Forensic Application of STR Polymorphic Markers

S. Nakamura, T. Sawaguchi and A. Sawaguchi

Department of Legal Medicine, Tokyo Women's Medical College, Tokyo 162, Japan

Introduction

STR (short tandem repeat) polymorphisms are powerful tools for human identification, paternity analysis and genetic mapping. STR loci consist of short, repetitive sequence elements of 3 to 7 bases pairs in length, and may be amplified using the polymerase chain reaction (PCR).
In the present study, we analysed 6 STR systems CSF1PO, FESFPS, F13B, TH01, TPOX and vWF to obtain allele frequency data for a Japanese population living in Tokyo. The efficiency of these STR systems in paternity testing including postmortem paternity cases was also analysed.

Materials and methods

Blood samples were obtained from 150 healthy Japanese individuals living in Tokyo. DNA was isolated from EDTA-treated blood samples by proteinase K / phenol / chloroform extraction. Polymorphic STR loci tested in the present study were indicated in Table 1. PCR amplification was performed using the GenePrint™ STR Systems (Promega Corporation, USA) and technical manual #TMD004 provided by the manufacturer. The electrophoresis was carried out on 4% denaturing polyacrylamide gels with the Sequi-Gen II system (BIO-RAD, USA) at constant voltage of 1,500V. The bands were visualized by silver staining with Silver Stain Plus Kit (BIO-RAD, USA). The alleles were determined with the allelic ladder provided in the GenePrint™ STR Systems from Promega Corporation.

Table 1 Polymorphic STR Loci tested in the Present Study

Locus	Gene (Chromosomal Location)	Rpeat Sequence	Allele Size Range (bases)
CSF1PO	Human c-fms proto-oncogene (5q33.5 - p34)	AGAT	299 - 323
FESFPS	Human c-fes/fps proto-oncogene (15q25-qter)	AAAT	222 - 250
F13B	Human coagulation factor XIII B subunit (1q31-q32.1)	AAAT	169 - 185
TH01	Human tyrosine hydroxylase (11p15.5)	AATG	179 - 203
TPOX	Human thyroid peroxidase (2p13)	AATG	232 - 248
vWF	Human von Willebrand factor (12p12-qter)	AGAT	139 - 167

Results and discussion

In 150 Japanese subjects, a total of 8 alleles for CSF1PO, 6 alleles for FESFPS, 5 alleles for F13B, 6 alleles for TH01, 6 alleles for TPOX and 8 alleles for vWF was observed.

The most common alleles were 12 for CSF1PO, 11 for FESFPS, 10 for F13B, 9 for TH01, 8 for TPOX and 17 for vWF. The distribution of 6 STR allele frequencies, heterozygosities, polymorphism information contents (PIC) and matching probabilities (pM) were indicated in Table 2. The combined matching probability of 6 STR systems was estimated as $9.2X10^{-6}$ and the combined power of discrimination was therefore 99.99908%. The combinational resolution of these 6 STR systems has been shown to be a powerful tool in personal identification.

The distribution of 6 STR allele frequencies in the various populations were compared. Allele frequency profiles for CSF1PO, FESFPS, TPOX and vWF in the present study were essentially the same as those indicated in Caucasians (Hammond et al. 1994; Puers et al. 1993a; Kimpton et al. 1992), and the allele frequency profiles for CSF1PO, FESFPS and TH01 from the present study showed a good agreement with Asians data obtained from Hammond et al. (1994) and Puers et al. (1993b). In F13B system the frequency of allele 10 obtained from the present study was higher than that obtained from Caucasian (Nishimura and Murray 1992), and the variant allele 9.3 in TH01 system was a rare allele in the Japanese and also in Asian whereas this allele was the most common in Caucasian (Puers et al. 1993b).

A postmortem paternity case was analysed by conventional systems and DNA profiling. The results were presented in Table 3. Routine paternity testing consisted of the conventional serological analysis of the red cell antigen systems ABO, MNSs, Rhesus, P, Duffy, Kidd and Lewis, the serum proteins GC, HP, BF, IF, C1R, C2, C6, C7, C81 and PLG, the red cell enzymes ACP, PGM1, ESD, GPT and PGD, and leucocyte antigens HLA-A, -B, -C by means of standard techniques. The probability of fatherhood by means of conventional systems was calculated as 83.25%. The simultaneous analysis of 4 STR systems following separate amplifications was presented in Fig. 2. From DNA profiling, the cumulative probability of fatherhood was calculated as 99.85%. An unequivocal conclusion was obtained.

From the present study, these STR systems are useful genetic markers for paternity tests and individual identification in forensic analysis.

Table 2 Analysis of Japanese population with six STR loci

Locus	CSF1PO	FESFPS	F13B	TPOX	TH01	vWF
			Allele frequency (%)			
Allele 6	-	-	-	-	20.16	-
Allele 7	0.97	-	0.33	-	29.84	-
Allele 8	2.43	-	4.68	48.64	4.65	-
Allele 9	6.80	1.39	20.33	5.78	41.09	-
Allele 9.3	-	-	-	-	2.71	-
Allele 10	26.21	13.89	74.33	3.06	1.55	-
Allele 11	17.96	33.33	0.33	36.06	-	-
Allele 12	35.44	30.56	-	4.76	-	-
Allele 13	9.22	18.06	-	1.70	-	0.47
Allele 14	0.97	2.77	-	-	-	17.67
Allele 15	-	-	-	-	-	4.65
Allele 16	-	-	-	-	-	18.60
Allele 17	-	-	-	-	-	28.83
Allele 18	-	-	-	-	-	22.79
Allele 19	-	-	-	-	-	6.06
Allele 20	-	-	-	-	-	0.93
Hetero-zygosity (%)	75.95	74.26	40.40	62.66	69.83	79.32
PIC	0.72	0.70	0.36	0.56	0.65	0.76
pM	0.094	0.110	0.404	0.206	0.144	0.074

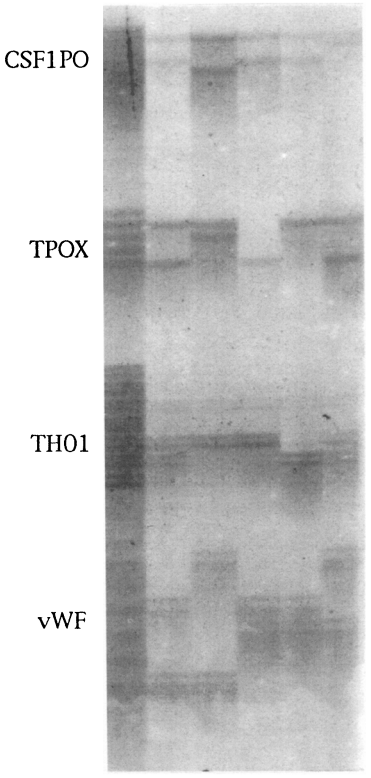

Fig. 2 Silver detection of 4 STR Loci.

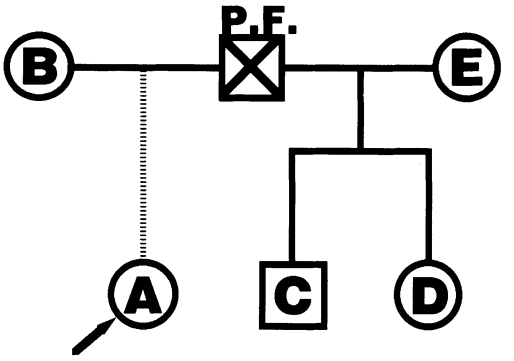

Fig. 1 Paternity testing with six STR loci in postmortem paternity case.

Table 3 Results of paternity testing

Markers	Probability of Fatherfood
Conventional Markers	0.8325
D1S80	0.7866
CSF1PO	0.7298
FESFPS	0.6923
F13B	0.5140
TH01	0.6729
TPOX	0.5069
vWF	0.7293
Cumulative	0.9985

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