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Population study of Y-chromosome STR haplotypes in Japanese from the Tokushima

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Abstract The Y-chromosome short tandem repeat (STR) systems including DYS19, DYS389I/II, DYS390, DYS391, DYS392, DYS393, DYS385 and YCAII were investigated in 121 Japanese males (Tokushima area). Analysis of the nine loci permitted classification of the samples into 107 haplotypes, and the haplotype diversity was estimated to be 0.989. DYS385 showed the highest gene diversity (GD) value (GD=0.946), while DYS391 showed the lowest GD value (GD=0.27). Furthermore, in DYS392, we found a new mutant allele that added a T within the repeat region (a mutation in the repetitive region), and the allele was designated 13.1 according to the sequence structure. We also detected allele 20 in DYS392 as a new allele. In DYS385, a new allele 26 that has not been previously reported was observed.

Keywords Y-chromosome STR · Mutation in the repetitive region · New allele · Japanese population · Tokushima

Introduction

Y-chromosome short tandem repeat (STR) polymorphisms are male-specific and therefore inherited from father to son.

Thus, STR polymorphisms are potential tools for personal identification in paternity tests for male infants and in sexual assault cases [1–3]. Recently, many papers providing worldwide population data for Y-STR have been published [4–7]. Moreover, the detection of an unknown allele and a mutation in the repetitive region of this sequence had been reported [8, 9]. This study generated and evaluated haplotype databases for nine Y-STRs, such as the minimal haplotypes (DYS19, DYS389I/II, DYS390, DYS391, DYS392, DYS393, DYS385) [10] and YCAII in Tokushima, Japan. When a mutation or new allele was suspected, the allele was confirmed by direct sequencing.

Materials and methods

Samples

Blood samples of 121 unrelated males were obtained from healthy volunteers in Tokushima, Japan. Genomic DNA was extracted according to the modified phenol–chloroform method [11].

Y-STRs examination

Coamplification of the loci was performed in PCR multiplexes: triplex I (DYS19, DYS385, YCAII), triplex II (DYS390, DYS391, DYS393), triplex III (DYS389I/II, DYS392) [12, 13]. Forward primers were 5'-end labeled with the following dyes: 5-Fam to triplex I, VIC to triplex II, NED to triplex III. PCR amplifications were performed on a thermocycler (Gene Amp PCR system 9700, Applied Biosystems, Weiterstadt, Germany).

PCR products were separated by electrophoresis on 4% acrylamide denaturing gels and ABI 310 Genetic Analyser (Applied Biosystems, Weiterstadt, Germany). Gene diversities (GD values) and haplotype diversities (HD values) were calculated according to Nei [14].

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Table 1 Extended Y-chromosomal STR haplotypes found among untreated males from a Japanese population ($n=121$)

Haplotype	DYS19	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393	DYS385	YCAII	Number
1	13	12	29	24	10	13	13	12/15	19/19	1
2	13	13	29	25	10	11	16	13/14	19/19	1
3	13	13	29	24	10	11	14	14/14	18/19	1
4	13	13	29	24	10	11	15	13/14	19/19	1
5	13	13	29	24	10	11	14	13/14	19/19	2
6	13	13	29	24	10	11	14	13/16	19/19	1
7	13	13	29	24	10	11	14	13/15	19/19	1
8	13	13	30	24	10	11	14	13/14	19/19	2
9	13	13	30	24	10	11	13	13/17	19/19	1
10	13	14	30	24	11	11	14	14/14	19/19	1
11	13	14	30	25	6	14	14	16/20	19/23	1
12	13	14	31	24	11	11	13	13/17	19/21	1
13	14	12	27	23	10	14	12	13/19	19/20	1
14	14	12	28	25	10	14	12	13/19	19/19	1
15	14	12	28	25	10	11	12	13/19	19/19	1
16	14	13	29	22	10	13	13	10/20	19/23	1
17	14	13	29	23	10	13	14	12/14	18/19	1
18	14	14	32	23	10	14	13	11/12	19/19	1
19	15	12	28	24	10	20 ^a	14	12/13	19/20	1
20	15	12	28	23	10	13.1 ^b	12	13/17	19/22	1
21	15	12	28	24	10	13	12	12/19	19/22	1
22	15	12	29	25	10	13	12	12/18	18/21	1
23	15	12	29	25	10	11	13	12/14	19/21	2
24	15	12	29	23	10	12	13	12/17	22/22	1
25	15	12	29	23	11	12	12	13/16	19/22	1
26	15	12	29	25	10	13	12	14/19	19/23	1
27	15	13	28	22	10	13	13	10/21	19/23	1
28	15	13	29	22	10	13	12	11/15	19/22	1
29	15	13	29	22	10	13	13	11/18	19/23	1
30	15	13	29	21	10	11	13	14/14	20/21	1
31	15	13	30	23	10	11	14	11/19	22/23	1
32	15	13	30	22	10	13	12	10/19	19/23	1
33	15	13	30	22	10	13	13	10/20	19/24	1
34	15	13	30	24	10	11	13	12/12	18/19	1
35	15	13	30	25	10	11	13	14/17	19/21	1
36	15	13	31	24	10	13	11	13/26 ^a	19/22	1
37	15	13	32	25	10	11	13	12/14	19/21	1
38	15	14	29	22	10	13	13	10/20	19/23	1
39	15	14	29	21	10	13	13	10/20	19/23	1
40	15	14	29	23	10	13	13	10/18	19/25	1
41	15	14	29	22	11	15	13	11/20	19/23	1
42	15	14	29	23	10	13	13	10/19	19/23	1
43	15	14	30	23	10	13	13	10/20	19/23	1
44	15	14	30	22	10	13	13	10/20	19/23	3
45	15	14	30	24	11	11	13	13/18	19/21	1
46	15	14	30	22	11	13	13	10/18	19/23	1
47	15	14	30	23	10	11	14	11/17	23/24	1
48	15	14	30	23	10	11	14	11/19	22/23	1
49	15	14	30	23	10	14	13	10/20	19/22	1
50	15	14	30	23	10	12	13	12/19	19/22	1
51	15	14	30	22	11	13	13	10/20	19/23	1
52	15	14	31	26	10	11	12	12/17	20/21	1
53	15	14	31	25	11	11	13	13/17	19/21	1
54	15	14	31	22	10	13	13	10/21	19/23	1

Table 1 (continued)

Haplotype	DYS19	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393	DYS385	YCAII	Number
55	15	14	31	24	10	11	13	13/17	19/20	1
56	15	14	31	22	10	13	13	10/19	19/23	1
57	15	14	32	25	10	13	15	13/19	19/22	1
58	15	14	32	25	11	11	13	13/14	19/22	2
59	15	15	31	22	10	13	13	10/20	19/23	2
60	16	12	28	23	10	13	12	13/20	18/22	1
61	16	12	28	25	11	13	12	14/16	19/23	1
62	16	12	28	23	10	12	13	13/18	22/22	1
63	16	13	28	23	10	13	13	10/18	19/23	4
64	16	13	28	23	10	13	13	11/17	19/23	1
65	16	13	28	22	10	14	12	11/11	19/22	1
66	16	13	29	25	9	11	13	13/19	19/21	1
67	16	13	29	25	10	13	12	13/18	19/23	1
68	16	13	30	25	10	11	13	13/17	19/22	1
69	16	13	31	25	10	11	13	13/17	19/22	1
70	16	13	31	25	10	11	13	12/14	19/21	1
71	16	14	28	23	10	13	13	10/19	19/21	1
72	16	14	29	23	10	13	13	10/18	19/23	1
73	16	14	29	23	11	13	13	10/18	19/23	1
74	16	14	29	23	11	13	14	10/18	19/23	1
75	16	14	29	24	10	13	13	10/17	19/23	2
76	16	14	29	23	10	13	13	10/17	19/23	3
77	16	14	29	23	10	13	13	10/20	19/23	1
78	16	14	29	22	10	13	13	10/21	19/23	1
79	16	14	29	24	10	13	13	10/18	19/23	1
80	16	14	29	23	10	14	13	10/18	19/23	1
81	16	14	29	23	10	14	13	10/16	19/23	1
82	16	14	29	23	10	13	13	11/18	19/23	1
83	16	14	30	23	10	12	12	12/16	19/22	1
84	16	14	30	23	10	13	13	10/18	18/23	1
85	16	14	30	23	10	13	13	10/18	19/23	1
86	16	14	30	23	10	11	14	11/17	23/24	1
87	16	14	30	22	10	12	15	11/19	22/22	1
88	16	14	31	25	10	11	13	13/18	19/22	1
89	17	12	28	24	10	13	12	14/22	18/18	1
90	17	12	28	24	10	13	12	12/19	19/23	1
91	17	12	29	25	10	13	12	12/20	19/23	2
92	17	12	29	25	10	13	12	12/18	19/23	1
93	17	13	28	23	10	11	15	11/21	22/22	1
94	17	13	29	23	10	13	12	14/22	23/23	1
95	17	13	30	24	11	11	13	13/17	19/22	1
96	17	13	30	26	10	11	13	13/18	19/21	1
97	17	13	30	25	10	11	13	15/19	19/21	1
98	17	13	30	25	11	11	13	13/18	19/21	1
99	17	13	31	25	10	11	13	13/17	19/22	1
100	17	14	30	25	10	11	13	15/17	19/22	1
101	17	14	30	24	11	11	12	13/18	19/21	1
102	17	14	31	25	10	11	13	13/16	19/22	1
103	17	14	31	25	10	11	13	13/17	19/22	1
104	17	14	31	25	11	11	13	13/17	19/21	1
105	17	14	31	24	10	11	13	13/19	19/21	1
106	17	14	31	25	10	11	13	13/17	22/22	1
107	17	14	31	26	10	11	13	13/20	19/21	1

Haplotype diversity: 0.989

^aNew allele^bRepeat-based mutation

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