test and an exact test. Some statistical properties for discriminating power were calculated, and the differences in those distributions at 15 STR loci were examined statistically with Genepop software.

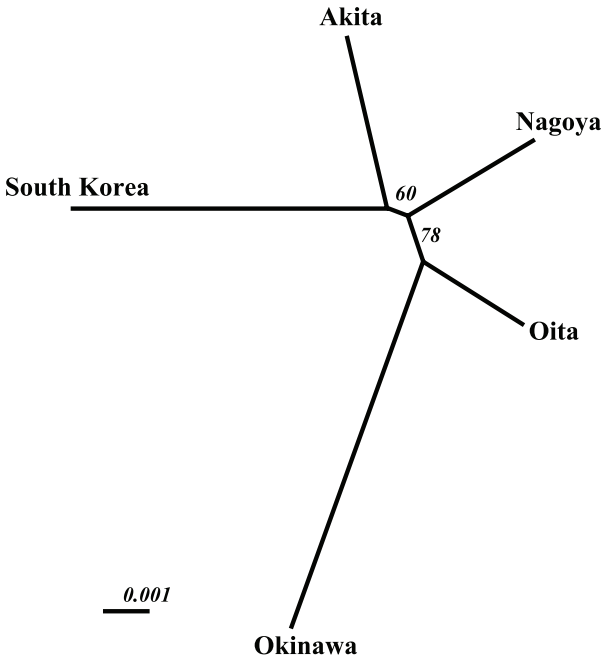

Fig. 1. Neighbor-joining tree based on Nei's D_A genetic distance for 15 STRs in four regional Japanese populations and a Korean population [2].

2.4. Constructing a phylogenetic tree

We calculated a Nei's D_A genetic distance with a Korean population [2] as an outer group, and constructed a tree based on the neighbor-joining method using neighbor-joining tree construction from allele frequency data (NJBAFD) software package.

3. Results and discussion

The allele frequencies at 15 STR loci were calculated in four regional Japanese populations. No significant deviations from HWE were observed using the three tests except a likelihood ratio test at D16S539 ($P=0.0335$) and homozygosity test at TPOX ($P=0.0217$) in Akita, a exact test at D18S51 ($P=0.0412$) in Oita, and homozygosity test at TH01 ($P=0.0117$) in Okinawa. However, these P-values are close to 0.05. The statistical properties for discriminating power in each population were also calculated. The allele frequency distributions were then compared pairwise and statistically, and the number of loci showing significant differences ($P<0.05$) were 5, 8, 4, 3, 1 and 2 between Akita and Okinawa, Nagoya and Okinawa, Oita and Okinawa, Akita and Oita, Nagoya and Oita, and Akita and Nagoya, respectively.

The neighbor-joining tree was constructed based on Nei's D_A genetic distance for 15 STRs in five populations including a Korean population [2] as an outer group (Fig. 1). Consequently, the genetic relationship in these five populations was very consistent with their geographical distribution. The genetic distances between the Okinawa population and the other Japanese populations were slightly farer than those between the Korean population and the other Japanese populations. The present study provides effective information to create a larger database in Japan, and on genetic relationships in/around Japan.

Acknowledgements

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