SHORT COMMUNICATION

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Y-Specific DNA polymorphisms of the *YAP* element and the locus *DYS19* in the Korean population

Received: March 18, 1998 / Accepted: May 19, 1998

Abstract The Y *Alu* polymorphic (*YAP*) element (*DYS287*) and the Y-linked tetranucleotide microsatellite locus *DYS19* were examined in samples from a total of 455 unrelated males in the Korean population. The frequency of the *YAP*⁺ allele was found to be 1.3% (6/455) in the Korean population. These results are consistent with previous reports that showed the *YAP* element to be absent in most Asian populations, with the exception of the high frequency of the *YAP*⁺ allele in the Japanese population. All five common alleles at the *DYS19* locus were identified in this study. The C allele was the most frequent (197/455), followed by the D (119/455), B (78/455), E (41/455), and A (20/455) alleles. Seven combination haplotypes (*DYS287/DYS19*) were found, and the mean combination haplotype diversity in the Korean population appeared to be 0.71. Based on results of these two loci, Japanese and Korean populations may share some common genetic structure that could reflect recent gene flow and some amount of admixture of Y chromosomes between these two populations.

Keyword Y Chromosome · *DYS287* · *DYS19* · Chinese · Japanese · Korean

Introduction

With the exception of the pseudoautosomal region, the human Y chromosome is haploid and paternally inherited without recombination. Consequently, Y haplotypes constructed with alleles at multiple polymorphic sites can be used for studies of paternal lineages and population history

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in humans (Ngo et al. 1986; Hammer 1994; Spurdle et al. 1994a).

The presence or absence of the Y *Alu* polymorphic (*YAP*) insert (*DYS287*) has proved to be a useful marker for human population studies (Batzer and Deininger 1991; Perna et al. 1992; Hammer 1994, 1995; Hammer et al. 1997). The *YAP* insertion is likely to have originated only once, so it has the feature of a unique polymorphism (Hammer 1994). Significant differences in the frequencies of the *YAP* insertion were detected among worldwide populations (Hammer 1994; Hammer et al. 1997). The *DYS19* microsatellite polymorphism is due to variation in the number of GATA repeats (Roewer et al. 1992; Santos et al. 1993, 1996b; Gomolka et al. 1994). It is known to be a useful tool for studying human population structure. There are at least nine different alleles, including five common alleles (A–E) that exhibit remarkable frequency differences in worldwide populations (Santos et al. 1993, 1996b; Gomolka et al. 1994; Hammer and Horai 1995). Therefore, these two genetic markers have been used to compare genetic similarities in different human ethnic groups. Here, we report results for the frequency of these two Y-linked specific markers in the Korean population.

Subjects and methods

We studied samples from a total of 455 unrelated Korean males for the detection of allelic variations at the loci *YAP* element (*DYS287*) and *DYS19*. DNA was extracted from buccal cells (Richards et al. 1993) from volunteer donors in Korea. The *YAP* insertion and *DYS19* alleles were detected by polymerase chain reaction (PCR) amplification, using flanking primers described by Hammer and Horai (1995) and Roewer et al. (1992). The *YAP*⁺ (~460bp) or *YAP*[−] fragment (~150bp) of PCR products was electrophoresed in 2% agarose gel (Fig. 1). In the case of *DYS19* alleles, the DNA fragments derived from PCR amplification were separated by using 10% polyacrylamide gel electrophoresis (PAGE) for 10h at 100V. Following the PAGE, the frag-

Fig. 1 Detection of insertion of *YAP* element in polymerase chain reaction (PCR) products. Electrophoresis in a 2% agarose gel of restriction fragments of PCR product produced by digestion with *TaqI*. Lane 1, DNA size markers; lane 2, [*YAP*⁺] KK1 sample; lane 3, female; lane 4, [*YAP*⁺] KK1 digested with *TaqI*; lane 5, *YAP*⁻ control

Fig. 2 Polyacrylamide gel electrophoresis of PCR products amplified with specific flanking primers of *DYS19*. Fifteen individuals displaying different allelic classes at the *DYS19* locus in increasing order of size. Lane 1, DNA size markers; lanes 2–4, A (186 bp); lanes 5–7, B (190 bp); lanes 8–10, C (194 bp); lanes 11–13, D (198 bp); lanes 14–16, E (202 bp)

Fig. 3 Nei's genetic distance (Nei 1972) for the *DYS19* locus in several Asian populations. Nei's standard genetic distance (D) in the table body was based on allele frequency of *DYS19*

Population	(1)	(2)	(3)	(4)	(5)	(6)
(1) Chinese						
(2) Indians	0.0159					
(3) Japanese	0.1550	0.2265				
(4) Koreans	0.0291	0.0693	0.0478			
(5) Mongolians	0.0116	0.0237	0.2181	0.0603		
(6) Tibetans	0.1147	0.1153	0.1273	0.0854	0.1850	

ments were visualized using a simple ethidium bromide staining and ultraviolet light (Fig. 2).

The mean haplotype (gene) diversity was calculated for each group as $h = n(1 - \sum x_i^2)/(n - 1)$, where n represents the number of chromosomes sampled and x_i is the frequency of the i th haplotype (Nei 1987). The PHYLIP Package (Felsenstein 1993) was used to compute genetic distances (Nei 1972) for use in distance matrix programs based on Y chromosome haplotype frequencies.

Results

Polymorphisms at the loci *YAP* element and *DYS19*. The frequencies of the *YAP* element and *DYS19* alleles in this study are given, with the results of previous surveys, in Table 1. The frequency of the *YAP* element insertion in the 455 Korean males was 1.3% (6/455). This result for the Korean population is consistent with previous reports that showed the *YAP* element to be absent in most Asian and Oceanic populations, with the exception of the highly polymorphic Japanese and Tibetans (Hammer 1994; Spurdle et al. 1994b; Hammer and Horai 1995; Hammer et al. 1997). The six *YAP*⁺ chromosomes detected in this study were

associated with long adenine residues (41 bp–50 bp) at the 3' end of the *Alu* sequence (poly(A) tail) (data not shown). These chromosomes probably belong to haplotype 3, which is frequently found in the Japanese population (Hammer 1995; Hammer et al. 1997).

All five common alleles (A–E) at the *DYS19* locus were identified in the Korean population (Table 1). The C allele was the most frequent (197/455), followed by the D (119/455), B (78/455), E (41/455), and A (20/455) alleles. The predominance of the C allele in this study is similar to that in previous reports from other surveys of Asian, Australian aboriginal people, and indigenous African populations (Gomolka et al. 1994; Muller et al. 1994; Hammer and Horai 1995; Hammer et al. 1997). Nei's genetic distances, based on the frequencies for *DYS19* alleles (Table 1), show that Koreans appeared to have the closest genetic relationship with Chinese (Fig. 3). The Japanese population is more similar to Koreans than to several other Asian populations.

A total of seven haplotypes were found with respect to the *YAP/DYS19* combination in the Korean sample (Table 2). The mean combination haplotype diversity was 0.71. In this survey, the haplotype H3 (–/C) had the most common frequency (42.9%). Among the seven *YAP/DYS19* combination haplotypes, *YAP*⁻ chromosomes were associated with all of five *DYS19* alleles, while the *YAP*⁺ allele was

Table 1 Allele frequencies of two Y-chromosome specific loci in several Asian populations

Population (<i>n</i>)	No. (%) of <i>YAP</i> allele		No. (%) of <i>DYS19</i> allele						
	+	–	Z	A	B	C	D	E	F
Chinese (76) ^a	1 (1.3)	75 (98.7)	1 (1.3)	3 (3.9)	21 (27.6)	29 (38.2)	19 (25.0)	3 (3.9)	0
Indians (43) ^b	0	43 (100.0)	0	1 (2.3)	15 (34.9)	17 (39.5)	8 (18.6)	2 (4.7)	0
Japanese (132) ^c	55 (41.7)	77 (58.3)	0	10 (7.6)	4 (3.0)	63 (47.7)	32 (24.2)	22 (16.7)	1 (0.8)
Koreans (455) ^d	6 (1.3)	449 (98.7)	0	20 (4.4)	78 (17.1)	197 (43.3)	119 (26.2)	41 (9.0)	0
Mongolians (46) ^e	1 (2.2)	45 (97.8)	0	1 (2.2)	14 (30.4)	15 (32.6)	12 (26.1)	2 (4.3)	2 (4.3)
Tibetans (30) ^b	14 (46.7)	17 (53.3)	0	0	7 (23.3)	20 (66.7)	3 (10.0)	0	0

^aHammer et al. (1997); ^bHammer and Karafet (unpublished data); ^cHammer and Horai (1995); ^dPresent study; ^eSantos et al. (1996a)

^{*}Most are Taiwanese of Chinese ancestry (Horai et al. 1987; Hammer and Horai 1995)

Table 2 Comparison of frequencies for the *YAP/DYS19* combination haplotype in the Korean population

Haplotype number	Combination haplotype (<i>YAP/DYS19</i>)	No. (%) (<i>n</i> = 455)
H1	–/A	20 (4.4)
H2	–/B	78 (17.1)
H3	–/C	195 (42.9)
H4	–/D	115 (25.3)
H5	–/E	41 (9.0)
H6	+/C	2 (0.4)
H7	+/D	4 (0.9)

Mean haplotype diversity values appeared to be 0.71 in the Korean population

associated only with the C and D alleles of *DYS19* (Table 2). This result is consistent with a previous report that, in East Asia, the *YAP*⁺ chromosome (haplotype 3) is mainly associated with the C and D alleles of *DYS19* and was never found with the *DYS19* B allele (Hammer et al. 1997).

Discussion

The genetic structure of the Korean population. The Korean population appears to have an extremely low frequency of the *YAP*⁺ allele (6/455) (Table 1). This result is consistent with earlier reports showing that most East Asian populations have very low frequencies of the *YAP*⁺ allele (less than 2%). The notable exception to this pattern was the finding of high frequencies of the *YAP*⁺ allele in Japan (41.7%) and Tibet (46.7%) (Hammer and Horai 1995; Hammer et al. 1997). On the other hand, the *DYS19* data presented here indicate that the Korean population is more similar to the Chinese. The Japanese population appears to have a closer genetic relationship to Koreans than to the other surveyed Asian populations (Fig. 3). Most authors generally agree that Koreans are considered as a North Asian group (Nei and Roychoudhury 1993; Cavalli-Sforza et al. 1994). Earlier reports of the frequencies of the *YAP/DYS19* combination haplotypes (Table 1) suggest that Japanese and Tibetan populations shared a common ancestral population. These results may reflect the “out of northeast-Asia hypothesis” of the origin of Japanese populations (Hammer and Horai 1995; Nei 1995).

The *YAP/DYS19* combination haplotype diversity in the Korean population ($h = 0.71$) was found to be lower than that in other Asian populations. A global survey has shown that sub-Saharan Africans have the highest diversity (0.878) of this mean combination haplotype, followed by Asians (0.779), Europeans (0.736), and indigenous Australasians (0.718) (Hammer et al. 1997). The low diversity level detected in our Korean sample may be due to the homogenizing influence of migrations from northeast Asia (Nei and Roychoudhury 1993; Cavalli-Sforza et al. 1994). Indeed, the high Japanese diversity levels in Asians (0.835) have been explained as resulting from a combination of an ancient population (Jomon people) subject to a more recent admixture (Yayoi people) (Hammer and Horai 1995; Hammer et al. 1997).

Based on the results for these two loci, it seems that Korean and Japanese populations may share some common genetic structure that is rare or not present in the other groups. Earlier surveys of *DXYS5Y* (47z) polymorphisms in Africa, East Asia, and Europe suggest that the Y2 allele is restricted to Korea and Japan (Nakagome et al. 1992; Mathias et al. 1994). A similar overall pattern was detected in a much larger sample of Koreans ($n = 480$, Y2 = 4.6%, W. Kim and D.J. Shin, unpublished data). This similarity may be because there has been some admixture between these two populations. This is also consistent with the hypothesis that the Y2 allele tracks male lineages that originated in Korea and migrated to Japan (Hammer and Horai 1995). Hammer and Horai (1995) reported that there were high frequencies of the *YAP*⁺ haplotype in Hokkaido and in Okinawa, with lower frequencies in southwestern Japan, the region closest to the Korean peninsula. There is also evidence for a migration of Yayoi people from the Korean peninsula to Japan about 2300 years ago (Chard 1974). An extremely low frequency of *YAP*⁺ alleles (1.3%) in the Korean population provides additional evidence to support this hypothesis.

Based on the results shown here, the 6 *YAP*⁺ chromosomes detected in our survey had the long poly (A) tail (41–50 dA-nucleotides) at the 3' end of the *Alu* sequence. This is consistent with their belonging to *YAP* haplotype 3, which is frequently found in Japanese populations (Hammer 1995; Hammer et al. 1997). This result could reflect a recent gene flow from Japan to Korea. Therefore, we suggest that the rate of admixture for *YAP*⁺ chromosomes

from Japan to Korea seems to be lower than that of the apparently large infusion of *YAP*⁺ chromosomes with the Yayoi migration, starting 2300 years ago, from Korea to Japan.

Acknowledgments We thank Dr.M.F. Hammer for help and advice during the course of this work. This study was supported by the Basic Science Research Institute Program (BSRI-96- No. 4440) of the Ministry of Education, Republic of Korea.

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