



Genetic relationships among East and Southeast Asian populations using 14 Y-STR markers

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Abstract. We analyzed the genotypes and haplotypes at 14 short tandem repeat (STR) loci on Y-chromosome (Y-STRs) in 10 East and Southeast Asian populations including two regional Japanese populations (Nagoya and Okinawa) using two multiplex typing systems. The allele frequency at each locus was calculated in each population and then compared among the populations. We also estimated the gene diversity at each locus and the haplotype diversity in each population. As a result, Japanese populations seemed to be genetically less diverse than other Asian populations. Furthermore, we constructed a tree based on the neighbor-joining method using a genetic distance D_A . The resultant tree was comprised of three clusters (Japanese, Chinese and Southeast Asian) and coincided well with their geographical distributions. © 2004 Elsevier B.V. All rights reserved.

Keywords: STR; Y-chromosome; Asian; Genetic relationship

1. Introduction

Short tandem repeats on Y-chromosome (Y-STRs) are generally used for male genotyping in sexual assault cases and sibship analyses. We reported previously on the haplotypes of Y-STRs in two regional Japanese populations [1,2]. Such haplotype analyses have also been utilized to examine relationships among various human populations [3]. We analyzed haplotypes at 14 Y-STRs (DYS389I, DYS439, DYS435, DYS19, DYS460, Y-GATA-H4, DYS391, DYS392, DYS438, DYS437, DYS393, DYS389II, DYS390, and DYS385) using two multiplex typing systems from DNA samples in two regional Japanese (Nagoya and Okinawa), six regional Chinese (Beijing, Shaanxi, Jiangsu, Hunan, Fujian, and Guangdong), one Thai (Bangkok), and one Burmese (Yangon) populations. In the present study, the allele frequencies at 14 Y-STR loci were calculated for each population and compared among those populations. Corresponding haplotype analyses

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were also performed, and their genetic relationships were examined by constructing a phylogenetic tree based on their allele frequencies at 14 Y-STR loci.

2. Materials and methods

2.1. DNA samples

Male blood samples were collected from two regions (207 in Nagoya and 87 in Okinawa) in Japan, six (77 in Beijing, 41 in Shaanxi, 58 in Hunan, 52 in Guangdong, 55 in Fujian, and 71 in Jiangsu) in China, one (119 in Bangkok) in Thailand, and one (91 in Yangon) in Burma. DNA samples were extracted using a usual organic extraction method or with a QIAamp DNA Blood Mini Kit (QIAGEN).

2.2. PCR amplification and typing

We analyzed a total of 14 Y-STRs with two multiplex PCR-based typing systems. Ten loci, DYS389I, DYS439, DYS435, DYS19, DYS460, Y-GATA-H4, DYS391, DYS392, DYS437, and DYS438 were analyzed with a 10plex system originally devised by Ruitberg and Butler, which we modified to change DYS436 to DYS389I with a newly designed primer set (Y-10plex). Six loci, DYS393, DYS19, DYS389II, DYS390, DYS391, and DYS385, were genotyped with Y-PLEX 6. Since DYS19 and DYS391 were included in both systems, a total of 14 Y-STRs were genotyped. The PCR products were analyzed with a Genetic Analyzer 310, and genotyped semi-automatically using Genotyper 2.5 software with the allelic ladder markers we made for Y-10plex.

2.3. Diversity estimates

The allele frequencies at 14 Y-STRs were calculated, and their haplotypes were aligned. The gene diversity at each locus and the haplotype diversity in each population were also estimated.

2.4. Constructing a phylogenetic tree

We calculated a Nei's D_A genetic distance, 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 haplotypes at 14 Y-STRs were analyzed using two multiplex typing systems, and the allele frequencies at 14 Y-STR loci were calculated in each population. Some alleles were apparently distributed differently at some loci between the populations. For example, the frequencies of the alleles 14 at DYS389I, 11 at DYS460 and 11 at DYS392 in the two Japanese populations were apparently higher than those in the other populations. On the other hand, the allele distributions at DYS391 did not seem to be diverse in all populations. The gene diversities were also estimated from these allele frequencies. Although the values at all loci were almost similar, those at DYS391, DYS437 and

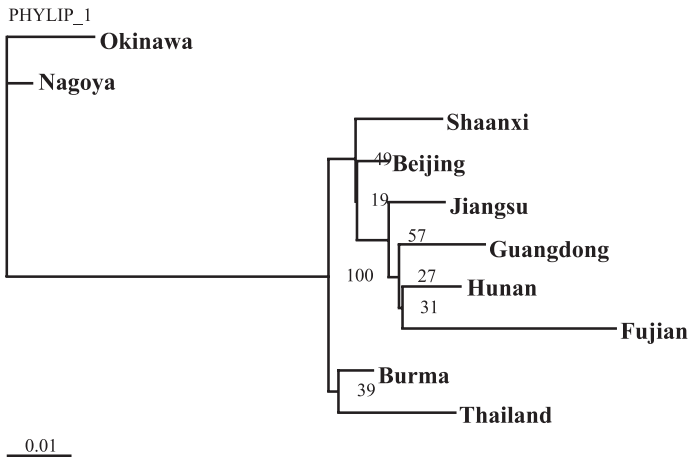


Fig. 1. Neighbor-joining tree based on Nei's D_A genetic distance for the allele frequencies at 14 Y-STRs in 10 East and Southeast Asian populations.

DYS435 in the two Japanese populations seemed to be slightly lower than those in the other populations. Furthermore, when we compared the haplotypes at 14 Y-STRs among all the populations, the haplotypes observed to be more than one in number in the two Japanese populations were not observed in the other Asian populations. Conversely, those in the other populations were not observed in the Japanese populations. The haplotype diversities in the Japanese populations were slightly lower than those in the other populations.

The neighbor-joining tree constructed based on Nei's D_A genetic distance for 14 Y-STRs showed three clusters, each comprised of a Japanese, Chinese and Southeast Asian population, and their genetic relationships coincided well with their geographical distribution as shown in Fig. 1. The genotype and haplotype analyses for Y-STRs such as those in the present study should provide valuable information on human genetic relationships.

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