SHORT COMMUNICATION

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Population study of the STRs HUMCD4 and HUMF13A1 in Catalonia (Northeast Spain)

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Abstract Allele and genotype frequencies were determined in a population sample from Catalonia (Northeast Spain) for two short tandem repeat loci (HUMCD4 and HUMF13A1), using the polymerase chain reaction (PCR). After denaturing PAG electrophoresis, 6 alleles were identified for HUMCD4 in a sample of 157 unrelated individuals, and 11 alleles for HUMF13A1 in a sample of 141 individuals. No deviation from Hardy-Weinberg equilibrium was found. The HUMCD4 and HUMF13A1 loci demonstrated a heterozygosity of 0.6815 and 0.7305 respectively.

Key words STR · HUMCD4 · HUMF13A1 · Population genetics · Paternity testing

Introduction

In order to obtain data from a Western Mediterranean population, allele frequencies and genotype distributions of the STRs HUMCD4 (Edwards et al. 1991) and HUMF13A1 (Polymeropoulos et al. 1991) were studied in samples from Catalonia (Northeast Spain).

Material and methods

DNA was extracted from blood samples by the phenolchloroform-isoamyl alcohol method (Auxbel et al. 1987).

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D. W. M. Schwartz · B. Glock University Clinic for Blood Group Serology and Transfusion Medicine, University of Vienna, Austria The PCR reaction mixture contained 5–15 ng of template DNA, $10 \times$ buffer (50 mM KCl, 10 mM Tris-HCl pH 9, Triton X-100), 1.5 mM MgCl₂, 1.5 pmol of each primer (HUMCD4: Edwards et al. 1991, and HUMF13A1: Polymeropoulos et al. 1991) and 1 U Taq polymerase, diluted to a total volume of 25 µl with distilled water.

Amplification conditions for HUMCD4 were : 94° C 1 min, 60° C 45 s; 72° C 1 min 30 s for 30 cycles and for HUMF13A1 95^{\circ}C 1 min, 60° C 1 min, 72° C 2 min for 30 cycles.

For both systems separation was carried out on 6% (w/v acrylamide/bisacrylamide) polyacrylamide denaturing high-performance DNA sequencing gels (Ready Mix Gel ALF grade, Pharmacia). The electrophoresis was carried out on the Automated Laser Fluorescent (ALF) DNA sequencer (Pharmacia) at 1450 V, 38 mA, 45 W and 50° C with laser power at 3 mW for 220 min.

The alleles observed for the HUMCD4 system were typed following the nomenclature applied by Glock et al. (1996). Allele designation for the HUMF13A1 system followed allele sizes described by Lygo et al. (1994).

Possible divergence from Hardy-Weinberg equilibrium (HWE) was determined by the exact test proposed by Guo and Thompson (1992). The Catalonia data were compared with other sample populations using an RxC contingency table χ^2 test for homogeneity.

Results and discussion

Genotype and allele frequencies for HUMCD4 and HUMF13A1 obtained in the Catalonian population sample are shown in Table 1.

A total of 14 different genotypes from 6 common alleles were observed for HUMCD4 locus. The alleles 6 and 8 were not observed. The distribution of the genotypes was in Hardy-Weinberg equilibrium (Exact test: P = 0.9111).

A total of 23 genotypes from 11 common alleles were observed for HUMF13A1. The sample studied showed no deviation from Hardy-Weinberg equilibrium (Exact test: P = 0.0786).

Table 1 Allele frequency distribution for HUMCD4 (n = 157) and HUMF13A1 (n = 141) in a Catalonian population and statistical parameters for the two STR systems

Allele	HUMCD4	HUMF13A1	
3.2		0.0709	
4	0.3280	0.0319	
5	0.2834	0.1843	
6		0.3191	
7	0.0095	0.3475	
8		0.0106	
9	0.2961		
10	0.0605		
11	0.0223		
12			
13		0.0035	
14		0.0035	
15		0.0212	
16		0.0035	
17		0.0035	
System	h	PD	CE
HUMCD4	0.6815	0.869	0.466
HUMF13A1	0.7305	0.887	0.506

h: Heterozygosity value

PD: power of discrimination

CE: chance of exclusion

 Table 2
 Comparison of different populations for the HUMF13A1 system

Galicia	$\chi^2 = 1.864$ df = 6 P = 0.91318		
Dutch population	$\chi^2 = 2.257$ df = 6 P = 0.8946	$\chi^2 = 3.036$ df = 6 P = 0.8034	
Norwegians	$\chi^2 = 0.966$ df = 6 P = 0.9868	$\chi^2 = 0.811$ df = 6 P = 0.9897	$\chi^2 = 2.583$ df = 6 P = 0.8592
	Catalonia (present study)	Galicia	Dutch population

Although the nomenclature employed by us for the HUMCD4 system is slightly different, the results obtained from Catalonia are similar to other Caucasoid populations studied (Edwards et al. 1991).

For HUMF13A1, the data obtained in Catalonia were compared with data from populations of Galicia (Pestoni et al. 1995), Netherlands (Sjerps et al. 1995) and Norway (Dupuy et al. 1991) and the results (Table 2) reveal that there is uniformity in European populations for this system.

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