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SHORT COMMUNICATION

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Population data for 12 STR loci in Hong Kong Chinese

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Abstract The allele distributions at the 12 short tandem repeat (STR) loci D3S1358, HUMvWA, HUMFIBRA/FGA, HUMTHO1, HUMTPOX, HUMCSF1P0, D5S818, D13S317, D7S820, D8S1179, D21S11 and D18S51 have been determined for 284 unrelated Chinese in Hong Kong. The combined probability of identity for the 12 STR loci was about 4.1×10^{-14} and the overall probability of excluding paternity 0.999978. None of the 12 loci were found to deviate from Hardy-Weinberg expectations according to the results of the exact test. There was also little evidence for association of alleles between loci. The results demonstrate that the loci are useful for forensic human identification and parentage testing for the Chinese population in Hong Kong.

Keywords Forensic DNA typing · Polymerase chain reaction (PCR) · Short tandem repeat (STR) · Chinese · Allele frequencies

Introduction

Allele frequencies for some short tandem repeat (STR) loci have been reported for the Chinese populations in the north-east, east and south-east of mainland China and in Taiwan [1, 2, 3, 4]. A study of allelic frequency data for 12 STR loci, D3S1358, HUMvWA, HUMFIBRA/FGA, HUMTHO1, HUMTPOX, HUMCSF1P0, D5S818, D13S317, D7S820, D8S1179, D21S11 and D18S51, was conducted in the Hong Kong Chinese population. Commercially available kits that enable multiplex PCR of the 12 STR loci in 2 reactions were employed with the PCR products analysed using capillary electrophoresis. The power of discrimination, combined match probability or probability of identity, probability of excluding paternity, observed heterozygosity and unbiased estimates of expected heterozygosity and standard errors were obtained. Possible divergences from Hardy-Weinberg equilibrium and linkage equilibrium were investigated statistically.

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Materials and methods

Sample preparation

Specimens were obtained from 284 unrelated Chinese in Hong Kong, which included 119 whole blood samples and 165 buccal scrapings. DNA was extracted from the specimens by the Chelex extraction method [5]. The quantity of extracted DNA was estimated using the ACES 2.0^{+} human DNA quantitation system according to the manufacturer's procedure (User's manual from Life Technologies).

PCR amplification and STR typing

Amplification by PCR of the STR loci using 1–2.5 ng of extracted DNA was performed using the AmpF*l*STR Profiler and Profiler Plus kits, according to the manufacturer's recommendations (AmpF*l*STR Profiler and Profiler Plus user's manuals from PE/ABD), except that the reaction volume was 25 µl. All PCR reactions were performed in a PE Biosystems GeneAmp 2400 thermal cycler. An aliquot of 1–2.5 µl of each PCR product was denatured with formamide and electrophoresis was carried out on an ABI PRISM 310 genetic analyzer using the recommended protocol (AmpF*l*STR Profiler and Profiler Plus user's manuals from PE/ABD). The length of the amplified DNA fragments was determined using the GeneScan Analysis 2.1 software based on an internal lane ROX size standard, GeneScan-350 or GeneScan-500 from PE Biosystems. Allele designations were determined using the Genotyper 2.1 software by comparison of the sample fragments with those of the allelic ladders.

Statistical analysis

The Hardy-Weinberg and linkage equilibria were examined using the Fisher exact test [6]. The test has high statistical power to detect disequilibrium as reported by Zaykin et al. [7], and so the com-

Table 1 Allele frequencies for 12 STR loci in Hong Kong Chinese (*OH* observed heterozygosity, *EH* unbiased estimate of the expected heterozygosity and *SE* its standard error, *PD* power of discrimination, *PE* probability of excluding paternity)

Allele	D3S1358 (*n* = 284) Frequency	vWA (*n* = 282) Frequency	FGA (*n* = 283) Frequency	TH01 (*n* = 275) Frequency	TPOX (*n*=275) Frequency	CSF1PO (*n* = 268) Frequency	D5S818 (*n* = 284) Frequency	D13S317 (*n*=284) Frequency	D7S820 (*n* = 278) Frequency	D8S1179 (*n* = 276) Frequency	D21S11 (*n*=275) Frequency	D18S51 (*n* = 269) Frequency
6	–	–	–	0.100	–	–	–	–	–	–	–	–
7	–	–	–	0.316	–	0.009	0.035	0.004	0.002	–	–	–
8	–	–	–	0.053	0.545	0.002	0.005	0.306	0.121	–	–	–
9	–	–	–	0.440	0.100	0.037	0.072	0.130	0.068	0.002	–	–
9.3	–	–	–	0.029	–	–	–	–	–	–	–	–
10	–	–	–	0.060	0.022	0.239	0.252	0.155	0.162	0.138	–	0.004
11	–	–	–	0.002	0.313	0.261	0.255	0.278	0.376	0.121	–	–
12	–	–	–	–	0.020	0.362	0.210	0.099	0.230	0.118	–	0.045
13	0.002	0.004	–	–	–	0.082	0.165	0.023	0.034	0.167	–	0.156
14	0.033	0.254	–	–	–	0.006	0.005	0.005	0.007	0.152	–	0.184
15	0.331	0.035	–	–	–	0.002	–	–	–	0.196	–	0.203
16	0.326	0.156	–	–	–	–	–	–	–	0.098	–	0.126
17	0.239	0.266	0.002	–	–	–	–	–	–	0.007	–	0.104
18	0.056	0.160	0.025	–	–	–	–	–	–	0.002	–	0.056
19	0.011	0.106	0.065	–	–	–	–	–	–	–	–	0.035
20	0.002	0.020	0.044	–	–	–	–	–	–	–	–	0.024
21	–	–	0.131	–	–	–	–	–	–	–	–	0.024
21.2	–	–	0.004	–	–	–	–	–	–	–	–	–
22	–	–	0.178	–	–	–	–	–	–	–	–	0.024
22.2	–	–	0.004	–	–	–	–	–	–	–	–	–
23	–	–	0.189	–	–	–	–	–	–	–	–	0.009
23.2	–	–	0.004	–	–	–	–	–	–	–	–	–
24	–	–	0.166	–	–	–	–	–	–	–	–	0.004
24.2	–	–	0.007	–	–	–	–	–	–	–		–
25	–	–	0.110	–	–	–	–	–	–	–	–	–
25.2	–	–	0.007	–	–	–	–	–	–	–	–	–
26	–	–	0.048	–	–	–	–	–	–	–	–	0.002
26.2	–	–	0.005	–	–	–	–	–	–	–	–	–
27	–	–	0.011	–	–	–	–	–	–	–	0.004	–
28	–	–	0.002	–	–	–	–	–	–	–	0.067	–
28.2	–	–	–	–	–	–	–	–	–	–	0.004	–
29	–	–	–	–	–	–	–	–	–	–	0.253	–
30	–	–	–	–	–	–	–	–	–	–	0.224	–
30.2	–	–	–	–	–	–	–	–	–	–	0.005	–
31	–	–	–	–	–	–	–	–	–	–	0.096	–
31.2	–	–	–	–	–	–	–	–	–	–	0.067	–
32	–	–	–	–	–	–	–	–	–	–	0.027	–
32.2	–	–	–	–	–	–	–	–	–	–	0.191	–
33	–	–	–	–	–	–	–	–	–	–	0.004	–
33.2	–	–	–	–	–	–	–	–	–	–	0.047	–
34.2	–	–	–	–	–	–	–	–	–	–	0.011	–
OH	0.711	0.780	0.803	0.727	0.567	0.739	0.768	0.803	0.777	0.859	0.854	0.848
EH	0.723	0.791	0.867	0.689	0.594	0.735	0.794	0.778	0.759	0.854	0.828	0.866
SE	0.027	0.023	0.021	0.028	0.030	0.027	0.024	0.025	0.026	0.021	0.023	0.021
PD	0.873	0.934	0.968	0.854	0.768	0.885	0.926	0.917	0.908	0.961	0.949	0.965
PE	0.474	0.610	0.732	0.450	0.330	0.499	0.592	0.569	0.548	0.702	0.662	0.731

monly used χ^2 goodness-of-fit test [8, 9] was not attempted. The frequency of each allele was obtained using the standard gene counting method. Unbiased estimates of expected heterozygosities and the standard errors were calculated using the method as described by Nei and Roychoudhury [10] and compared to the observed heterozygosities. The power of discrimination [11] and the probability of excluding paternity [12] for each locus, and the combined probability of identity [13] and the overall probability of excluding paternity for the 12 loci were also evaluated.

Results and discussion

The observed allele frequencies for the 12 STR loci in the Chinese population in Hong Kong are shown in Table 1. The observed heterozygosity and the unbiased estimate of the expected heterozygosity for each locus were also calculated (Table 1). The observed values and the estimates are in good agreement, especially after taking into account the standard errors of the estimates, which are approximately 0.025. The power of discrimination and the probability of excluding paternity were evaluated for each locus and they are also reported in Table 1. The power of discrimination ranges from about 0.77 to 0.97 and the combined probability of identity for the 12 loci is about 4.1×10^{-14} which is extremely discriminating. The probability of excluding paternity spans from about 0.33 to 0.73 and the combined probability of excluding paternity for the 12 loci is 0.999978.

The data were tested for Hardy-Weinberg and linkage equilibrium. Since the size of the sample was not large (284 specimens), we only tested them for pair-wise linkage equilibrium. For linkage equilibrium at any three or more loci, the associated frequency table would be too sparse for a conclusive result to be obtained and so it was not attempted here. Table 2 presents the *p*-values of the exact test results [6]. For the single loci, all the *p*-values are higher than the commonly adopted 5% rule, indicating that there is no evidence of departure from Hardy-Weinberg equilibrium. For dependencies between the loci, five pairs of loci namely, 1 and 3 (D3S1358 and FGA), 1 and 12 (D3S1358 and D18S51), 2 and 3 (vWA and FGA), 3 and 6 (FGA and CSF1P0), and 3 and 12 (FGA and D18S51) indicated some degree of linkage disequilibrium. However, the associated *p*-values of 2.8%, 4.9%, 2.4%, 1.3% and 2.2%, respectively are not too small. Other combinations of loci seem to be in linkage equilibrium. Statistically, when the 5% significance rule is adopted, it is not unusual to have 5 significant results for a total of 78 tests even under a situation of total independence. Furthermore, none of the results were found to be significant if the 1% level was taken.

In conclusion, databases for the 12 STR loci D3S1358, vWA, FGA, THO1, TPOX, CSF1P0, D5S818, D13S317, D7S820, D8S1179, D21S11 and D18S51 have been established for the Hong Kong Chinese, which will be of great use both for forensic identity and parentage testing in the Hong Kong community.

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Table 2 *P* values of the exact tests for Hardy-Weinberg and linkage equilibrium for 12 loci (*1* D3S1358, *2* vWA, *3* FGA, *4* THO1, *5* TPOX, *6* CSF1P0, *7* D5S818, *8* D13S317, *9* D7S820, *10* D8S1179, *11* D21S11, *12* D18S51)

Locus combination	*P* value	Locus combination	*P* value
1	0.853	3/10	0.104
2	0.999	3/11	0.428
3	0.572	3/12	0.022*
4	0.377	4/5	0.284
5	0.832	4/6	0.512
6	0.968	4/7	0.162
7	0.993	4/8	0.551
8	0.505	4/9	0.696
9	0.860	4/10	0.672
10	0.224	4/11	0.297
11	0.912	4/12	0.463
12	0.205	5/6	0.758
1/2	0.280	5/7	0.338
1/3	0.028*	5/8	0.169
1/4	0.442	5/9	0.476
1/5	0.774	5/10	0.203
1/6	0.396	5/11	0.685
1/7	0.376	5/12	0.497
1/8	0.798	6/7	0.994
1/9	0.690	6/8	0.663
1/10	0.108	6/9	0.954
1/11	0.708	6/10	0.464
1/12	0.049*	6/11	0.979
2/3	0.024*	6/12	0.218
2/4	0.867	7/8	0.861
2/5	0.807	7/9	0.753
2/6	0.556	7/10	0.663
2/7	0.706	7/11	0.570
2/8	0.401	7/12	0.063
2/9	0.961	8/9	0.948
2/10	0.696	8/10	0.675
2/11	0.579	8/11	0.793
2/12	0.079	8/12	0.220
3/4	0.584	9/10	0.783
3/5	0.557	9/11	0.998
3/6	0.013*	9/12	0.641
3/7	0.168	10/11	0.629
3/8	0.248	10/12	0.449
3/9	0.526	11/12	0.280

*Significant value

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