

POPULATION STUDY FOR THE HLA-DQA1, LDLR, GYPA, HBGG, D7S8 AND GC LOCI IN NORTH-EAST OF SPAIN

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System and locus: PM loci: low density protein receptor (LDLR)
glycophorin A (GYPA)
hemoglobin G gammaglobin (HBGG)
D7S8
group specific component (GC)
HLA-DQA1 locus

Population and sample: Catalonia (N.E. of Spain)
N: 146 (PM loci), 195 (HLA-DQA1)

Results:

Primers and PCR amplification conditions: Amplitype® PM and the HLA-DQ α PCR Amplification and Typing kit (Perkin-Elmer) in a Linus DualCycler thermocycler. All samples for the HLA-DQA1 locus were single amplified.

Characterization of the alleles: by hybridization to filter strips carrying immobilised allele specific oligonucleotide DNA probes (reverse dot-blot) using the strips and recommendations of the manufacturer.

Results:

Table 1. Allele frequencies

Locus	Allele	Frequency	Allele	Frequency	Allele	Frequency
LDLR	A	0.48288	B	0.51712		
GYPA	A	0.51027	B	0.48973		
HBGG	A	0.46233	B	0.51712	C	0.02055
D7S8	A	0.55137	B	0.44863		
GC	A	0.33219	B	0.16096	C	0.50685
HLA-DQA1	1.1	0.16154	1.2	0.13846	1.3	0.05897
	2	0.20000	3	0.12051	4	0.32051

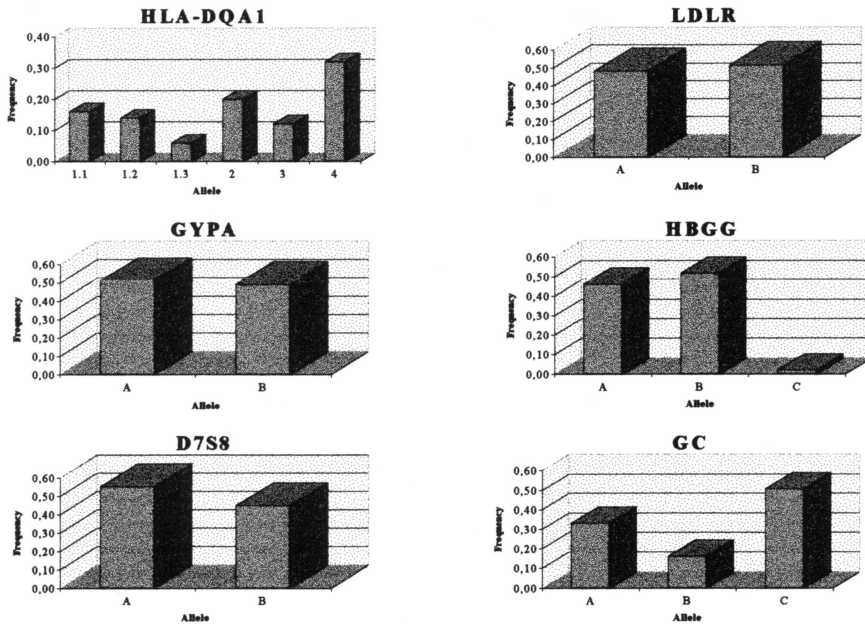


Figure 1. Distribution of HLA-DQA1 and PM loci alleles in North-East of Spain

Table 2. Observed genotypes

	Gen.	Obs.	Gen.	Obs.	Gen.	Obs.
LDLR	A-A	38	A-B	65	B-B	43
GYPA	A-A	39	A-B	71	B-B	36
HBGG	A-A	28	A-B	75	A-C	4
	B-B	37	B-C	2		
D7S8	A-A	49	A-B	63	B-B	34
GC	A-A	18	A-B	12	A-C	49
	B-B	5	B-C	25	C-C	37
HLA-DQA1	1.1-1.1	6	1.1-1.2	7	1.1-1.3	4
	1.1-2	16	1.1-3	6	1.1-4	18
	1.2-1.2	5	1.2-1.3	2	1.2-2	13
	1.2-3	7	1.2-4	15	1.3-1.3	1
	1.3-2	3	1.3-3	2	1.3-4	10
	2-2	8	2-3	8	2-4	22
	3-3	5	3-4	14	4-4	23

Table 3. Evaluation

Locus	Power of discrimination	Power of exclusion	Allelic diversity h (\pm s.e.) [3]	P value
	PD [1]	PE [2]		Chi square test
HLA-DQA1	0.929	0.599	0.796 \pm 0.029	0.91
LDLR	0.625	0.187	0.501 \pm 0.041	0.19
GYP A	0.625	0.187	0.502 \pm 0.041	0.74
HBGG	0.653	0.213	0.520 \pm 0.041	0.61
D7S8	0.622	0.186	0.496 \pm 0.041	0.12
GC	0.770	0.322	0.609 \pm 0.040	0.68
Combined	0.9997	0.885		

Comments:

The distribution of phenotypes is in agreement with Hardy-Weimberg expectations for the six loci. The Amplitype® PM and the HLA-DQ α PCR Amplification and Typing kit provide two important advantages: they can be used as a fast and high sensitive screening of the samples; on the other hand, we need less template DNA to type six loci at a time with a high combined power of discrimination.

We use in routinely forensic casework this multiplex system.

References:

1. Jones DA (1972) Blood samples: probability of discrimination. *J Forensic Sci Soc* 12:355-359
2. Garber RA, Morris JW (1983) General equations for the average power of exclusion for genetic systems of n codominants alleles in one-parent and no-parent cases of disputed parentage. In: Walker RH (ed) *Inclusion probabilities in parentage testing*. American Association of Blood Banks, Arlington, pp 277-280
3. Nei M, Roychoudhury AK (1974) Sampling variances of heterozygosity and genetic distance. *Genetics* 76:379-390