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SHORT COMMUNICATION

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DXS10011: a hypervariable tetranucleotide STR polymorphism on the X chromosome

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Abstract The locus DXS10011 is a polymorphic system with a tetranucleotide repeat sequence located on the human X chromosome. The distribution of allele frequencies was examined in 334 Japanese and 171 German individuals and a total of 36 alleles was detected in the two population groups. This STR polymorphism will be a useful marker for linkage analysis.

Key words DXS10011 · X chromosome · STR polymorphism · Tetranucleotide repeat · Population data

Introduction

The short tandem repeat (STR) system DYS384 was first described by Gerken et al. (accession number in the DDBJ and GeneBank databases, L29968), and allele frequencies in a Japanese population have been published [1]. Recently, we showed that this STR system was located on the X chromosome [2] and the locus was renamed DXS10011 (Matsuki T, accession ID GDB:312393). Many STR loci on the Y chromosome have been studied [3] but data for the allele distribution of STR loci on the X chromosome are almost lacking.

In the present study, we report data for the allele frequencies for this STR system in Japanese and German individuals.

Materials and methods

EDTA blood samples were collected from 334 unrelated Japanese (212 male, 122 female) and 171 unrelated Germans (127 male, 44 female) for a population study. EDTA blood samples were also collected from 48 Japanese in 14 families for family studies. DNA was extracted using the phenol-chloroform method [4].

PCR primers

Primer 1 (GAAA strand):5′-GGAGTGAACTCTGAAAAAAAA-3′
Primer 2 (TTTC strand): 5′-TGAAATCATCTATCTTTCTTTC-3′

PCR conditions

PCR was performed in a total reaction volume of 10 μl containing 10 ng genomic DNA, 10 mM Tris-HCl buffer (pH 8.3), 50 mM KCl, 1.5 mM $MgCl_2$, 200 μM each dNTP, 4 pmol each primer and 0.5 U Taq polymerase (Takara, Ohtsu, Japan). Amplification was carried out for 32 cycles using a DNA Thermal Cycler Model 480 (Perkin-Elmer, Foster City, Calif.) with denaturation for 1 min at 94 °C, annealing for 30 s at 56 °C and extension for 30 s at 72 °C.

Analysis of PCR products

Electrophoresis was carried out on native polyacrylamide gels (8%T, 5%C, size 270 × 200 × 1 mm) in TBE buffer for 16 h at 240 V and bands were visualized by staining with SYBR Green I (Molecular

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Fig. 1 Sequences of regular and inter-alleles of DXS10011. The sequences in *bold type* correspond to the STR region

Regular alleles

– GAA **(GAAA)$_n$** GAAGGAAAG (GAAG) –

Inter-alleles

– GAA **(GAAA) GA (GAAA)$_6$ (GAGA) (GAAA)$_m$** GAAGGAAAG (GAAG)–

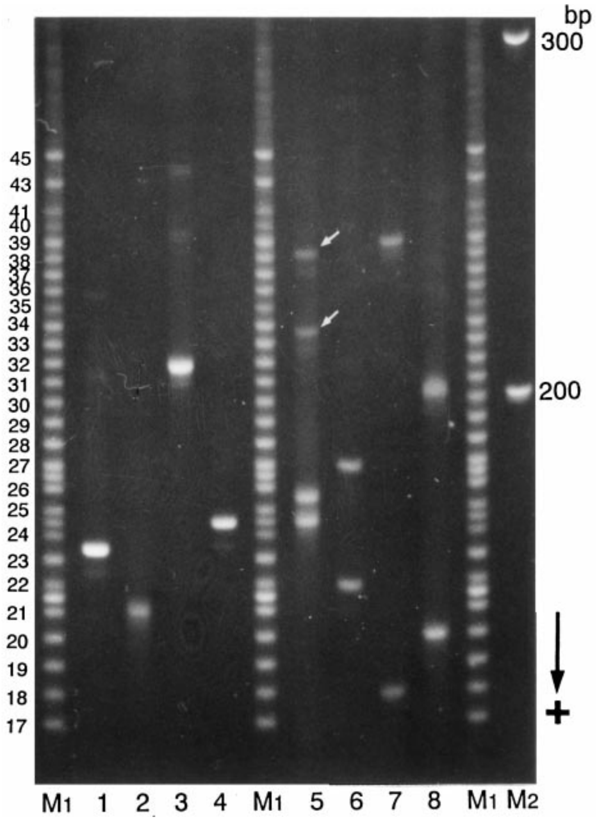

Fig. 2 Banding patterns of PCR products. *Lanes 1–4* male samples, *lanes 5–8* female samples; *lane 1* 23.2, *lane 2* 21, *lane 3* 32, *lane 4* 24.2, *lane 5* 24.2-25.2, *lane 6* 22-27, *lane 7* 18-39, *lane 8* 20-30.2, *lane M_1* allelic ladder marker, *lane M_2* DNA size marker (Superladder-Low, Gensura Laboratories, USA). *Arrows* indicate hetero-duplex bands

Probes, Rockland, ME). Some PCR products were directly sequenced using a Taq Dye Deoxy Terminator Cycle Sequencing Kit (Perkin-Elmer) with the ABI PRISM 310 Genetic Analyzer (Perkin-Elmer).

Results and discussion

In 671 chromosomes for the Japanese and German populations, 36 alleles including 8 inter-alleles were detected. The nucleotide sequences of regular alleles and the inter-alleles are shown in Fig. 1. Figure 2 shows that each allele was clearly and unambiguously distinguished by comparison with the corresponding allelic ladder band. The size and frequencies of the alleles are shown in Table 1. The frequency profiles are essentially similar in the male and female samples in the present study. The genetic transmission on the X chromosome was confirmed in 12 two-generation and 2 three-generation families. This STR system will be useful as genetic marker for linkage analysis and personal identification.

Table 1 Size and allele frequencies of the DXS10011 from 671 chromosomes

Allele	Size (bp)	Japanese	German
15	137	– –	1 (0.005)
17	145	3 (0.007)	– –
18	149	2 (0.004)	4 (0.019)
19	153	11 (0.024)	1 (0.005)
20	157	24 (0.053)	3 (0.014)
20.2	159	2 (0.004)	– –
21	161	20 (0.044)	4 (0.019)
21.2	163	9 (0.020)	6 (0.028)
22	165	26 (0.057)	1 (0.005)
22.2	167	9 (0.020)	14 (0.065)
23	169	29 (0.064)	6 (0.028)
23.2	171	15 (0.033)	19 (0.088)
24	173	22 (0.048)	6 (0.028)
24.2	175	40 (0.088)	8 (0.037)
25	177	15 (0.033)	7 (0.033)
25.2	179	12 (0.026)	3 (0.014)
26	181	29 (0.064)	14 (0.065)
26.2	183	4 (0.009)	3 (0.014)
27	185	24 (0.053)	8 (0.037)
28	189	29 (0.064)	14 (0.065)
29	193	33 (0.072)	24 (0.112)
30	197	28 (0.061)	17 (0.079)
30.2	199	1 (0.002)	– –
31	201	20 (0.044)	16 (0.074)
32	205	11 (0.024)	10 (0.047)
33	209	5 (0.011)	10 (0.047)
34	213	4 (0.009)	7 (0.033)
35	217	7 (0.015)	2 (0.009)
36	221	4 (0.009)	3 (0.014)
37	225	4 (0.009)	4 (0.019)
38	229	4 (0.009)	– –
39	233	2 (0.004)	– –
40	237	5 (0.011)	– –
41	241	1 (0.002)	– –
43	249	1 (0.002)	– –
45	257	1 (0.002)	– –
Total		456 (1.001)	215 (1.003)

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