Allele frequencies of HLA-DQα, LDLR, GYPA, HBGG, D7S8 and GC in the resident and autochthonous populations of the Basque Country

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Systems and loci: HLA-DQα (6p21.3)
LDLR (19p13.1→p13.3)
GYPA (4q28→q31)
HBGG (11q)
D7S8 (7cen→q22)
GC (4q11→q13)

Populations and sample size: Basque Country resident population N = 208
Basque Country autochthonous population N = 206

Methods

DNA was extracted by the chelex method. The amplification by PCR and the typing by reverse dot-blot methodology using ASO probes were performed according to the AmpliType HLA DQα and AmpliType PM PCR Amplification and Typing kit protocol provided by the manufacturer (Perkin-Elmer Cetus). All samples carrying the allele 4 in the HLA-DQα system were digested using 2 restriction enzymes (Rsa I and Fok I) [1]. The digested fragments were separated by vertical discontinuous polyacrylamide gel electrophoresis using the Tris-chloride/Tris-glycine buffer system [2] and detected by silver stain.

Results

Observed allele frequency distribution for HLA-DQα locus

Allele	Autochthonous	Residents
1.1	0.190	0.156
1.2	0.180	0.184
1.3	0.101	0.087
2	0.204	0.158
3	0.108	0.153
4.1	0.180	0.238
4.2	0.034	0.021
4.3	0.002	0.002

HWE test for independence on HLA-DQα locus

	Autochthonous	Residents
Obs. Homozygosity	20.7 %	19.8 %
Exp. Homozygosity[a] [3]	16.4 %	16.9 %
Homozygosity test[b] [4-6]	0.095	0.265
Likelihood ratio test[b] [3,7-8]	0.224	0.839
Exact test[b] [9]	0.205	0.900

[a] Expected homozygosity is an unbiased estimate
[b] These values are probability values

Observed allele frequency distribution for PM loci

Allele	Autochtonous	Residents
LDLR*A	0.524	0.438
LDLR*B	0.476	0.562
GYPA*A	0.541	0.514
GYPA*B	0.459	0.486
HBGG*A	0.524	0.534
HBGG*B	0.476	0.459
HBGG*C	0.000	0.007
D7S8*A	0.515	0.575
D7S8*B	0.485	0.425
GC*A	0.330	0.339
GC*B	0.104	0.096
GC*C	0.566	0.565

HWE test for independence on PM loci in a Basque Country autochthonous sample population

	LDLR	GYPA	HBGG	D7S8	GC
Obs. Homozygosity	47.6 %	51.9 %	55.3 %	46.6 %	38.4 %
Exp. Homozygosity[a]	50.0 %	50.2 %	50.0 %	49.9 %	43.8 %
Homozygosity test[b]	0.487	0.621	0.125	0.341	0.113
Likelihood ratio test[b]	0.471	0.663	0.147	0.364	0.033*
Exact test[b]	0.570	0.663	0.116	0.429	0.035*

[a] Expected homozygosity is an unbiased estimate
[b] These values are probability values

HWE test for independence on PM loci in a Basque Country resident sample population

	LDLR	GYPA	HBGG	D7S8	GC
Obs. Homozygosity	54.8 %	46.2 %	61.1 %	48.6 %	47.1 %
Exp. Homozygosity[a]	50.7 %	49.9 %	49.4 %	51.0 %	44.2 %
Homozygosity test[b]	0.232	0.277	0.001*	0.482	0.396
Likelihood ratio test[b]	0.253	0.277	0.003*	0.476	0.095
Exact test[b]	0.253	0.337	0.003*	0.476	0.054

[a] Expected homozygosity is an unbiased estimate
[b] These values are probability values

Discussion

The genotype frequency distributions for most of the loci do not deviate from HWE based on the homozygosity test, likelihood ratio test and the exact test. Those loci that depart from HWE are marked with an asterisk. It should be noted that even for the highly significant departure from HWE for HBGG in the resident population, there would still be little impact for forensic identity purposes. The differences between observed and expected genotype frequencies in this case would not substantially alter the estimated rarity of a DNA profile and thus would not result in bias to an accused individual. An interclass correlation test analysis [10] demonstrated that there is little evidence for correlation between the alleles at any pairs of loci (data not shown).

In conclusion, a Basque Country population database has been established for six PCR-based polymorphic loci. The data can be used for deriving estimates of multiple locus profile frequencies for identity testing purposes using the product rule under the assumption of independence.

References

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