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SHORT COMMUNICATION

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Analysis of the STR myelin basic protein locus in Koreans

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Abstract Allele und genotype frequencies and the mutation rate of a short tandem repeat locus, the myelin basic protein (MBP) gene, were studied in 973 unrelated Koreans. The alleles were distributed in two discrete regions, one in a high molecular weight region (A) above 190 bp and the other in a low molecular weight region (B) below 150 bp. In a heterozygote, two alleles were found in each region. In region A 13 alleles were found and in region B 7 alleles. Most alleles showed a difference of 4 bp, but three interalleles were found in region A. Allele frequencies in Koreans differed from those reported for Germans and Portuguese. Sets of alleles, one from each region, were linked and transmitted to the offspring. A total of 36 haplotypes and 148 genotypes was identified. In 763 gametes of 550 families, whose parent-child relationship was confirmed using other serological and DNA systems, all alleles were transmitted in a Mendelian fashion, and no mutations were observed. The polymorphism information content (PIC) in Koreans was calculated as 0.833 for region A and 0.718 for region B. The power of discrimination (PD) was 0.959 for region A and 0.901 for region B. No significant deviation from Hardy-Weinberg equilibrium could be observed for this system.

Key words STR · Myelin basic protein · Koreans

Introduction

Many short tandem repeat (STR) loci showing a high degree of polymorphism have recently been revealed, and they have become promising markers for individual identification and paternity testing. The STR myelin basic protein (MBP) locus is peculiar in that a polymorphism of two introns can be amplified using one pair of primers (Boylan et al. 1990), which are 5′ to the human MBP gene [18q23-pter] with a $[TGGA]_n$ repeat. For a genetic polymorphism to be used in practical case work, a survey of a given population is necessary. In this study we present data for allele and genotype distributions and mutation rates for MBP in 973 unrelated Korean individuals.

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Materials and methods

DNA was extracted from fresh blood and placenta samples by phenol/chloroform extraction. A total of 238 placenta samples and 735 unrelated Koreans were selected, and family studies for 763 gametes in 550 families were carried out. Additional VNTR and STR markers and a serological test for HLA were performed to confirm pedigrees. The PCR reaction was carried out using two primers described by Polymeropoulous et al. (1992): 5′-GGA CCT CGT GAA TTA CAA TC-3′ and 5′-ATT TAC CTA CCT GTT CAT CC-3′. DNA (20 ng) was amplified in a total volume of 20 µl reaction mixture consisting of 2 µM of each primer, 2 mM dNTP mixture, 2.5 units Taq polymerase, 50 mM KCl, 657 mM Tris-HCl (pH 8.3), 1.5 mM $MgCl_2$ and 0.1% Triton X-100. Amplification condi-

Table 1 Observed allele frequencies for the myelin basic protein locus in region A and region B in 973 unrelated Koreans.

	Region A		Region B	
	Allele	Frequency	Allele	Frequency
	< 1	0.001	7	0.221
	3	0.116	8	0.0001
	3.2	0.0001	9	0.120
	4	0.062	10	0.220
	5	0.189	11	0.357
	6	0.069	12	0.073
	7	0.113	13	0.005
	7.2	0.001		
	8	0.238		
	9	0.128		
	9.2	0.0001		
	10	0.073		
	11	0.005		
Power of Discrimination (PD)	0.959		0.901	
Heterozygosity (HT)	0.857		0.745	
Polymorphism information content (PIC)	0.833		0.718	

Table 2 Observed numbers and haplotype frequencies of the MBP locus in 763 unrelated Koreans

region B region A	7 (frequency)	8 (frequency)	9 (frequency)	10 (frequency)	11 (frequency)	12 (frequency)	13 (frequency)	sum (frequency)
< 1	1 0.0006							1 0.0006
3	9 0.0058		170 0.1114	1 0.0006				180 0.1179
3.2			1 0.0006					1 0.0006
4	22 0.0144		6 0.0039	55 0.036	5 0.0032			88 0.0576
5	282 0.1847		3 0.0019	4 0.0026	1 0.0006			290 0.19
6	19 0.0124		6 0.0039	75 0.0491	1 0.0006			101 0.0661
7				15 0.0098	141 0.0923	3 0.0019		159 0.1041
7.2				1 0.0006	1 0.0006			2 0.0013
8	1 0.0006			151 0.0989	152 0.0996	79 0.0517		383 0.2509
9	1 0.0006			40 0.0262	136 0.0891	22 0.0144	7 0.0045	206 0.1349
9.2					1 0.0006			1 0.0006
10					100 0.0655	8 0.0052	1 0.0006	109 0.0714
11					4 0.0026		1 0.0006	5 0.0032
sum	335 0.2195		186 0.1218	342 0.2241	542 0.3551	112 0.0733	9 0.0058	1526

tions were: 95° C for 10 s, 55° C for 10 s, 72° C for 1 s for 27 cycles on a 9600 GeneAmp PCR system (Perkin Elmer Corporation, Norwalk, USA). The amplified fragments were separated on a 12% polyacrylamide gel and visualized by silver staining.

Results and discussion

The PCR products were visualized as discrete bands in two separate regions: a high molecular weight region (A) ranging from 197 bp to 241 bp, and a low molecular weight region (B) ranging from 121 bp to 145 bp. A heterozygous person had four bands, two in region A and two in region B. A total of 13 alleles in region A and 7 alleles in region B were found. Three 'interalleles' which did not exactly match the allelic ladder, were found in region A. These interalleles with a size difference of less than 4 bp could be easily differentiated. Allele frequencies from the region A and B are shown in Table 1. In region A 50 and in region B 20 genotypes were noted and there was no significant deviation from Hardy-Weinberg expectations (region A; $\chi^2 = 28.67$, df = 27, $p = 0.377$, region B; $\chi^2 = 11.31$, df = 14, $p = 0.661$). Comparison with reports on German (Möller et al. 1994) and Portuguese (Gusmao et al. 1996) showed a obvious population differences. A deficiency of observed heterozygotes in region B reported in Portuguese was not found. Mendelian inheritance was confirmed and no mutations were found by family studies.

The STR MBP locus is peculiar in that two repetitive sequences of a TGGA motif are separated by a non-repetitive sequence, which can be co-amplified with two primers. This increases the usefulness of the STR MBP in individual identification. Both region A and region B were demonstrated to be forensically useful, but as they were closely linked, the haplotype must be considered for statistical analysis. A total of 36 haplotypes and 148 genotypes was found (Table 2) when considering the two regions simultaneously in family studies of 763 gametes, and the degree of polymorphism seemed to be quite high compared to other VNTR or STR loci (heterozygosity = 0.9017, PD = 0.983).

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