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Analysis of the VNTR locus D1S80 in a Japanese population

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Summary. A population study on the VNTR locus D1S80 was carried out in a sample of 377 unrelated Japanese individuals living in and around Gifu Prefecture (central region of Japan). A total of 29 different alleles was distinguished. Alleles 18, 24 and 30 were found to be the 3 most common alleles in this population sample and their frequencies were 0.147, 0.200 and 0.162, respectively. Out of the 435 possible phenotypes, 120 were observed. The observed heterozygosity was 0.88 and the power of discrimination was 0.98. No significant deviations from Hardy-Weinberg equilibrium could be found in this Japanese population sample.

Key words: AMPFLP – D1S80 locus – Japanese population data

Zusammenfassung. Eine populationsgenetische Studie über den VNTR Locus D1S80 wurde an einer Stichprobe von 377 unverwandten Japanern durchgeführt, welche innerhalb und in der Umgebung der Gifu-Präfektur (Zentral-Japan) leben. Insgesamt wurden 29 verschiedene Allele unterschieden. Die Allele 18, 24 und 30 waren die 3 häufigsten Allele in dieser Populationsstichprobe, und ihre Frequenzen waren 0,147, 0,200 und 0,162. Von insgesamt 435 möglichen Phänotypen wurden 120 beobachtet. Die beobachtete Heterozygotie-Rate war 0,88 und der Diskriminations-Index betrug 0,98. In der untersuchten japanischen Stichprobe wurden keine signifikanten Abweichungen vom Hardy-Weinberg-Gleichgewicht gefunden.

Schlüsselwörter: AMPFLP – D1S80-Locus – Japanische Populationsdaten

Introduction

The majority of population studies on the VNTR locus D1S80 have been carried out on Caucasians (e.g. Budowle et al. 1991; Rand et al. 1992; Sajantila et al. 1992), and other population data are scarce. This paper presents allelic data for the D1S80 locus in a sample of 377 unrelated Japanse individuals living in and around Gifu Prefecture (central region of Japan).

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Materials and methods

DNA was extracted from blood obtained from 377 unrelated Japanese individuals living in and around Gifu Prefecture, using a phenol-chloroform extraction method (Maniatis et al. 1982). PCR of the D1S80 locus (Budowle et al. 1991) was performed in a MiniCycler™ (MJ Research, MA, USA). Temperature cycling conditions were the same as those described by Alonso et al. (1993). The amplified fragments were electrophoretically separated according to the method of Kasai et al. (1992) with modifications and visualized by either silver staining (Budowle et al. 1991) or ethidium bromide staining. Alleles were determined by the D1S80 Allelic Ladder (Cetus) and MS allelic ladder kindly supplied by Prof. Dr. B. Brinkmann (Institut für Rechtsmedizin, Westfälische Wilhelms Universität, Münster, Germany).

Results and discussion

A total of 29 different D1S80 alleles was observed in a sample of 377 unrelated Japanese individuals living in and around Gifu Prefecture (Table 1, Fig. 1). Alleles 18, 24 and 30 were found to be the 3 most common alleles with frequencies of 0.147, 0.200 and 0.162, respectively.

Table 1. D1S80 alleles observed in 377 unrelated Japanese

Allele	No.	Allele	No.	Allele	No.
14	2	24	151	34	5
15	3	25	18	35	1
16	29	26	6	36	5
17	16	27	34 (1)	37	5
18	111	28	69	38	1
19	12	29	27	39	3
20	14	30	122	40	0
21	17	31	58 (3)	41	2
22	7 (1)	32	13	42	5
23	5	33	6	43	7

The number of "interalleles" is shown in parentheses

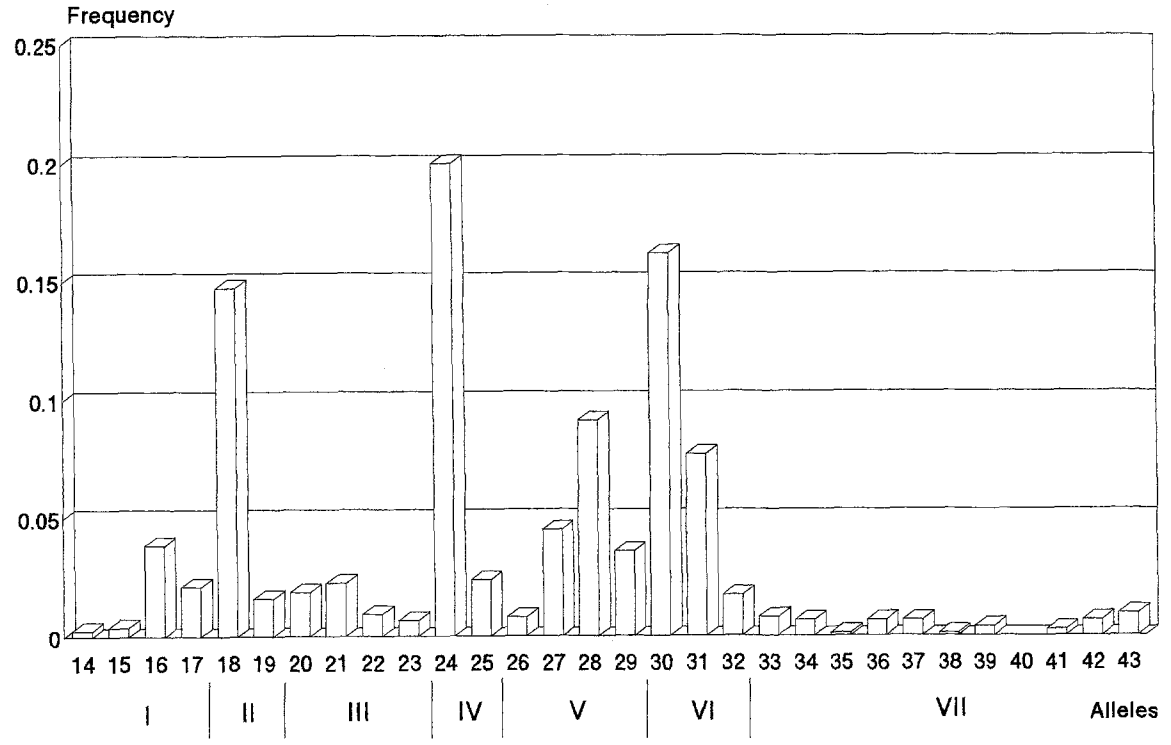

Fig. 1. D1S80 allele frequencies in 377 unrelated Japanese. I–VII indicate the groups of alleles used to estimate the Hardy-Weinberg equilibrium

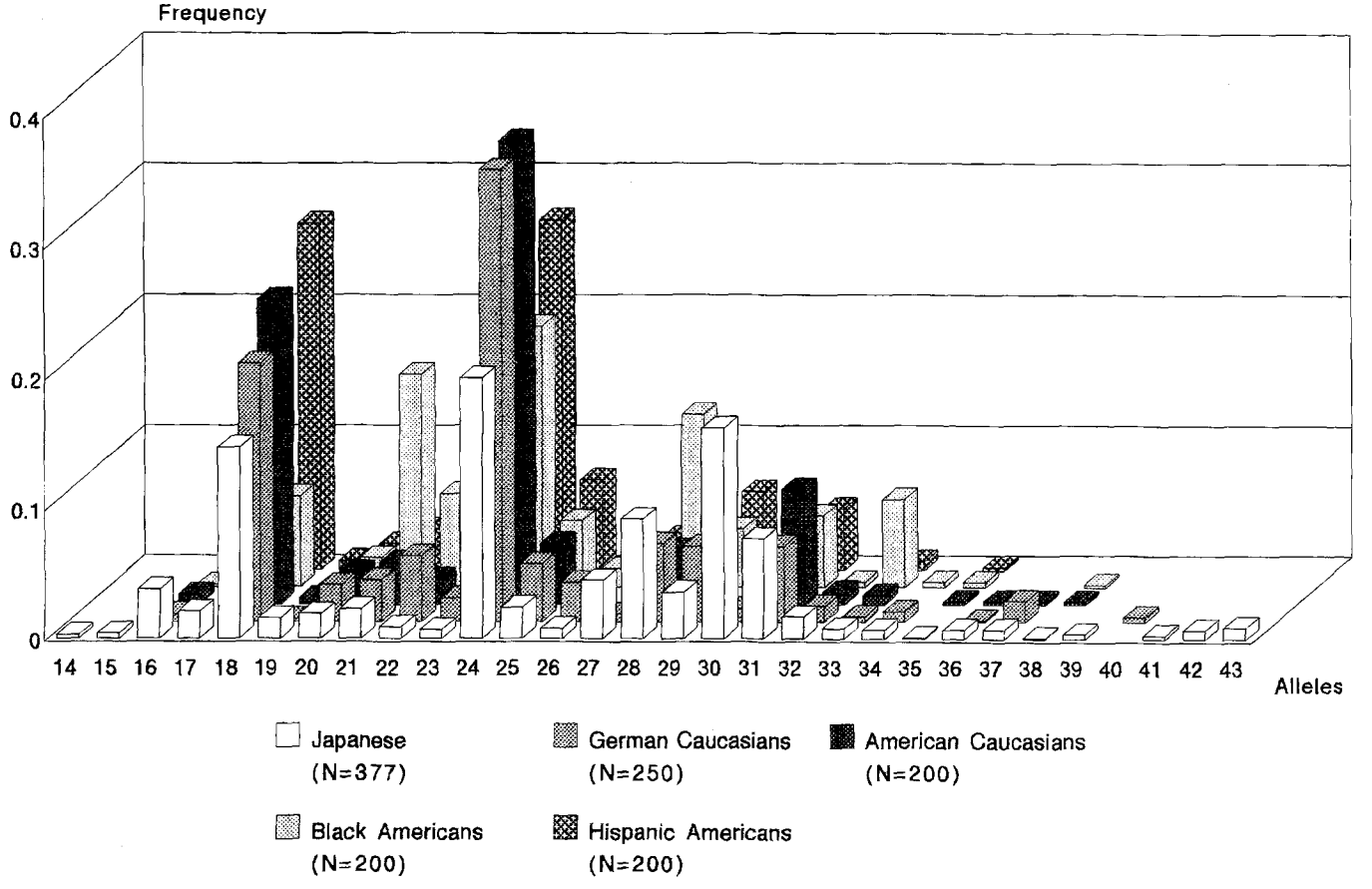

Fig. 2. Comparison of D1S80 allele frequencies from 5 population samples, i.e. Japanese (this study), German Caucasians (Schnee-Griese et al. 1993), American Caucasians, Black Americans and Hispanic Americans (Eisenberg and Maha 1991). The allele designation of Eisenberg and Maha (1991) has been aligned to the designation used in the study of Schnee-Griese et al. (1993) and in this study

"Interalleles" as described by Skowasch et al. (1992) were observed between alleles 22 and 23, between alleles 26 and 27, and between alleles 31 and 32. The "interalleles" were classified according to the closest alleles in the allelic ladder (Table 1). A total of 120 genotypes out of 435 possible genotypes was observed. As expected from the allele frequencies, the most common genotype was 24–30 with a frequency of 0.085. The χ^2 test was carried out to estimate if this population sample conforms to the Hardy-Weinberg equilibrium. Because of the large number of genotypes, 7 allele groups giving 28 genotype classes were formed by binning some alleles together (Brenner and Morris 1990) (Fig. 1). No significant deviations from Hardy-Weinberg equilibrium could be found ($\chi^2 = 27.70$; $df = 21$; $0.1 < P < 0.2$). The observed and expected heterozygosities were 0.88 and 0.89, respectively. The power of discrimination (PD) was 0.98. The expected heterozygosity and PD were calculated according to the formula described by Kloosterman et al. (1993).

A qualitative comparison of the allelic data between different populations showed that the distribution of allele frequencies in the Japanese is trimodal with 3 peaks at the alleles 18, 24 and 30, whereas the distribution in the Caucasians and the Hispanic Americans are bimodal with the 2 peaks at the alleles 18 and 24 (Fig. 2). The distribution in the Japanese is similar to that in the Black Americans with regard to the trimodality, however, 2 of the 3 peaks are different (Fig. 2). It is considered that the high frequency of allele 30 with alleles 18 and 24 is characteristic of the D1S80 allele distribution in the Japanese (Mongolian) population, while it must be taken into account that the population sample size and source in this study are insufficient to present a significant difference.

Analysis of the D1S80 locus for individual identification appears to be more valuable in Japanese population because there are many alleles more evenly distributed, and the good values of heterozygosity and PD.

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