J Forensic Sci, January 2006, Vol. 51, No. 1
doi:10.1111/j.1556-4029.2005.00039.x
Available online at: www.blackwell-synergy.com

FOR THE RECORD

Hiroyuki Ohtaki,[1] Ph.D.; Toshimichi Yamamoto,[2] Ph.D.; Shogo Yoshikawa,[1]M.S.; Takashi Yoshimoto,[2] Ph.D.; Rieko Uchihi,[2] Ph.D.; and Yoshinao Katsumata,[2] M.D.

Allele Frequency Distributions at Three Pentanucleotide Repeat STRs in a Japanese Population

POPULATION: Japanese.

KEYWORDS: forensic science, DNA typing, population genetics, short tandem repeat, Japanese population, penta B, penta E, penta C

Blood samples were collected from 300 unrelated healthy individuals residing in Japan (Nagoya city). DNA was extracted from each sample by the usual organic method.

One nanogram of template DNA was amplified using the Penta BEC Multiplex Primers (Promega, Madison, WI) following the manufacturer's instructions (1). The amplification included 30 cycles using the GeneAmp PCR system 9700 (Applied Biosystems, Foster City, CA). Amplified products were electrophoresed on an ABI PRISM 310 genetic analyzer (Applied Biosystems), and were analyzed using the GeneScan analysis 3.1 software (Applied Biosystems). The analyzed data were semi-automatically genotyped using the Genotyper 2.5 Software (Applied Biosystems), using the template made for this multiplex system temporally from the loading size data of each allele at those loci.

Tests for Hardy–Weinberg equilibrium (HWE) were carried out using the homozygosity test (2), likelihood ratio test (3), and exact test (4). The statistical properties of observed heterozygosity, expected heterozygosity (5), power of discrimination (6), polymorphism information content (7), and paternity exclusion rate (8) were calculated.

The allele frequency distributions of three short tandem repeat (STRs) in 300 Japanese are shown in Table 1. All genotype data for the three loci can be downloaded by any interested party from the following web site: http://www.med.nagoya-u.ac.jp/legal/jalm/BECdata.xls

p-values for HWE and the statistical properties of 3 STRs are shown in Table 2. The allele and genotype distributions at three STRs did not deviate from HWE.

The statistical data of three STRs were found to be relatively high values. The combined powers of discrimination and paternity exclusion rate were 0.99991 and 0.9653, respectively. Two Penta E variant alleles, one base shortened from the regular alleles 19 and 20 in size (alleles 18.4 and 19.4, respectively), were observed in the Japanese population. Sequence analyses revealed that both the variant alleles changed to $(GAAAA)_7(AAAA)(GAAAA)_{11}$ and $(GAAAA)_6(AAAA)(GAAAA)_{13}$, respectively, almost similar to the previous report (9).

TABLE 1—*Allele frequency distributions of three STRs in a Japanese population (n = 300).*

Allele	Penta B	Penta E	Penta C
5	—	0.100	0.067
6	—	—	—
7	0.005	—	—
8	0.003	0.018	0.087
9	0.182	0.007	0.315
10	0.095	0.042	0.058
11	0.163	0.120	0.343
12	0.338	0.133	0.107
13	0.090	0.027	0.023
14	0.077	0.050	—
15	0.032	0.133	—
16	0.012	0.090	—
17	—	0.087	—
18	—	0.065	—
18.4	—	0.002	—
19	—	0.030	—
19.4	—	0.002	—
20	—	0.037	—
21	—	0.032	—
22	—	0.017	—
23	0.003	0.005	—
24	—	0.005	—

STR, short tandem repeat.

[1] Criminal Investigation Laboratory, Aichi Prefectural Police Headquarters, Naka-ku, Nagoya 460-8502, Japan.
[2] Department of Legal Medicine and Bioethics, Graduate School of Medicine, Nagoya University, Showa-ku, Nagoya 466-8550, Japan.

TABLE 2—*p-values for Hardy–Weinberg equilibrium and statistical properties of three STRs.*

	Penta B	Penta E	Penta C
H	0.937	0.589	0.328
LR	0.857	0.717	0.623
ET	0.682	0.765	0.489
Ob. H	0.803	0.920	0.733
Ex. H	0.802	0.911	0.756
PD	0.936	0.985	0.905
PIC	0.777	0.905	0.720
PE	0.602	0.819	0.519

STR, short tandem repeat; H, homozygosity test based on 10,000 shufflings; LR, likelihood ratio based on 10,000 shufflings; ET, exact test based on 10,000 shufflings; Ob. H, observed heterozygosity; Ex. H, expected heterozygosity; PD, power of discrimination; PIC: polymorphic information content; PE, paternity exclusion rate.

Acknowledgments

This work was supported in part by Grants-in-Aid for Scientific Research from the Ministry of Education, Culture, Sports, Science and Technology of Japan (No. 15590577).

References

1. Promega Corporation. Penta BEC multiplex primers product information. Madison, WI: Promega Corporation; 2004.
2. Weir BS. Independence of VNTR alleles defined as fixed bins. Genetics 1992;130:873–87.
3. Chakraborty R, Fornage M, Guegue R, Boerwinkle E. Population genetics of hypervariable loci: analysis of PCR based VNTR polymorphism within a population. In: Burke T, Dolf G, Jeffreys AJ, Wolff R, editors. DNA fingerprinting: approaches and applications. Berlin: Birkhauser Verlag; 1991:127–43.
4. Guo SW, Thompson EA. Performing the exact test of Hardy–Weinberg Proportion for multiple alleles. Biometrics 1992;48:361–72.
5. Nei M. Estimation of average heterozygosity and genetic distance from a small number of individuals. Genetics 1978;89:583–90.
6. Grunbaum BW, Selvin S, Pace N, Black DM. Frequency distribution and discrimination probability of twelve protein genetic variants in human blood as functions of race, sex, and age. J Forensic Sci 1978;23:577–87.
7. Botstein D, White RL, Scolnick M, Davis RW. Construction of a genetic linkage map in man using restriction fragment length polymorphisms. Am J Hum Genet 1980;32:314–31.
8. Chakraborty R, Stivers DN. Paternity exclusion by DNA markers: effects of paternal mutations. J Forensic Sci 1996;41:671–7.
9. Mizuno N, Sekiguchi K, Sato H, Kasai K. Variant alleles on the Penta E locus in the PowerPlex16 Kit. J Forensic Sci 2003;48:358–61.

Additional information and reprint requests:
Toshimichi Yamamoto, Ph.D.
Department of Legal Medicine and Bioethics
Graduate School of Medicine
Nagoya University
65 Tsuruma-cho, Showa-ku, Nagoya 466-8550
Japan
Tel.: +81-52-744-2119
Fax: +81-52-744-2121
E-mail: yamachan@med.nagoya-u.ac.jp