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SHORT COMMUNICATION

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Haplotype frequencies of nine Y-chromosome STR loci in the Taiwanese Han population

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Abstract Haplotype frequencies of nine Y-chromosome STR loci (DYS19, DYS385, DYS388, DYS389I, DYS389II, DYS390, DYS391, DYS392 and DYS393) in the Taiwanese Han population were established. A total of 183 unrelated individuals produced 162 haplotypes, of which 146 were unique, 1 was found in 5 individuals, 2 were found in 3 individuals and 13 were found in 2 individuals. The haplotype diversity (99.99%) and discrimination capacity (88.5%) were calculated. A family study of 109 father/son pairs in 100 families showed 2 mutational events in the DYS389II locus and 1 in the DYS392 locus.

Keywords STR polymorphism · Y-STR · Population genetics · Taiwanese

Introduction

Y-chromosome STRs are used in forensic analysis for a number of reasons: most of the Y chromosome is recombination-free during meiosis, they are inherited paternally and transferred unchanged from generation to generation, unless mutations occur. This is most useful in parentage testing especially in deficiency cases and male offender identification in male/female DNA mixtures of sexual assault cases [1, 2]. They can also be used in human evolution, genealogical and population studies [3, 4, 5]. In this study, nine Y-STR loci (DYS19, DYS385, DYS388, DYS389I, DYS389II, DYS390, DYS391, DYS392 and DYS393) were screened to establish haplotype data for the Taiwanese Han population. The Taiwanese Han population is comprised of individuals who migrated from mainland China in a number of waves of immigration, the earliest being approximately 400 years ago from the Fukien and Kwangtung provinces and the descendants of these people now constitute approximately 86% of the population [6]. In 1949 there was a large-scale migration from all 35 mainland provincial areas. People derived from this migration now constitute approximately 13% of the population [6]. There is a large degree of admixture between these two groups of Han population.

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Materials and methods

Population samples

Blood samples of 183 unrelated individuals and 109 father/son pairs in 100 families of the Taiwanese Han population were collected. DNA was extracted by the salt-chloroform method [7] and quantified by the QuantiBlot kit (Roche Molecular Systems, Alameda, Calif.).

PCR amplification

PCR amplifications were performed in one quadruplex, one triplex and two singleplex reactions. The loci used in the quadruplex were DYS19, DYS389I/II and DYS390, for the triplex DYS391, DYS392 and DYS393, and the single loci DYS385 and DYS388. All primers used were those described by Kayser et al.[1] and one of each pair was fluorescently labeled and different fluorescent dyes were used in the quadruplex and triplex amplifications. PCR amplifications were performed in a 25 μl reaction mixture which contained 20 ng of extracted genomic DNA, reaction buffer (10 mM Tris-HCl, pH 8.3, 2.5 mM $MgCl_2$, 50 mM KCl, 0.01% gelatin), 1 unit of AmpliTaq Gold DNA polymerase (Applied Biosystems) and 0.06–0.3 μM of each primer. The PCR amplifications were all carried out using the programme of 95 °C for 10 min followed by 40 cycles of 94 °C for 1 min, 55 °C for 1 min and 72 °C for 2 min in a thermal cycler.

Table 1 A list of the 162 haplotypes found in 183 unrelated individuals in the Taiwanese Han population

Haplotype No.	DYS19	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393	DYS385	DYS388	N
1	13	9	25	24	11	14	12	13–14	10	1
2	13	9	25	25	10	13	12	12–18	12	1
3	13	9	27	23	10	12	12	12–16	12	1
4	13	10	26	24	9	14	13	15–22	12	1
5	13	11	27	24	9	14	14	15–21	12	1
6	14	9	24	23	10	14	12	13–22	10	1
7	14	9	24	24	9	14	13	11–13	12	1
8	14	9	24	25	10	13	12	13–20	12	1
9	14	9	24	25	11	14	12	13–19	10	2
10	14	9	25	22	10	14	12	15–18	10	2
11	14	9	25	23	10	12	12	11–16	12	1
12	14	9	25	23	10	13	12	16–18	10	1
13	14	9	25	23	10	14	12	15–15	10	1
14	14	9	25	23	10	14	12	15–18	10	1
15	14	9	25	23	10	14	12	15–19	10	2
16	14	9	25	24	10	14	12	13–17	10	1
17	14	9	25	24	10	14	12	13–18	10	1
18	14	9	25	24	10	14	12	13–19	10	2
19	14	9	25	24	10	14	12	13–20	10	1
20	14	9	25	24	10	14	12	14–17	10	1
21	14	9	25	24	10	14	12	14–18	10	1
22	14	9	25	24	10	14	12	14–19	10	1
23	14	9	25	24	11	14	13	14–18	10	1
24	14	9	25	25	10	14	12	13–17	10	2
25	14	9	25	25	10	14	12	13–18	10	1
26	14	9	26	22	10	14	12	15–18	12	1
27	14	9	26	23	11	14	12	15–17	10	1
28	14	9	26	23	12	14	13	13–13	12	1
29	14	9	26	24	10	13	12	13–18	10	1
30	14	10	26	23	10	11	15	11–18	13	1
31	14	10	26	23	10	14	12	13–18	10	1
32	14	10	26	23	10	14	13	13–13	12	1
33	14	10	26	23	10	15	13	9–13	12	1
34	14	10	26	24	10	11	12	13–16	14	1
35	14	10	27	23	11	12	12	12–12	12	1
36	14	10	27	24	10	14	12	13–18	10	1
37	14	10	28	24	10	13	12	14–21	12	1
38	14	11	26	25	11	14	12	14–19	10	1
39	14	11	27	25	11	13	12	13–19	12	1
40	14	11	27	26	11	13	12	13–18	12	1
41	14	11	28	23	10	14	13	11–12	13	1
42	15	8	24	22	9	14	13	13–13	12	1
43	15	9	24	23	10	12	12	12–16	12	1
44	15	9	24	23	10	14	12	13–13	12	1
45	15	9	24	23	10	14	13	13–13	12	5
46	15	9	24	25	10	13	12	12–19	12	1
47	15	9	24	26	10	13	12	12–19	12	1
48	15	9	25	21	10	11	12	11–17	13	1
49	15	9	25	22	10	14	13	13–14	12	1
50	15	9	25	23	10	13	12	12–18	11	1
51	15	9	25	23	10	13	12	12–19	10	1
52	15	9	25	23	10	13	14	12–14	12	1
53	15	9	25	23	10	14	12	12–13	10	1
54	15	9	25	23	10	14	12	13–13	12	1
55	15	9	25	23	10	14	13	12–12	12	1
56	15	9	25	23	10	14	13	12–13	12	2
57	15	9	25	23	10	14	13	12–14	12	1

Table 1 (continued)

Haplotype No.	DYS19	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393	DYS385	DYS388	N
58	15	9	25	23	11	12	12	12–17	12	1
59	15	9	25	23	11	13	13	13–13	12	1
60	15	9	25	23	11	14	13	13–13	12	2
61	15	9	25	23	11	14	13	13–14	12	1
62	15	9	25	24	10	13	12	13–21	12	1
63	15	9	25	24	10	13	13	13–20	12	1
64	15	9	25	24	10	13	15	14–17	12	1
65	15	9	25	24	10	14	12	14–18	10	3
66	15	9	25	24	10	14	12	15–18	10	1
67	15	9	25	24	10	14	12	15–19	10	1
68	15	9	25	24	11	13	14	13–16	13	1
69	15	9	25	24	11	14	12	14–18	10	1
70	15	9	25	25	10	13	12	12–19	12	2
71	15	9	26	23	10	14	13	12–13	13	1
72	15	9	26	23	10	14	13	13–13	12	1
73	15	9	26	23	10	14	14	13–13	12	1
74	15	9	26	23	11	13	12	12–20	12	1
75	15	9	26	23	11	14	13	13–13	12	2
76	15	9	26	23	11	14	13	13–14	12	1
77	15	9	26	23	11	14	13	13–15	12	1
78	15	9	26	23	11	14	14	13–13	12	1
79	15	9	26	23	12	14	13	13–13	12	1
80	15	9	26	24	11	13	13	12–19	12	1
81	15	9	26	24	11	14	13	12–13	12	1
82	15	9	26	25	10	14	12	14–17	12	1
83	15	9	27	23	10	12	12	12–12	12	1
84	15	9	27	23	10	12	12	12–17	12	1
85	15	9	27	25	9	13	12	12–19	12	1
86	15	10	25	23	10	13	14	12–18	12	1
87	15	10	26	22	11	13	14	12–21	12	1
88	15	10	26	23	9	15	14	12–18	12	1
89	15	10	26	23	10	11	15	12–17	13	1
90	15	10	26	23	10	13	14	12–20	12	1
91	15	10	26	23	10	14	12	15–19	10	1
92	15	10	26	23	11	11	15	11–18	13	1
93	15	10	26	23	11	14	13	13–13	12	1
94	15	10	26	24	10	13	12	13–20	12	1
95	15	10	26	24	10	13	12	13–21	12	2
96	15	10	26	24	10	13	13	12–17	12	1
97	15	10	26	25	10	11	13	12–13	13	1
98	15	10	26	25	11	14	12	13–21	12	1
99	15	10	27	22	10	14	12	13–15	12	1
100	15	10	27	22	10	14	13	14–14	12	1
101	15	10	27	22	10	15	13	11–14	12	1
102	15	10	27	23	10	11	13	11–18	13	1
103	15	10	27	23	10	13	12	12–19	12	1
104	15	10	27	24	10	14	12	12–17	12	1
105	15	10	27	25	10	13	12	13–19	12	1
106	15	10	27	25	10	14	12	12–17	12	1
107	15	10	27	25	11	11	13	12–13	14	1
108	15	10	28	23	10	13	12	12–19	12	1
109	15	10	28	23	10	13	14	14–20	12	1
110	15	10	28	23	10	14	12	12–19	12	1
111	15	11	26	24	10	13	14	11–19	12	1
112	15	11	27	23	10	11	16	11–19	13	1
113	15	11	27	23	11	14	13	11–12	13	1
114	15	11	27	24	10	13	12	13–20	12	1

Table 1 (continued)

Haplotype No.	DYS19	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393	DYS385	DYS388	N
115	15	11	27	24	10	13	13	14–20	12	1
116	15	11	27	24	10	13	14	12–17	12	1
117	15	11	27	24	10	13	15	13–18	12	2
118	15	11	27	25	10	13	14	13–19	12	2
119	15	11	27	25	11	13	14	14–20	12	1
120	15	11	28	25	10	13	14	14–20	12	1
121	15	11	29	23	10	13	12	12–19	12	1
122	16	9	24	23	10	14	13	13–13	12	1
123	16	9	24	23	10	14	13	13–14	12	1
124	16	9	24	23	11	12	12	12–15	12	1
125	16	9	25	23	10	13	12	11–19	12	1
126	16	9	25	23	10	14	13	13–13	12	1
127	16	9	25	24	11	13	11	14–18	12	1
128	16	9	25	25	10	13	12	12–17	12	1
129	16	9	25	25	10	13	12	12–19	12	1
130	16	9	25	25	10	13	12	12–20	12	1
131	16	9	25	25	10	13	12	13–21	12	1
132	16	9	25	25	10	15	12	12–20	12	1
133	16	9	25	26	11	13	12	11–20	12	1
134	16	9	26	23	11	14	13	13–13	12	1
135	16	9	26	24	10	13	12	13–20	12	1
136	16	9	27	25	10	13	12	12–19	12	2
137	16	9	29	25	10	13	12	12–19	12	1
138	16	10	25	24	10	13	13	13–17	12	1
139	16	10	25	25	10	13	12	12–12	12	1
140	16	10	26	23	10	11	14	11–17	13	1
141	16	10	26	23	10	13	12	11–18	12	1
142	16	10	26	24	10	11	14	11–17	13	1
143	16	10	26	24	10	13	12	12–16	12	1
144	16	10	26	25	10	13	12	12–20	12	1
145	16	10	27	23	10	11	14	11–19	13	1
146	16	10	27	23	10	12	11	12–16	12	1
147	16	10	27	24	10	13	12	12–18	12	1
148	16	10	27	24	11	13	14	13–18	12	1
149	16	10	28	23	11	14	13	13–14	12	1
150	16	10	28	24	10	13	12	12–19	12	1
151	16	11	27	24	10	13	12	12–18	12	1
152	16	11	27	24	10	13	14	12–18	12	3
153	16	11	27	24	10	13	14	12–19	12	1
154	16	11	27	24	11	13	12	12–12	12	1
155	17	9	25	23	11	14	12	13–18	10	1
156	17	9	25	24	10	13	12	13–20	12	1
157	17	9	26	24	11	13	12	12–18	12	1
158	17	10	25	24	10	13	13	13–17	12	1
159	17	10	25	24	10	13	13	13–18	12	1
160	17	10	26	24	10	13	12	12–17	12	1
161	17	10	27	23	10	13	12	12–19	12	1
162	17	10	27	25	10	13	12	13–19	12	1

Electrophoresis of PCR products

PCR products were separated and detected in 4% denatured polyacrylamide gels and with ROX500 (Applied Biosystems) as internal standard on a PRISM 377 DNA sequencer (Applied Biosystems). PCR products of the two singleplex reactions were loaded together in one lane, because they were labeled with different fluorescent dyes.

Designation of alleles

Every allele fragment with the same size of PCR products in each locus was amplified by single locus PCR with non-labeled primer pairs. These PCR products were then separated on 3% low melting agarose gels, sliced out and purified by the alcohol precipitation method. The purified PCR products were sequenced by cycle sequencing using fluorescent dideoxynucleotides in the BigDye Ter-

minator Cycle Sequencing Ready Reaction kit (PE Applied Biosystems, Foster City, Calif.). The two primers used for PCR amplification were used as sequencing primers for both side extensions. The cycle sequencing products were separated by 5% denatured Long Ranger gels (FMC BioProducts, Rockland, Me.) and detected using a PE Applied Biosystems 373A DNA sequencer. The nomenclature of the alleles was determined by the number of repeat units presented in sequencing results.

Statistical methods

The haplotype diversity was calculated according to Nei [8] and the discrimination capacity was the percentage proportion of the different haplotypes.

Results and discussion

Haplotypes of the 9 Y-STR loci according to the nomenclature of number of repeat unit observed in sequencing results are shown in Table 1 and 162 haplotypes were found in 183 unrelated individuals in the Taiwanese Han population. Among these haplotypes, 146 were unique, 1 was found in 5 individuals, 2 were found in 3 individuals and 13 were found in 2 individuals. The haplotype diversity was calculated as 99.99% with a discrimination capacity of 88.5%.

A family study of 109 father/son pairs in 100 families showed 2 mutational events at the locus DYS389II (from allele 26 to 27) and 1 in DYS392 locus (from allele 13 to 14). The alleles of mutation father/son pairs were sequenced to confirm the mutation events. The paternity was confirmed by the paternity index value of 8.7×10^7, 1.1×10^6 and 1.4×10^7 in the Taiwanese Han population, respectively, by testing HLA-DQA1, PM markers (LDLR, GYPA, HBGG, D7S8 and GC), and 9 STR loci (D3S1358, vWA, FGA, THO1, TPOX, CSF1PO, D5S818, D13S317 and D7S820). The age distribution of the parents at the birth of the child is shown in Table 2. The mutation rates found in this study for DYS389II and DYS392 (2 in 109 meioses for the DYS389II locus and 1 in 109 meioses for the DYS392 locus) are in agreement with the previous studies of Heyer et al. [9] and Kayser et al. [10]. With the exception of the locus DYS385, DYS389 and DYS392 were the only loci in which mutations were observed, when examining 8 Y-STR loci in 213 generations in the study by Heyer et al. [9]. Kayser et al. [10] reported 4 mutation loci found among 11 loci. In the DYS389II locus, 1 mutation event was observed in 53 meioses. Even though a limited number of meioses were screened in these three studies, the DYS389 locus was found in each study to have a higher mutation rate than the other Y-STR loci.

Table 2 The age distribution of the parents at the birth of the child in 100 families with 109 sons

Father		Mother	
Age (years)	No. of meioses	Age (years)	No. of meioses
< 19	2	< 19	2
19–24	9	19–24	14
25–29	18 (A)	25–29	32 (B, C)
30–34	22 (B)	30–34	21
35–39	20	35–39	8 (A)
≧ 40	12 (C)	≧ 40	6
Unknown	26	Unknown	26
Total	109	Total	109

A, B are mutation events at the DYS389II locus and C occurred at the DYS392 locus. The youngest father and mother were 17 and 16 years old, respectively, and the oldest father and mother were aged 56 and 41 years, respectively.

In conclusion, nine Y-STR loci can provide a high discrimination capacity for the identification of male DNA. In paternity cases of male offspring, these Y-STR loci can give powerful male lineage expectation, however, conclusions must be made carefully because of the high risk of mutation.

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