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SHORT COMMUNICATION

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Population study of Aymara Amerindians for the PCR-DNA polymorphisms HUMTH01, HUMVWA31A, D3S1358, D8S1179, D18S51, D19S253, YNZ22 and HLA-DQα

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Abstract Allele and genotype frequencies for eight DNA polymorphisms (HUMTH01, HUMVWA31A, D3S1358, D8S1179, D18S51, D19S253, YNZ22 and HLA-DQα) were determined in a population sample of Aymara Indians from Bolivia using PCR. No deviations of the observed allelic frequencies from Hardy-Weinberg equilibrium were found for all the systems studied. Significant differences in the allele frequencies were found between the Aymara and Quechua populations only for HUMVWA31A, which suggests a certain degree of genetic differentiation between the two populations.

Key words HUMTH01 · HUMVWA31A · D3S1358 · D8S1179 · D18S51 · D19S253 · YNZ22 · HLA-DQα · Aymara

Introduction

DNA polymorphisms have been widely studied in many human populations [1, 2]. However, there is a scarcity of data for autochthonous South American Indians. The aim of this study was to report the allele frequency distributions for HUMTH01, HUMVWA31A, D3S1358, D8S1179, D18S51, D19S253, YNZ22 and HLA-DQα in the Aymara population.

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The Aymara people constitute a large South American Indian group living in the vast windy Titicaca plateau of the Central Andes of modern Peru and Bolivia. The Aymara studied live at an altitude of 3835 m in the provinces of Pacajes and Murillo in the department of La Paz (Bolivia).

Material and methods

The biological samples analysed consisted of two hairs with root bulbs taken from unrelated individuals of both sexes from the Aymara population. DNA was extracted with Chelex 100 using the method described by Walsh et al.[3]. The HLA-DQα typing was done with the AmpliType kit. Genotyping methods used for the other PCR-DNA polymorphisms were as described in previous studies on Catalonian populations [1, 4, 5]. For the D19S253 system, the alleles were scored following the terminology used by De Stefano et al. [6].

Possible divergence from Hardy-Weinberg equilibrium (HWE) was determined by using the exact test proposed by Guo and Thompson [7].

From a forensic point of view, the power of discrimination (PD)[8], heterozygosity value (h)[9] and the "a priori" chance exclusion value (CE)[10] were calculated. The Aymara data were compared with those from a Quechua population using a $R \times C$ contingency table χ^2-test for homogeneity [11].

Results and discussion

Allele frequencies for the eight DNA systems studied in the Aymara population sample are shown in Table 1. The distribution of the genotypes for all loci were in Hardy-Weinberg equilibrium. The results were compared with those of Quechua Indians, previously studied by our group [12], as another important population living in the same geographic area.

The comparisons between the Aymara and Quechua have been carried out only with the four commonly published polymorphisms (HUMTH01, HUMVWA31A, YNZ22 and HLA-DQα). No statistically significant differences were observed for HUMTH01 ($P = 0.7755$), YNZ22 ($P = 0.1508$), and HLA-DQα ($P = 0.7699$) but significant differences were observed for the HUMVWA31A system

Table 1 Allele frequency distribution for HUMTH01, HUMVWA31A, D3S1358, D8S1179, D18S51, D19S253, YNZ22 and HLA-DQα", in Aymara population and statistical parameters for the eight systems

Allele	HUMTH01 *n* = 115	HUMVWA31A *n* = 110	D3S1358 *n* = 117	D8S1179 *n* = 115	D18S51 *n* = 104	D19S253 *n* = 114	YNZ22 *n* = 117	HLA-DQα *n* = 57
1.1								0.026
1.2								0.009
1.3								
2							0.009	
3							0.030	0.474
4							0.812	0.491
5							0.047	
6	0.217						0.021	
7	0.600					0.079	0.004	
8						0.004	0.009	
9	0.013			0.009			0.009	
9.3 + 10	0.170							
10				0.052	0.010	0.105	0.017	
11				0.048	0.005	0.140	0.013	
12				0.126	0.154	0.482	0.009	
13				0.335	0.197	0.158	0.004	
14		0.014	0.026	0.230	0.197	0.031	0.013	
15		0.041	0.603	0.170	0.111		0.004	
16		0.395	0.261	0.026	0.091			
17		0.450	0.103	0.004	0.125			
18		0.059	0.004		0.053			
19		0.036	0.004		0.034			
20		0.005			0.014			
21					0.010			
HWE exact test	*P* = 0.5136	*P* = 0.4596	*P* = 0.9603	*P* = 0.1801	*P* = 0.7088	*P* = 0.6289	*P* = 0.0701	*P* = 0.6390

Table 2 Statistical parameters of medico-legal interest (*h* heterozygosity value, *PD* power of discrimination, *CE* chance of exclusion)

System	h	PD	CE
HUMTH01	0.566	0.722	0.310
HUMVWA31A	0.637	0.798	0.370
D3S1358	0.560	0.744	0.308
D8S1179	0.787	0.923	0.587
D18S51	0.862	0.963	0.715
D19S253	0.707	0.880	0.493
YNZ22	0.337	0.553	0.204
HLA-DQα	0.538	0.673	0.231
Total		0.999	0.989

(*P* = 5.017E-04). To confirm the results for this locus all samples were typed twice. At the moment, there is no explanation for this difference. However, the similarities observed suggest some genetic affinities between the two groups. Thus, from a genetic point of view, except for the HUMVWA31A system, the allele distributions for the rest of the systems analysed are comparable with those observed in the Quechua population. Nevertheless, the analysis of more polymorphisms will be necessary to determine if genetic differences exist between the two groups.

For forensic purposes, statistical parameters of medico-legal interest were calculated from the gene frequencies obtained in our population (Table 2).

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