

HLA-DQA1 POLYMORPHISM IN TWO PORTUGUESE POPULATION SAMPLES FROM LISBON AND THE SOUTH OF PORTUGAL

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INTRODUCTION

The HLA-DQA1 polymorphism provides a powerful tool to forensic analysis.

The purpose of this study was to analyse the HLA-DQA1 locus in view of its application in forensic casework, as recommended by the DNA Commission of the International Society for Forensic Haemogenetics (1992).

We describe the allele and genotype frequencies of two portuguese population samples from Lisbon and from the South of Portugal that have been investigated for HLA-DQA1 locus by the polymerase chain reaction (PCR), (Saiki et al, 1986; Comey et al,1991), using allele specific oligonucleotides and reverse dot-blot methodology (Cetus).

MATERIAL AND METHODS

Blood samples from 120 unrelated individuals from Lisbon and 104 unrelated individuals from the South of Portugal were studied.

DNA was extracted from the blood stains using a chelating resin according to the method developed by Singer-Sam et al, (1989).

Amplification of the DNA samples and allele typing were performed using the Amplitype HLA-DQ Alpha Forensic Amplification and Typing Kit (Perkin-Elmer Cetus).

RESULTS AND DISCUSSION

The distribution of observed and expected HLA-DQA1 genotypes and the allele frequencies for the two portuguese population samples are shown in Table 1, and no deviation from Hardy-Weinberg equilibrium was observed.

The similarity between the heterozygosity rate and the allelic diversity value (h) of both population samples is in good agreement with Hardy-Weinberg equilibrium (Table 2).

Table 1. Distribution of observed and expected HLA-DQA1 genotypes and allele frequencies.

GENOTYPE	LISBON (n=120)		SOUTH PORTUGAL (n=104)	
	observed	expected	observed	expected
1.1-1.1	6	4.22	1	1.88
1.1-1.2	4	4.69	4	4.58
1.1-1.3	0	4.50	2	3.23
1.1-2	6	6.37	6	5.25
1.1-3	9	6.19	5	3.90
1.1-4	14	14.81	9	7.27
1.2-1.2	1	1.31	3	2.78
1.2-1.3	4	2.50	5	3.92
1.2-2	2	3.54	8	6.38
1.2-3	3	3.44	4	4.74
1.2-4	10	8.23	7	8.83
1.3-1.3	0	1.20	2	1.38
1.3-2	6	3.40	5	4.50
1.3-3	4	3.30	5	3.35
1.3-4	10	7.90	3	6.23
2-2	3	2.41	2	3.66
2-3	6	4.68	6	5.44
2-4	8	11.20	10	10.13
3-3	2	2.27	0	2.02
3-4	7	10.86	9	7.53
4-4	15	13.00	8	7.01
χ^2 df=15	15.81 0.25<p<0.50		9.09 0.75<p<0.90	

	ALLELES					
	1.1	1.2	1.3	2	3	4
Lisbon	0.1875	0.1042	0.1000	0.1417	0.1375	0.3291
South Portugal	0.1346	0.1635	0.1154	0.1875	0.1394	0.2596

Table 2. Statistical parameters.

	PD	h	Heterozygosity
Lisbon	0.931	0.800	0.775
South Portugal	0.942	0.824	0.846

The power of discrimination (PD) was estimated for Lisbon population sample (0.931), and for the South of Portugal population sample (0.942).

The data showed that for both samples the most frequent allele was HLA-DQA1*4 and the less frequent allele was HLA-DQA1*1.3. Differences in allele distributions were observed for the HLA-DQA1*1.1 allele (Lisbon= 0.1875; South Portugal=0.1346) and the HLA-DQA1*1.2 allele (Lisbon= 0.1042; South Portugal=0.1635)

The allele frequencies distribution of both population samples were similar to other Caucasian data, mainly from Portugal and Spain (Lareu et al, 1993; Martin et al, 1993; Pascal et al, 1993; Sanz et al, 1993).

Acknowledgements: The authors are grateful to the Department of Biology, Institute of Legal Medicine, University of Coimbra for providing useful information.

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