

ALLELE FREQUENCY DISTRIBUTION OF 15 PCR-BASED DNA POLYMORPHISMS IN THE POPULATION OF GALICIA (NW SPAIN)

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Systems and loci: HLA DQA1 (6p21.3), LDLR (Cr 19), GYPA (Cr 4), HBGG (Cr 11), D7S8, GC (Cr 4), pMCT (D1S80), YNZ22 (D17S5), COL2A1 (12q14.3), 3'ApoB (Cr 2), HUMTH01 (11p15.5-p15), HUMVWA31/A (12p12-12pter), HUMF13A1 (6p24-25), HUMFES/FPS (15q25-qter), HUMLPL (8q22).

Population and sample size: 140 to 250 unrelated people from Galicia (NW Spain).

Methods:

Primers and PCR amplification conditions: HLA DQA1 and pMCT (Lareu et al. 1993), LDLR, GYPA, HBGG, D7S8 and GC (AmpliType PM PCR Amplification and Typing kit (Perkin Elmer)), YNZ22 (Rand et al. 1992), COL2A1 (Wu et al. 1990), 3'ApoB (Boerwinkle et al. 1989), HUMTH01, HUMVWA31/A, HUMF13A1, HUMFES/FPS and HUMLPL (Pestoni et al. 1995).

Method of detection: HLA DQA1, LDLR, GYPA, HBGG, D7S8 and GC (Dot-blot with ASO probes - AmpliType HLA DQ α and PM kits (Perkin-Elmer)), pMCT and YNZ22 (PAGE (PhasGels 8-25, Pharmacia) followed by silver-staining), COL2A1, 3'ApoB, HUMTH01, HUMVWA31/A, HUMF13A1, HUMFES/FPS and HUMLPL (ALF DNA sequencer, Pharmacia).

Results:

Allele frequencies:

HLA DQA1 (n=178)

Allele	Frequency	Allele	Frequency	Allele	Frequency
0101	0.1826	0103	0.0955	0301	0.0871
0102	0.1966	0201	0.1039	0501	0.3343

χ^2 : 13.3124; p:0.5781

POLYMARKER (n=143)

Locus	Allele	Frequency	Locus	Allele	Frequency	Locus	Allele	Frequency
LDLR	A	0.3986	GYP	A	0.4825	D7S8	A	0.5594
	B	0.6014		B	0.5175		B	0.4406

χ^2 :0.009; p:0.924

χ^2 :0.188; p:0.665

χ^2 :0.007; p:0.934

Locus	Allele	Frequency	Locus	Allele	Frequency
HBGG	A	0.5909	GC	A	0.2762
	B	0.3986		B	0.1748
	C	0.0105		C	0.5490

χ^2 :1.861; p:0.602

χ^2 :0.553; p:0.907

D1S80 (n=149)

Allele	Frequency	Allele	Frequency	Allele	Frequency
16	0.0034	23	0.0235	30	0.0101
17	0.0067	24	0.3557	31	0.0403
18	0.2617	25	0.0302	32	0.0034
19	0.0034	26	0.0034	33	0.0101
20	0.0336	27	0.0201	34	0.0067
21	0.0302	28	0.0268	36	0.0101
22	0.0436	29	0.0738	37	0.0034

Exact test: p:0.193

YNZ22 (n=137)

Allele	Frequency	Allele	Frequency	Allele	Frequency
1	0.0365	5	0.0438	9	0.1058
2	0.1934	6	0.0511	10	0.0620
3	0.1934	7	0.0146	11	0.0182
4	0.2518	8	0.0255	12	0.0036

Exact test: p:0.168

COL2A1 (n=164)

Allele	Frequency	Allele	Frequency	Allele	Frequency
11	0.1311	13	0.5579	15	0.0244
12	0.0366	14	0.2409	16	0.0091

Exact test: p:0.674

3'ApoB (n=183)

Allele	Frequency	Allele	Frequency	Allele	Frequency
25	0.0027	35	0.2322	43	0.0191
29	0.0055	36	0.0027	45	0.0574
30	0.0055	37	0.3852	47	0.0956
31	0.0765	39	0.0301	49	0.0109
33	0.0574	41	0.0191		

Exact test: p:0.124

HUMTH01 (n=234)

Allele	Frequency	Allele	Frequency	Allele	Frequency
5	0.0021	7	0.1731	9	0.1774
6	0.2115	8	0.1325	9.3	0.3034

Exact test: p:0.928

HUMVWA31/A (n=158)

Allele	Frequency	Allele	Frequency	Allele	Frequency
14	0.1297	17	0.2247	20	0.0095
15	0.1108	18	0.2247		
16	0.2595	19	0.0411		

Exact test: p:0.857

HUMF13A1 (n=143)

Allele	Frequency	Allele	Frequency	Allele	Frequency
3.2	0.0874	7	0.3671	12	0.0035
4	0.0385	8	0.0070	15	0.0175
5	0.1748	10	0.0035	16	0.0070
6	0.2832	11	0.0035	17	0.0070

Exact test: p:0.708

HUMFES/FPS (n=124)

Allele	Frequency	Allele	Frequency	Allele	Frequency
8	0.0121	10	0.3306	12	0.1815
9	0.0081	11	0.4193	13	0.0484

Exact test: p:0.723

HUMLPL (n=113)

Allele	Frequency	Allele	Frequency	Allele	Frequency
1	0.0885	3	0.2611	5	0.0796
2	0.3717	4	0.1903	6	0.0088

Exact test: p:0.695

Comments: All the systems were in Hardy-weinberg equilibrium. No significant differences with other caucasian populations were found except for HLA DQA1. No mutations were found for any of the systems in a total of 95 paternal meioses and 120 maternal meioses. Anomalous electrophoretic mobility was observed for HUMF13A1 and 3'ApoB systems in non-denaturing PAGE.

Systems	<i>h</i>	PD	CE	Systems	<i>h</i>	PD	CE
HLADQA1	0.7914	0.9211	0.5932	YNZ22	0.8427	0.9453	0.6828
LDLR	0.4755	0.6141	0.1823	COL2A1	0.6133	0.7946	0.3689
GYPA	0.5174	0.6247	0.1873	HUMTH01	0.7859	0.9195	0.5737
HBGG	0.5454	0.6307	0.1960	HUMVWA31/A	0.8033	0.9308	0.6052
D7S8	0.4895	0.6214	0.1857	HUMF13A1	0.7475	0.8956	0.5226
GC	0.5944	0.7643	0.3174	HUMFES/FPS	0.6821	0.8392	0.4172
D1S80	0.7936	0.9342	0.6189	HUMLPL	0.7465	0.8932	0.5160
3'ApoB	0.7764	0.9238	0.5889				

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