

Study of the HLA DQ α polymorphism in the population of Catalonia (Spain)

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INTRODUCTION

We report here the results of a survey of HLA DQ α polymorphism studied by the polymerase chain reaction (PCR) in 110 samples from people living in Catalonia (North-east Spain). In agreement with others, we conclude that it is a useful marker in forensic evidence analysis and in paternity testing.

MATERIAL AND METHODS

The study was carried out with blood donor samples obtained from a Blood Bank, "Hospital Clínic", Barcelona, and from unrelated paternity cases. The HLA DQ α alleles were characterized after enzymatic PCR amplification of specific DNA sequences, using allele specific oligonucleotide probes (ASO probes) and reverse dot-blot methodology (Cetus 1990). The method identifies four common types of alleles called DQ α 1, 2, 3, 4, and distinguishes three genetic variants (subtypes) of allele 1 (DQ α 1.1, 1.2, 1.3), defining a total of twenty-one different genotypes.

RESULTS AND DISCUSSION

Calculation of the phenotype distribution and gene frequencies indicates that there is no deviation from the Hardy-Weinberg equilibrium (Chi-square of 8.64 with 15 degrees of freedom, and $p > 0.75$). The results of population studies are given in Table 1. The gene frequencies of the twenty-one genotypes range from less than 0.09 (allele 1.3) to 0.15 (alleles 1.1 and 1.2) except for allele 4 in which the frequency was 0.37.

When compared with the frequencies given from Cetus and Roche Biomedical Laboratories (Cetus 1990), the distribution of some of the variants shows few differences between Catalans and other caucasians, but larger ones in relation to other ethnic groups.

We have performed the HLA DQ α polymorphism in 52 paternity testing cases. 35 cases of paternity practically proved ($W \geq 99.73\%$) using conventional markers were confirmed, supporting the assumed autosomal codominant way of inheritance. The percentage of exclusions (11 incompatibilities in 17 cases of

non paternity 64.7%) using this method proved the theoretical a priori chance exclusion value (C.E.= 0.59) obtained from gene frequencies in our population.

Table 1. Hardy Weinberg equilibrium for the locus HLA DQ α

Types	Obs.	Exp.	Dif.	$\frac{2}{X}$
1.1 - 1.1	3	2.63	0.37	0.05
1.1 - 1.2	3	5.10	-2.10	0.86
1.1 - 1.3	5	3.25	1.75	0.95
1.1 - 2	5	4.17	0.83	0.16
1.1 - 3	4	3.55	0.45	0.06
1.1 - 4	11	12.67	-1.67	0.22
1.2 - 1.2	2	2.47	-0.47	0.09
1.2 - 1.3	2	3.15	-1.15	0.42
1.2 - 2	6	4.05	1.95	0.94
1.2 - 3	4	3.45	0.55	0.09
1.2 - 4	14	12.30	1.70	0.23
1.3 - 1.3	1	1.00	-0.00	0.00
1.3 - 2	3	2.58	0.42	0.07
1.3 - 3	3	2.20	0.80	0.30
1.3 - 4	6	7.83	-1.83	0.43
2 - 2	1	1.66	-0.66	0.26
2 - 3	0	2.82	-2.82	2.82
2 - 4	11	10.06	0.94	0.09
3 - 3	2	1.20	0.80	0.53
3 - 4	8	8.57	-0.57	0.04
4 - 4	16	15.28	0.72	0.03
Total	110	109.99		8.64
HLA DQ α 1.1 =	0.1545			2
HLA DQ α 1.2 =	0.1500			X = 8.64
HLA DQ α 1.3 =	0.0955			df = 15
HLA DQ α 2 =	0.1227			P > 0.75
HLA DQ α 3 =	0.1045			CE = 0.59
HLA DQ α 4 =	0.3727			EM value = 9.55

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