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RESTRICTION FRAGMENT LENGTH POLYMORPHISMS OF X CHROMOSOME AMONG JAPANESE POPULATION

Hisao UEYAMA,[1] Wataru SHIRAHASE,[2] Takashi TAGA,[2] and Morimi SHIMADA[2]

Departments of [1]Medical Biochemistry and [2]Pediatrics, Shiga University of Medical Science, Seta, Otsu 520–21, Japan

Summary Restriction fragment length polymorphisms were studied among the Japanese using 13 polymorphic DNA probes on the X chromosome. For 6 probes (pPA4B, cpX203, p58-1, pHPGK-7e, cpX289 and 7b) the allelic frequencies were the same as those for Caucasians, but they were quite different ($p<0.01$) for 4 probes (dic56, pOTC (*Msp*I), pTAK8B and pXG-16 (*Hin*dIII)). No polymorphisms were observed for 4 probes (pG95α1-7dIII/RI (n (chromosome number studied)=54), pXG-16 (*Taq*I) (n=50), p8 (n=108), and pXG-17 (n=76). These results suggest that not a small number of DNA probes currently available are useless for linkage analysis in Japan.

INTRODUCTION

Restriction fragment length polymorphisms (RFLPs) are powerful tools for carrier detection and prenatal diagnosis of inherited diseases, especially of genetic disorders with unidentified biochemical lesions such as Huntington's chorea (Guesella *et al.*, 1983).

Many polymorphic DNA probes have been reported on the X chromosome, but their usefulness depends on the frequency of RFLPs. Shimmoto *et al.* (1988) studied DNA probes detecting polymorphisms on the short arm of X chromosome and found that one of the probes (pD2, DXS43) did not show any polymorphisms among the Japanese. We examined DNA probes for the distal end of the long arm of X chromosome and found no *Hin*dIII polymorphism for the probe p114.12 (F8C) in Japanese subjects (Taga *et al.*, 1989). In this communication, we present the frequencies of RFLPs among the Japanese for 13 probes for the other part of the X chromosome.

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Table 1. RFLP for DNA probes of X chromosome.

Probe	Restriction enzyme	Japanese		Previous reports	
		Size (kb)	Frequency (n)	Size (kb)	Frequency
pPA4B	*Xba*I	11.0	0.52(32)	11.0	0.52
		9.5	0.48(30)	9.5	0.48
dic56	*Bcl*I	8.9	0.85(46)	8.9	0.44
		7.4	0.15(8)	7.4	0.56
pOTC	*Msp*I	6.6	0.31(17)	6.6	0.61
		6.2	0.69(37)	6.2	0.39
	*Taq*I	3.7	0.83(44)	3.7	(heterozygosity
		3.6	0.17(9)	3.6	=0.11)
pTAK8B	*Xba*I	5.0	0.14(9)	5.0	0.36
		3.3	0.86(57)	3.3	0.64
		0.9	*	0.9	*
pG95α1-7dIII/RI	*Taq*I	5.7	1.00(54)	5.7	0.95
		4.5	0.00(0)	4.5	0.05
cpX203	*Bgl*II	5.8	0.42(22)	5.8	0.36
		1.0	0.58(31)	1.0	0.64
pXG-16	*Taq*I	7.1	0.00(0)	7.1	0.24
		3.8	1.00(50)	3.8	0.74
		3.5	0.00(0)	3.5	0.02
	*Hin*dIII	9.5	0.82(40)	9.5	0.50
		5.5	0.18(9)	5.5	0.50
p58-1	*Msp*I	4.0	0.52(26)	4.0	0.65
		2.5	0.48(24)	2.5	0.35
p8	*Taq*I	15.0	1.00(108)	15.0	0.84
		9.0	0.00(0)	9.0	0.16
pXG-17	*Msp*I	4.0	0.00(0)	4.0	0.13
		3.6	1.00(76)	3.6	0.87
pHPGK-7e	*Xba*I/*Pst*I	6.0	0.36(20)	6.0	0.45
		4.0,2.0	0.64(35)	4.0,2.0	0.55
cpX289	*Pst*I	5.5	0.80(78)	5.5	0.67
		1.6	0.20(20)	1.6	0.33
7b	*Pst*I	12.0	0.23(12)	12.0	0.21
		9.0	0.77(40)	9.0	0.79

n, chromosome number; *, constant band.

MATERIALS AND METHODS

Extraction of high-molecular-weight DNA, Southern blotting and hybridization were performed as previously described (Taga *et al.*, 1989). The DNA probes studied were kindly supplied by Dr. Szabo (pXG-17), Dr. Kunkel (dic56), Dr. Kruse (pPA4B and pTAK8B), Dr. Pearson (cpX289), and Dr. White (pG95α1-7dIII/RI). The other 7 probes were obtained from American Type Culture Collection (ATCC). Arveiler *et al.* (1987) reported a *Pst*I RFLP detected with probe cpX73 (DXS162), but they actually worked with cpX289 (DXS159), which we also used in this study.

RESULTS AND DISCUSSION

Table 1 summarizes the frequency distribution among the Japanese for 13 DNA probes we studied.

No frequency differences were found between the previous report and this study for probes pPA4B, cpX203, p58-1, pHPGK-7e, cpX289, and 7b. However, the allelic frequencies were quite different ($p<0.01$) for probes dic56, pOTC (*Msp*I), pTAK8B, and pXG-16 (*Hin*dIII). As for the probes dic56 and pOTC, more frequent allele in Caucasians than the other was found to be less frequent in Japanese. The PIC values for probes dic56, pTAK8B, and pXG-16 (*Hin*dIII) are lower in Japanese than in Caucasians (0.49→0.26, 0.46→0.24, and 0.5→0.30, respectively). No polymorphisms were found for probes pG95α1-7dIII/RI, pXG-16 (*Taq*I), p8, and pXG-17. Shimmoto *et al.* (1988) reported no polymorphisms in Japanese for one probe (pD2, DXS43) among 9 they studied. We also found no polymorphisms

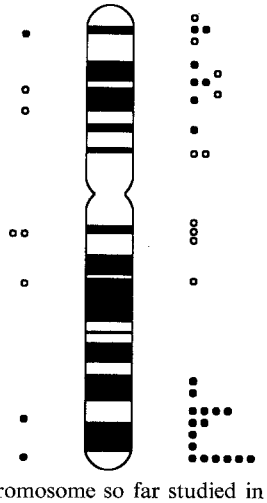

Fig. 1. RFLP probes for X chromosome so far studied in Japanese. ○, studied in this communication; ●, studied previously. RFLP probes which gave polymorphisms in Caucasians but not in Japanese are shown on the left.

for one probe (p114.12 (*Hin*dIII), F8C) among 12 we examined (Taga *et al.*, 1989). Kojima *et al.* (1987) reported the absence of four polymorphic sites in Japanese subjects, which are frequent in Caucasians, in coagulation factor IX gene. These results and those we obtained in this study suggest a significant difference in the allelic frequency between these ethnic groups. We also report here the allelic frequency of *Taq*I polymorphism detected by pOTC, which is not known yet even in Caucasians (Nussbaum *et al.*, 1986). As for *Msp*I polymorphisms detected by probe pOTC, neither the 5.1 kb nor the 4.4 kb band was observed in the Japanese or 5 Caucasian subjects.

Figure 1 shows the polymorphic probes for X chromosome so far studied in the Japanese, which cover from pter to qter regions of the chromosome. Similar systematic studies of polymorphic probes for autosomes are absolutely needed for linkage analysis in Japan, though there have been not a few reports on the ethnic differences (Prochownik *et al.*, 1983; Antonarakis *et al.*, 1985; Haneda *et al.*, 1986; Wainscoat *et al.*, 1986; Lubahn *et al.*, 1987; Suzuki *et al.*, 1988; Suehiro *et al.*, 1988; Ogasawara, 1988).

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