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SHORT COMMUNICATION

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The Y-27H39 polymorphism in a Japanese population

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Abstract We studied 184 Japanese males for the tetranucleotide TAGA repeat polymorphism at the Y-27H39 locus on the Y chromosome, and discovered a new allele, tentatively named F. Direct sequencing of Y-27H39 alleles revealed that the new allele (206 nt) is larger than allele E (202 nt) by 1 repeat unit. The six alleles differ only in the number of repeats, the flanking sequences being constant. The allele frequencies are different in Japanese and Caucasians.

Key words Y chromosome · Polymorphism · Population study · DNA sequencing

Introduction

The tetranucleotide repeat polymorphism at the Y-27H39 locus on the human Y chromosome (Roewer et al. 1992; Santos et al. 1993; Yamamoto et al. 1994) is attracting forensic interest because of the potential for sex identification as well as individual identification and paternity testing in deficiency cases. We studied the Y-27H39 polymorphism in a Japanese population and discovered a new allele.

Material and methods

Blood samples were taken from 184 Japanese males and DNA was extracted as described elsewhere (Lewin and Stewart-Haynes 1992). Two primers, H39.1–5′-CTACTGAGTTTCTGTTATAGT-3′ and H39.2–5′-ATGGCATGTAGTGAGGACA-3′ (Roewer et al. 1992) were used to amplify Y-27H39 alleles. The same primers, one of which was labeled with biotin, were then used to prepare the single-stranded Dynabeads-coupled templates (Dynal, Oslo) for direct sequencing of PCR products (fmol DNA sequencing system, Promega, Madison, Wis) as described previously (Wang et al. 1996). The PCR products were resolved on 5% polyacrylamide gels (Long Ranger, AT Biochem, Malvern, Pa) containing 7 M urea, and capillary blotted onto a Hybond-N+ membrane (Amersham, Tokyo). Alleles were visualized by probing with the primer H39.1 which was 3′-labeled with digoxigenin-dUTP, followed by reaction with alkaline phosphatase-labeled Fab fragments of anti-digoxigenin antibodies (DIG Oligonucleotide Tailing Kit, Boehringer Mannheim).

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Results and discussion

Since we found only one F allele in a sample of 184 individuals (Fig. 1 and Table 1) and no male relatives of the

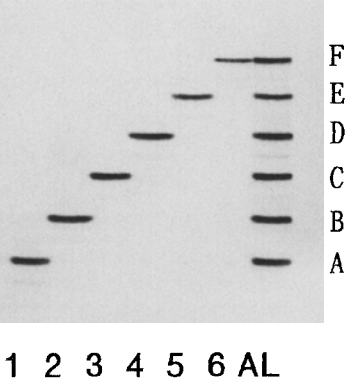

Fig. 1 The six alleles detected at Y-27H39 locus in a sample of 184 Japanese males. Lanes 1–6: alleles A to F. AL: allelic ladder

Table 1 Allele frequencies for Y-27H39 in Japanese and Caucasians

Allele	Length (nt)*	Frequency	
		Japanese	Caucasians*
A	186	0.054	0.19
B	190	0.071	0.49
C	194	0.457	0.24
D	198	0.190	0.07
E	202	0.223	0.01
F	206	0.005	–

* Santos et al. (1993)
* nt = nucleotide

Table 2 Sequence of alleles A-F from this study compared with the reported sequences from variable regions of Y-27H39 allele A

Allele	Repeat segment				
	1	2	3	4	5
	$(TA)_5$ GTNN $(TA)_5$	GTNN	$(TA)_6$	GTNNTT	$(TAGA)_3$ TANN $(TAGA)_{10}$ TATA*
A	----------	GT------	GT----------		GG $(TAGA)_{10}$ ---
B	----------	GT------	GT----------		GG $(TAGA)_{11}$ ---
C	----------	GT------	GT----------		GG $(TAGA)_{12}$ ---
D	----------	GT------	GT----------		GG $(TAGA)_{13}$ ---
E	----------	GT------	GT----------		GG $(TAGA)_{14}$ ---
F	----------	GT------	GT----------		GG $(TAGA)_{15}$ ---

* Sequence reported by Roewer et al. (1992)

proband were available for pedigree studies, we had to eliminate the possibility of the new allele having been amplified by Taq DNA polymerase errors. The F allele was reproducibly amplified from the original DNA sample. In addition, direct sequencing of the new allele showed it to be exactly 4 nt (a repeat unit of GATA) longer than allele E. The results prove the existence of this new allele at the Y-27H39 locus (Table 2). The hitherto detection of five alleles (A-E) in Caucasians (Santos et al. 1993) and six alleles (A-F) in Japanese (this study) cannot be interpreted as a racial difference because the data for Caucasians are from a relatively small sample size of 100 individuals. However, the frequencies of the alleles are different between the two populations (Table 1).

The Y-27H39 locus shows a regular repeat variability and a relatively high discriminating power of 0.697, and therefore appears especially useful for sex identification, paternity testing in deficiency cases and studies of paternal lineage. Recently, we have successfully applied Y-27H39 typing in a deficiency case of disputed paternity (Kishida et al. 1996).

Note added in proof We discovered another new allele G (210 nt) with a frequency of 1 in 200. Direct sequencing revealed this allele to be longer than allele F by one repeat unit (GATA).

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