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Genetic analysis of 18 STR loci on the X chromosome in a Japanese population

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Abstract

The allele frequencies of 18 short tandem repeat (STR) loci on the X chromosome have been analysed in 130 Japanese individuals living in Kanagawa by means of multiplex PCR and the ABI PRISM Linkage Mapping Set MD 10 Panel 28, followed by capillary electrophoresis using the ABI PRISM 310 genetic analyzer. Allele frequency profiles in this study were essentially the same as those for Caucasians. The theoretical heterozygosity levels of these STR loci, except for DXS1106 and DXS8055, ranged from 62.9% to 87.5%, and the means of the power of discrimination were estimated as 0.683 (male) and 0.848 (female). The combined resolutions of these 18 STR loci can be applied to routine casework.

Keywords: X chromosome; DNA polymorphism; STR; Population study; PCR

1. Introduction

Short tandem repeat (STR) polymorphisms are powerful tools for human identification, paternity analysis and genetic mapping. We have already analysed the AmpFLSTR Profiler Plus STR multiplex system to obtain allele frequency data for a Japanese population, and reported that this system was applicable to routine case work [1,2]. In the present investigation, we analysed the 18 STR loci DXS1227, DXS990, DXS986, DXS987, DXS993, DXS1073, DXS8091, DXS1106, DXS1047, DXS1001, DXS1068, DXS1214, DXS8055, DXS8051, DXS8043, DXS1060, DXS1226 and DXS991 to obtain allele frequency data for a Japanese population living in Kanagawa by means of multiplex PCR

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and the ABI PRISM Linkage Mapping Set MD 10 Panel 28 (PE Applied Biosystems), followed by capillary electrophoresis using the ABI PRISM 310 genetic analyzer.

2. Materials and methods

Blood samples were collected from 130 unrelated Japanese individuals living in Kanagawa, and genomic DNA was isolated using the Wizard Genomic DNA Purification Kit (Promega) from whole blood containing EDTA (1.5 mg/ml) as anticoagulant. Multiplex PCR amplification of these 18 STR loci was done using the ABI PRISM Linkage Mapping Set MD 10 Panel 28 according to the user's manual provided by the manufacturer [3]. The PCR products (1 μl) were mixed with 0.5 μl of GeneScan-400 HD [ROX] size standard (PE Applied Biosystems) and 12 μl of deionized formamide. These samples were denatured at 95 °C for 5 min, and electrophoresed using the ABI PRISM 310 Genetic Analyzer in a 47 cm, 50 μm i.d. capillary, filled with performance Optimized Polymer 4 (POP4, PE Applied Biosystems) at 15 kV for 24 min at 60 °C. The allele size determination and genotyping were performed using the GeneScan Analysis Software ver. 2.1. CEPH 1347-02 control DNA and CEPH Genotype Database were used as references for allele designation.

3. Results and discussion

The allele frequency distributions of 18 STR loci on the X chromosome have been analyzed in 130 Japanese individuals living in Kanagawa. The number of alleles and allele size ranges for each locus are summarized in Table 1. Six new rare alleles were detected in the Japanese population and were tentatively designated as DXS1227*7 (94 bp), DXS986*16 (147 bp), DXS993*9 (280 bp), DXS8091*10 (75 bp), DXS8051*13 (107 bp) and DXS1226*13 (275 bp). The most common alleles were 3 (53.8%) for DXS1227, 3 (41.6%) for DXS990, 4 and 11 (17.4%) for DXS986, 1 (43.7%) for DXS987, 3 (42.6%) for DXS993, 2 (47.2%) for DXS1073, 2 (54.3%) for DXS8091, 2 (78.7%) for DXS1106, 1

Table 1
The number of alleles and allele size ranges for the 18 STR loci on the X chromosome

Locus	Number of alleles observed for locus	Allele size ranges	Locus	Number of alleles observed for locus	Allele size ranges
DXS1227	7	80–92	DXS1001	7	187–207
DXS990	7	115–131	DXS1068	9	247–261
DXS986	14	147–183	DXS1214	8	283–299
DXS987	7	202–226	DXS8055	5	311–323
DXS993	8	270–290	DXS8051	9	99–129
DXS1073	11	306–332	DXS8043	7	147–169
DXS8091	8	67–89	DXS1060	10	241–259
DXS1106	5	123–131	DXS1226	12	279–303
DXS1047	11	148–170	DXS991	9	303–337

Table 2
Statistical properties of 18 STR loci in the Japanese population (n=130)

Locus	Heterozygosity	PIC	PD	
			Male	Female
DXS1227	0.629	0.579	0.624	0.813
DXS990	0.739	0.704	0.725	0.887
DXS986	0.875	0.862	0.875	0.967
DXS987	0.732	0.699	0.743	0.882
DXS993	0.678	0.632	0.710	0.832
DXS1073	0.655	0.598	0.678	0.810
DXS8091	0.662	0.567	0.612	0.869
DXS1106	0.365	0.346	0.230	0.635
DXS1047	0.838	0.819	0.802	0.958
DXS1001	0.797	0.767	0.794	0.925
DXS1068	0.767	0.735	0.751	0.915
DXS1214	0.784	0.756	0.760	0.921
DXS8055	0.178	0.170	0.205	0.298
DXS8051	0.718	0.672	0.680	0.929
DXS8043	0.709	0.653	0.711	0.848
DXS1060	0.854	0.837	0.830	0.962
DXS1226	0.857	0.841	0.854	0.980
DXS991	0.725	0.693	0.711	0.895

PIC: polymorphism information content, PD: power of discrimination.

(25.4%) for DXS1047, 1 (27.4%) for DXS1001, 1 (34.5%) for DXS1068, 1 (35.5%) for DXS1214, 2 (90.4%) for DXS8055, 3 (42.6%) for DXS8051, 1 (34.5%) for DXS8043, 1 (19.1%) for DXS1060, 9 (20.8%) for DXS1226 and 4 (45.2%) for DXS991. The distribution of genotypes of these 18 STR systems fitted the Hardy–Weinberg equilibrium ($0.50<P<0.95$). In general, a high number of alleles and high variability was observed, except for DXS1106 and DXS8055 loci, in the Japanese population. Allele frequency profiles in this study were essentially the same as those indicated in Caucasians obtained from the CEPH Genotype Database. In DXS8055 the allele frequency of DXS8055*2 obtained from this study was higher than that obtained from Caucasians. The heterozygosities, polymorphism information contents (PIC) and power of discrimination (PD) are given in Table 2. The theoretical heterozygosity levels of these STR loci, except for DXS1106 and DXS8055, ranged from 62.9% to 87.5%, the means of PD were estimated as 0.683 (male) and 0.848 (female). From these results, the 18 STR multiplex system on the X chromosome using the ABI PRISM Linkage Mapping Set MD 10 Panel 28 was applicable to routine casework.

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