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SHORT COMMUNICATION

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The polymorphisms of DYS388 and DYS392 on the Y chromosome in Japanese and German populations

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Abstract The analysis of a genotype survey in Japanese and German populations at the loci DYS388 and DYS392 located on the Y chromosome is reported. The gene diversities of DYS388 were 0.34 and 0.30 in the Japanese and German males, respectively, and six alleles were found in both groups. The gene diversities of DYS392 were 0.65 and 0.64 and the number of alleles was 8 and 9, respectively in the two populations. The distribution of DYS388 alleles in the Japanese population was different from the German population. The allele distribution of DYS392 showed significant differences among Asian populations.

Key words Trinucleotide repeat · Y chromosome · Population study

Introduction

In genetics, the sex chromosomes are especially useful for studies of human origins [7]. The short tandem repeats (STR) on the Y chromosome are very useful for sex identification as well as for individual identification and paternity testing [4]. In the present study the allele distributions of the polymorphic loci DYS388 (G00–365–729) and DYS392 (G00–456–509) were examined in Japanese and German populations.

Materials and methods

Blood samples were obtained from 200 Japanese and 111 German unrelated males. The protocol for DNA extraction [5], the primers [2, 6] and the polymerase chain reaction (PCR) amplifications were performed according to previous methods [2, 6]. For separation, 1 μl aliquot of PCR product was mixed with 0.5 μl of GeneScan Size Standard (GENESCAN-500 ROX) and 12 μl of deionized formamide. STR analysis was conducted with an ABI PRISM 311 Genetic Analyzer with Performance Optimized Polymer 4 (POP-4) and Genetic Analyzer capillaries. Each allele was confirmed by direct sequencing [6]. The gene diversity was analysed for informativeness of the STR locus on the Y chromosome [1, 3]. The allele frequencies of these loci were compared to those in other populations using the χ^2-test.

Results and discussion

DYS388

Genotyping of 200 Japanese and 111 German unrelated males revealed 6 distinct alleles in each population, dif-

Table 1 Allele frequency of DYS388 and DYS392 in Japanese and German populations

DYS388 number of repeats	Japanese	German
9	0.005	0.000
10	0.010	0.000
11	0.120	0.036
12	0.805	0.829
13	0.055	0.081
14	0.005	0.027
15	0.000	0.018
16	0.000	0.009
	$n = 200$	$n = 111$

χ^2-test in two populations $0.001 < p < 0.025$

DYS392 number of repeats	Japanese	German
7	0.005	0.009
8	0.005	0.009
9	0.000	0.000
10	0.085	0.018
11	0.530	0.523
12	0.145	0.108
13	0.205	0.279
14	0.020	0.027
15	0.000	0.018
16	0.005	0.009
	$n = 200$	$n = 111$

χ^2-test in the two populations $0.10 < p < 0.25$

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Table 2 The frequency of haplotypes of DYS388–DYS392 in Japanese and German populations

DYS388–DYS392 repeat number	Japan	Germany
9,11	0.005	0.000
10,10	0.005	0.000
10,13	0.005	0.000
11,10	0.010	0.000
11,11	0.070	0.018
11,12	0.015	0.009
11,13	0.020	0.009
11,14	0.005	0.000
12,70	0.005	0.009
12,80	0.005	0.009
12,10	0.065	0.018
12,11	0.425	0.414
12,12	0.125	0.090
12,13	0.165	0.243
12,14	0.010	0.018
12,15	0.005	0.018
12,16	0.005	0.009
13,10	0.005	0.000
13,11	0.025	0.045
13,12	0.005	0.009
13,13	0.015	0.018
13,14	0.005	0.009
14,11	0.005	0.018
14,13	0.000	0.009
15,11	0.000	0.018
16,11	0.000	0.009

fering in size by multiples of 3 bp (Tables 1 and 2). DNA sequencing revealed that these alleles were due to the number of ATT-repeats varying from 9–14 or 11–16 trinucleotides in the respective populations. The most common allele had a frequency of 0.805 in the Japanese and 0.829 in the German population and the gene diversities were 0.34 and 0.30, respectively. The allele distribution of DYS388 in the Japanese population was significantly different from that in Germans ($0.001 < p < 0.025$) or those reported by Kayser et al. [2] for Catalans ($0.005 < p < 0.01$) and for Basques ($0.01 < p < 0.025$), but the allele distributions did not differ between the European populations [2].

DYS392

In the Japanese and German populations, eight and nine alleles were observed respectively and the most common allele had a frequency of 0.530 and 0.523 (Tables 1 and 2). The gene diversity of this locus was 0.64 and 0.65, respectively. DNA sequencing revealed that these alleles were due to the number of ATT-repeats varying from 7–16 trinucleotides. The allele distributions of DYS392 were significantly different among populations, especially Japanese and Chinese ($p < 0.001$), Japanese and Mongolian ($p < 0.001$), Chinese and Mongolian ($p < 0.001$) [2].

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