THE DISTRIBUTION OF HLA DQA1 ALLELES IN THE POPULATION OF THE NORTH OF PORTUGAL

M. F. Pinheiro [1,2], M. L. Pontes [1]

1 Medico-Legal Institute, Oporto
2 Biomedical Sciences Institute Abel Salazar, University of Oporto

INTRODUCTION

DNA profiling has rapidly become a routine technique in forensic laboratories worldwide since its introduction in 1985. One of the PCR strategies that have been developed for forensic application is the dot-blot analysis of the human leucocyte antigen (HLA) DQα Locus (Saiki et al, 1986; Westwood et al 1990), now redesignated as DQA1.
The DQA1 typing system has been used to study many populations (Helmuth et al, 1990; Sullivan et al, 1992; Lareu et al, in press). We report here the allele and genotype frequencies distribution of a Portugal subgroup population, North of Portugal.

MATERIAL AND METHODS

Blood samples were taken from 325 unrelated individuals of routine paternity cases. DNA was extracted by a method previously described by Singer-Sam and col. (1989) using a chelating resin. Some of these samples were extracted using a phenol-chloroform procedure. PCR amplification was performed using HLA DQα Amplitype Kit (Cetus) and the protocols provied by the manufacturer. The cycling reaction was done in a programmable heat block (TC 480 Perkin Elmer - Cetus Thermal Cycler).
Typing of HLA DQA1 alleles was achieved using allele specific oligonucleotide probes (ASO probes) and reverse dot blot methodology (Cetus Amplitype User Guide).

RESULTS AND DISCUSSION

The genotype and allele frequencies distribution indicates that there is no deviation from the Hardy-Weinberg equilibrium.
The allele frequencies range from 0.0923 (allele DQA1 * 1.3) to 0.2477 (allele DQA1 * 04) (Table 1).

Table 1. Allele and genotype frequencies of the HLA DQA1 system in the population of North of Portugal

	Observed		Expected	
Genotypes	n	%	n	%
1.1 - 1.1	13	4.00	9.65	2.97
1.1 - 1.2	12	3.69	17.23	5.30
1.1 - 1.3	7	2.15	10.34	3.18
1.1 - 2	12	3.69	19.64	6.04
1.1 - 3	20	6.15	17.75	5.46
1.1 - 4	35	10.77	27.74	8.53
1.2 - 1.2	10	3.08	7.69	2.37
1.2 - 1.3	10	3.08	9.23	2.84
1.2 - 2	18	5.54	17.54	5.39
1.2 - 3	15	4.61	15.84	4.87
1.2 - 4	25	7.69	24.77	7.62
1.3 - 1.3	2	0.61	2.77	0.85
1.3 - 2	14	4.31	10.52	3.24
1.3 - 3	14	4.31	9.51	2.93
1.3 - 4	11	3.38	14.86	4.57
2 - 2	11	3.38	9.99	3.07
2 - 3	20	6.15	18.06	5.56
2 - 4	28	8.61	28.23	8.69
3 - 3	6	1.85	8.16	2.51
3 - 4	22	6.77	25.51	7.85
4 - 4	20	6.15	19.94	6.13
	Total 325			
1.1 = 0.1723	**2** = 0.1754		x^2 = 15,66	
1.2 = 0.1538	**3** = 0.1585		df = 15	
1.3 = 0.0923	**4** = 0.2477		P > 0,40	

The theoretical a priori chance exclusion value (C.E.) is 0.6399 from genotype data, the power of discrimination (P.D.) according to the equation suggested by Fisher (1951) is 0.9427. The allelic diversity value (h) as described by Nei and Rouchoudhoury (1974) is 0.8209 (Table 2).

Table 2. Statistical parameteres of HLADQA1 system.

CE	0.6399
PD	0.9427
h	0.8209
Heterozygosity	0.8092

When compared to other Caucasian data (Helmuth et al 1990; Gené et al, 1992; De Stefano et al 1992) the allelic frequencies of the most and fewest frequent alleles are quite similar, but other variants present some differences even in populations geographically very close, such as NW Spain and Central Portugal (Lareu et al, in press). Relatively to other ethnic populations these differences are significantly larger.

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