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Analysis of the STR loci HUMF13A01, HUMFXIIIB, HUMLIPOL, HUMTH01, HUMTPOX and HUMVWFA31 in a Japanese population

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Abstract Population studies on six short tandem repeat loci, HUMF13A01, HUMFXIIIB, HUMLIPOL, HUMTH01, HUMTPOX and HUMVWFA31 were carried out in a sample of unrelated Japanese individuals (n = 337–545) living in Gifu Prefecture (central region of Japan). Five alleles could be identified for HUMFXIIIB, six for HUMF13A01, HUMLIPOL, HUMTH01 and HUMTPOX, and eight for HUMVWFA31. For all/six loci no deviations from the Hardy-Weinberg equilibrium hypothesis were detected. The mean exclusion chance ranged from 0.22 to 0.60, the power of discrimination from 0.63 to 0.93, and the expected heterozygosity from 0.43 to 0.80. Allele frequency distributions for the loci in the Japanese sample were not similar to those in samples from other racial or ethnic groups except for the Chinese (for HUMTPOX). The results demonstrate that HUMTH01, HUMTPOX and HUMVWFA31 are more useful for forensic investigations in the Japanese population than the other three loci.

Key words PCR · Short tandem repeats · Japanese population data

Introduction

In recent years, allelic data for short tandem repeat (STR) loci have been obtained from many different populations (e.g. Edwards et al. 1992; Wiegand et al. 1993; Pestoni et al. 1995). This paper presents allele frequencies for the STR loci HUMF13A01, HUMFXIIIB, HUMLIPOL, HUMTH01, HUMTPOX and HUMVWFA31 in a Japanese population sample, a comparison of the data with other populations and an evaluation of the potential forensic utility of the six STR loci in Japanese.

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Materials and methods

Blood samples were obtained from 545 unrelated Japanese individuals living in Gifu Prefecture (central region of Japan). DNA was extracted using the phenol-chloroform method (Maniatis et al. 1982). Each locus was amplified using the GenePrint STR System (Promega Corporation, WI, USA) according to the technical manual (part # TMD004). The amplified products were separated in 4% denaturing polyacrylamide gels (300 mm long and 1 mm thick) according to the technical manual and visualized by silver staining (Budowle et al. 1991). Alleles were determined by comparison with the allelic ladders included in the kits and were designated according to the number of repeat units. In addition, the repeat numbers of alleles that were outside the ladders or did not align with the alleles in the ladder, were confirmed by sequencing with ΔTaq fluorescent dye-primer cycle sequencing kit (Amersham, Bucks.). Sequence analysis was performed on a SQ-5500-S DNA Sequencer (Hitachi Electronics Engineering, Tokyo, Japan).

To estimate if the Japanese population sample examined in this study conforms to the Hardy-Weinberg equilibrium, the conventional χ^2 test between observed and expected genotype frequencies was carried out for each locus. Examinations for population sample homogeneity were also done by the χ^2 tests of $2 \times C$ contingency tables. For all χ^2 tests, the software SAS, Ver. 5 (SAS Institute, NC, USA) was used to calculate P-values. The mean exclusion chance (MEC) was calculated using the computer programme described by Ohno et al. (1982). The power of discimination (PD), expected heterozygosity (H-exp) and the standard error (SE) were calculated according to Fisher (1951) and Edwards et al. (1992), respectively.

Results and discussion

Allele frequencies for the six STR loci in the Japanese population sample are shown in Table 1. Five alleles could be identified for HUMFXIIIB, six for HUMF13A01, HUMLIPOL, HUMTH01 and HUMTPOX, and eight for HUMVWFA31. No significant deviations from Hardy-Weinberg equilibrium could be found for all loci (Table 1). The repeat numbers of alleles confirmed by sequence data in this study were allele 3.2 in HUMF13A01, allele 11 in HUMFXIIIB, allele 9.3 in HUMTH01, and allele 14 in HUMTPOX, and at least three examples of each allele were used for the sequence analysis. The sequence data revealed that allele 3.2 in HUMF13A01 lacked a GT

Table 1 Allele frequencies for six STR loci in the Japanese population sample and χ^2 test for Hardy-Weinberg equilibrium

Allele	HUMF13A01 (n = 392)	HUMFXIIIB (n = 367)	HUMLIPOL (n = 337)	HUMTH01 (n = 545)	HUMTPOX (n = 486)	HUMVWFA31 (n = 493)
3.2	0.328					
4	0.098					
5	0.028					
6	0.543			0.242		
7	0.001	0.003	0.003	0.286		
8		0.065		0.049	0.451	
9		0.203	0.001	0.382	0.121	
9.3				0.031		
10		0.725	0.712	0.010	0.036	
11		0.004	0.101		0.349	
12	0.001		0.180		0.039	
13			0.003			0.003
14					0.004	0.186
15						0.032
16						0.194
17						0.284
18						0.213
19						0.071
20						0.017
χ^2	5.80	7.27	5.54	21.47	8.22	28.14
df	15	10	15	15	15	28
P	0.983	0.699	0.987	0.122	0.915	0.457

n: number of individuals analysed

Table 2 χ^2 comparisons of different populations with the Japanese data from this study

Population	n	χ^2	df	P
HUMF13A01				
Asian Americans (Hammond et al. 1994)	63	27.76	6	< 0.001
Caucasian Americans (Hammond et al. 1994)	174	457.29	9	< 0.001
African Americans (Hammond et al. 1994)	175	593.47	12	< 0.001
Mexican Americans (Hammond et al. 1994)	183	354.43	8	< 0.001
HUMFXIIIB				
Japanese (Sato 1995)	98	8.47	4	0.078
German Caucasians (Meyer et al. 1995)	50	147.45	5	< 0.001
HUMLIPOL				
Asian Americans (Hammond et al. 1994)	77	33.28	6	< 0.001
Caucasian Americans (Hammond et al. 1994)	189	112.99	5	< 0.001
African Americans (Hammond et al. 1994)	174	201.13	6	< 0.001
Mexican Americans (Hammond et al. 1994)	179	56.70	6	< 0.001
HUMTH01				
Japanese (Fujita et al. 1995)	210	5.36	6	0.498
Asian Americans (Puers et al. 1993 a)	77	50.24	6	< 0.001
Caucasian Americans (Puers et al. 1993 a)	186	337.56	6	< 0.001
African Americans (Puers et al. 1993 a)	185	194.81	5	< 0.001
Mexican Americans (Puers et al. 1993 a)	192	201.77	5	< 0.001
HUMTPOX				
Chinese (Huang et al. 1995)	116	12.76	5	0.026
Swiss Caucasians (Hochmeister et al. 1995)	100	18.86	5	0.002
HUMVWFA31				
German Caucasians (Möller et al. 1994)	321	45.51	8	< 0.001
African Americans (Sajantila et al. 1994)	101	121.89	8	< 0.001
Galicians (Pestoni et al. 1995)	158	46.31	7	< 0.001

n: number of individuals analysed in published population studies

Table 3 Forensic efficiency values of six STR loci in the Japanese population sample

Locus	H-obs	H-exp ± SE	MEC	PD
HUMF13A01	0.62	0.59 ± 0.025	0.32	0.76
HUMFXIIIB	0.43	0.43 ± 0.026	0.22	0.63
HUMLIPOL	0.45	0.45 ± 0.027	0.24	0.65
HUMTH01	0.70	0.71 ± 0.019	0.46	0.86
HUMTPOX	0.65	0.66 ± 0.022	0.40	0.82
HUMVWFA31	0.79	0.80 ± 0.018	0.60	0.93
Combined			0.95	0.99994

H-obs: observed heterozygosity, H-exp: expected heterozygosity, SE: standard error, MEC: mean exclusion chance, PD: power of discimination

dinucleotide in the flanking region ($[AAAG]_4A$--AAAA) and allele 9.3 in HUMTH01 lacked an adenine base in the repeat region in allele 10 ($[AATG]_6[-ATG]_1[AATG]_3$) as reported by Puers et al (1994, 1993a), respectively. All alleles determined as allele 11 in HUMFXIIIB possessed the sequence $[AAAT]_{11}$, and all alleles 14 in HUMTPOX were $[AATG]_{14}$.

A quantitative comparison of allele frequencies for the six STR loci between this study and other population studies is shown in Table 2. No significant differences ($P > 0.01$) were observed between our Japanese sample and other Japanese samples for HUMFXIIIB and HUMTH01, and the Chinese sample for HUMTPOX. However, the allele distributions of the Japanese population sample are significantly different from those of the other population samples.

Table 3 summarizes various statistical parameters of forensic interest calculated for the six STR loci in our Japanese population sample. The forensic efficiency values of the six loci varied considerably, mainly due to differences in the allele frequency distributions. The forensic efficiency values of HUMTH01, HUMTPOX and HUMVWFA31 were high when compared with those of HUMF13A01, HUMFXIIIB and HUMLIPOL. Additionally, the frequencies of the most common alleles in the three former loci were always less than 50% in our Japanese sample (Table 1). These facts show that HUMTH01, HUMTPOX and HUMVWFA31 are more useful loci for paternity testing and identification in the Japanese population. HUMTPOX is more disciminating in our Japanese sample than in other population samples, e.g. Chinese (H-exp: 0.59, MEC: 0.31, PD: 0.75) (Huang et al. 1995), German Caucasians (H-exp: 0.59, MEC: 0.34, PD: 0.77) (Puers et al. 1993b), and Swiss Caucasians (H-exp: 0.59, MEC: 0.35, PD: 0.78) (Hochmeister et al. 1995). This result suggests that HUMTPOX is a more appropriate locus for forensic use in the Japanese population than in the other populations compared.

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