Forensic Science International: Genetics 4 (2010) e105–e108

Contents lists available at ScienceDirect

Forensic Science International: Genetics

journal homepage: www.elsevier.com/locate/fsig

Announcement of Population Data

Genetic polymorphisms of eight X-chromosomal STR loci in the population of Japanese

Jian Tie *, Seisaku Uchigasaki, Shigemi Oshida

Division of Legal Medicine, Department of Social Medicine, Nihon University School of Medicine, 30-1 Oyaguchi, Kamimachi, Itabashi-ku, Tokyo 173-8610, Japan

ARTICLE INFO

Article history:
Received 11 December 2008
Received in revised form 6 April 2009
Accepted 3 September 2009

Keywords:
STR
X-chromosome
Allele frequencies
Haplotype
Genetic polymorphism

ABSTRACT

The genetic polymorphisms of eight X-chromosomal short tandem repeats (STR) DXS10135, DXS8378, DXS7132, DXS10074, HPRTB, DXS10101, DXS10134 and DXS7423 were analyzed in a sample of 492 unrelated males (283) and females (209) from the Japanese population. Multiplex PCR amplification was performed using the Mentype® Argus X-8 PCR amplification kit. The haplotype frequencies within the four linkage groups were studied for the 283 examined Japanese males. Allele frequencies of eight X-STR loci were calculated separately for males and females, and exact tests demonstrated no significant deviations from Hardy–Weinberg equilibrium. Several microvariant and rare alleles were observed, and forensic efficiency parameters were calculated. The combined powers of discrimination of the loci in men and women were 0.999995 and 0.9999999999988, respectively.

Population: Peripheral blood samples were obtained from 492 unrelated male (283) and female (209) individuals of Japanese population living in Tokyo.

DNA extraction: Genomic DNA was extracted from whole blood samples by using QIaamp DNA Micro Kit (Qiagen, Chatsworth, CA).

PCR: 1 ng of target DNA was amplified with a commercial Mentype® Argus X-8 PCR amplification kit (Biotype, AG, Dresden, Germany) that allows single-tube co-amplification and detection of eight X-STR loci (DXS10135, DXS8378, DXS7132, DXS10074, HPRTB, DXS10101, DXS10134, DXS7423 and amelogenin) using the GeneAmp PCR System 9700 (Applied Biosystems, USA).

Gene typing: The amplified products and reference ladders provided by the kit were analyzed by capillary electrophoresis on an ABI PRISM™ 310 Genetic Analyzer (Applied Biosystems, USA) employing the analytical software GeneMapper™ ID v3.2 (Applied Biosystems, USA). For fragment length determination of the products, DNA size standard 550 ROX (Biotype AG, Germany) was included as an internal lane and used for calibration.

Access to data: The complete data are available to any interested researchers via mail from author (jtie@med.nihon-u.ac.jp).

Results: The distribution of allele frequencies and the forensic efficiency parameters (PIC, HET, PD, PE, MEC) are shown in Table 1. The Mentype® Argus X-8 PCR amplification kit makes possible to examine DXS10135, DXS8378, DXS7132, DXS10074, HPRTB, DXS10101, DXS10134 and DXS7423 microsatellite markers, which belong to the four linkage groups of the X-chromosome in one multiplex reaction [1]. The haplotypes are shown in Table 2.

Quality control: Laboratory internal control standard and kit DNA control.

Analysis of data: The allele frequency of each locus was calculated separately for males and the females. The eight X-STR polymorphisms were analyzed statistically by calculating the heterozygosity (HET) [2], polymorphism information content (PIC) [3], power of discrimination (PD) [4], power of exclusion (PE) [5], mean exclusion chance (MEC) for trio cases (mother, daughter, putative father), and MEC for duo cases (daughter, putative father) [6,7]. Hardy–Weinberg (HW) equilibrium was analyzed using exact test [8].

Sequence analysis: For confirmation of the alleles not included in the standard ladder markers, the samples were amplified again with appropriate primers for the respective loci for sequence analysis. PCR products were analyzed on 6.5% polyacrylamide gels containing 7 M urea. The target allele bands were eluted from the gel [9]. Allele sequencing was performed on the PCR-amplified fragments by the dye terminator method using the ABI PRISM™ 310 Genetic Analyzer. Amplifications were performed using

* Corresponding author. Tel.: +81 3 39587776; fax: +81 3 39587776.
E-mail address: jtie@med.nihon-u.ac.jp (J. Tie).

1872-4973/$ – see front matter.
doi:10.1016/j.fsigen.2009.09.002

Table 1
Allele frequencies for the eight X-STR loci in the Japanese individuals.

Allele	DXS10135	DXS8378	DXS7132	DXS10074	HPRTB	DXS10101	DXS10134	DXS7423
8		0.0010						
9		0.0142			0.0051			
10		0.5356						
11		0.2907	0.0051		0.0467			0.0020
11.2					0.0051			
12		0.1270	0.0569		0.3140			
13		0.0244	0.1697	0.0020	0.4411			0.0061
14		0.0051	0.3537	0.0234	0.1311			0.2470
15		0.0020	0.2846	0.0681	0.0478			0.6911
16	0.0061		0.1037	0.1463	0.0071			0.0528
16.2				0.0010				
17	0.0203		0.0193	0.3364	0.0020			
18	0.0650		0.0041	0.2632				0.0010
19	0.0874		0.0020	0.1402				
20	0.1280		0.0010	0.0183				
21	0.0874			0.0010				
22	0.1250							
23	0.0711							
24	0.0732							
25	0.0569							
26	0.0457							
27	0.0579					0.0061		
27.2						0.0010		
28	0.0417					0.0254		
28.2						0.0081		
29	0.0417					0.0346		
29.2						0.0305		
30	0.0407					0.0874	0.0020	
30.2						0.0976		
31	0.0213					0.1799	0.0010	
31.2						0.1220		
32	0.0163					0.1697	0.0234	
32.2						0.0884		
33	0.0030					0.0854	0.0346	
33.2						0.0407		
34	0.0051					0.0173	0.0650	
34.2						0.0020		
35	0.0041						0.1504	
36	0.0020					0.0020	0.2144	
37							0.2093	
37.3							0.0203	
38						0.0020	0.1687	
38.3							0.0254	
39							0.0528	
40							0.0102	
40.3							0.0010	
42.3							0.0071	
43							0.0010	
43.2							0.0020	
43.3							0.0091	
44							0.0020	
PIC	0.9219	0.5604	0.7214	0.7477	0.6400	0.8837	0.8426	0.3977
HET	0.9265	0.6141	0.7536	0.7739	0.6877	0.8903	0.8523	0.4604
p	0.0040	0.1326	0.0588	0.5674	0.2195	0.0160	0.2347	0.4613
PD_F	0.9965	0.7723	0.9730	0.9020	0.9508	0.9922	0.9926	0.7919
PD_M	0.9222	0.6190	0.7510	0.7758	0.6809	0.8853	0.8523	0.4384
PE	0.8036	0.3416	0.5055	0.5339	0.4217	0.7258	0.6443	0.2225
MEC_T	0.9178	0.5506	0.7118	0.7370	0.6338	0.8764	0.8316	0.3973
MEC_D	0.8532	0.4037	0.5750	0.6044	0.4894	0.7893	0.7258	0.2625

PIC: polymorphism information content; HET: expected heterozygosis; PD_F: power of discrimination in female; PD_M: power of discrimination in male; PE: power of exclusion; MEC_T: mean exclusion chance for trio cases (mother, daughter, putative father); MEC_D: mean exclusion chance for duo cases (daughter, putative father); *p*-Values of the exact tests for Hard–Weinberg equilibrium.

primer sequences according GeneBank information (http://www.gdb.org) and literature [10,11].

Other remarks: The investigated set of eight X-STR markers possesses a combined power of discrimination 0.999995 for men and 0.9999999999988 for women. Three rare alleles in the locus DXS10135 (33, 35, 36), 2 rare alleles in the locus DXS8378 (8, 15), 3 rare alleles in the locus DXS7132 (11, 19, 20), 2 rare alleles in the locus DXS10074 (13, 21), 2 rare and microvariant alleles in the locus HPRTB (11.2, 17), 10 rare and microvariant alleles in the locus DXS10101 (27.2, 28.2, 29.2, 30.2, 31.2, 32.2, 33.2, 34.2, 36, 38) and 10 rare and microvariant alleles in the locus DXS10134 (30, 31, 37.3, 38.3, 40.3, 42.3, 43, 43.2, 43.3, 44) were observed in this study. When the allelic frequencies of Japanese compared to other Asian populations, no statistically significant differences were observed with reported Chinese data for DXS8378, DXS7132, and HPRTB, loci. In the comparison of two loci with Korean data, DXS7132, HPRTB loci showed no significant difference; however, DXS8378 locus was significantly different [12–15].