ALLELE DISTRIBUTION OF THE AMPLITYPE PM COAMPLIFICATION SYSTEM IN A POPULATION OF NORTHERN ITALY

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INTRODUCTION

Forensic application of any genetic marker requires a study to carry out a data base of the relevant population for a correct use of the analysis results. According to this guideline the allele distribution of 5 different PCR polymorphisms (LDLR, GYPA, HBGG, D7S8 and GC) were investigated.
LDLR, the Low Density Lipoprotein Receptor, is a two codominant allele (A and B) system that is placed on Cr. 19. His PCR product give band at 214 bp.
GYPA, Glycophorin A, is a system with two alleles (A and B) but additional low frequency alleles are identified in the African American population. It is placed on CR. 4 and his PCR product is setting at 190 bp.
HBGG, the Hemoglobin G Gammaglobin, with a PCR product of 172 bp, is a three (A, B and C) system placed on Cr. 11.
D7S8, a locus of Cr. 7, shows two alleles (A and B) at 151 bp.
Finally GC, the Group Specific Component (GC) is composed by three different alleles (A, B and C) corrisponding to the IEF: 2, 1F and 1S, located on Cr. 4, and are visible at 138 bp.

MATHERIAL AND METHODS

For genotype frequency determination DNA was extracted from whole blood (drawn in EDTA tubes) from 100 unrelated individuals living in Brescia area (Northern Italy)
DNA extraction and purification was carried out by standard protocols using Phenol/ Chloform reagents. The samples (100 microlitres total volume) have been amplified by using Amplitype PM Kit by Perkin Elmer following the manufacturer' s recommended protocol . Amplification was carried out with DNA Thermal Cycler - Perkin Elmer ; the amplified fragments were controlled in a 2% agarose gel electrophoresis in TBE 1X and the allele resolution was performed with the reverse Dot Blot technique.

RESULTS AND DISCUSSION

The observed genotypes , the expecrted and the allele frequencies of the systems are shown in Table 1 (LDLR, GYPA and D7S8) and Table 2 (HBGG and GC). The results of the statistical analysis demonstate no deviation from Hardy Weinberg expectation.
In LDLR system the most frequent allele is B, while, either in GYPA or in D7S8 is A.
About the two systems with three alleles we note that in HBGG the most frequent allele is A followed by B and C, while in GC, C is followed by A and B.
Oue allele distribution is similar to other Caucasian population except for HBGG system where the data obtained are different from Swiss population.

TABLE 1

Genotypes	Observed	Expected	X	Allele Frequencies
LDLR				
AA	16	15.61	0.0097	A = 0.395
AB	37	37.60	0.0044	
BB	47	47.79	0.0130	B = 0.605
Total	100	100	0.0271	0.7<p<0.9
GYPA				
AA	34	30.25	0.4649	A = 0.550
AB	24	20.25	0.6945	
BB	42	49.50	1.1363	B = 0.450
Total	100	100	2.2957	0.1< p<0.2
D7S8				
AA	44	42.90	0.0282	A = 0.655
AB	13	11.90	0.1017	
BB	43	45.20	0.1071	B = 0.345
Total	100	100	0.2370	0.5<p<0.7

TABLE 2

Genotypes	Observed	Expected	X	Allele Frequencies
HBGG				
AA	20	25.50	1.1863	A = 0.505
AB	60	48.99	2.4698	
BB	18	23.50	1.2955	B = 0.485
AC	1	1.01	0.0001	
BC	1	0.97	0.0000	C = 0.010
CC	-	0.01	0.0100	
Total	100	100	4.9616	0.1<p<0.2
GC				
AA	7	8.12	0.1545	A = 0.285
AB	7	10.83	1.3545	
BB	3	3.61	0.1030	B = 0.190
AC	36	29.93	1.2310	
BC	25	19.95	1.2783	C = 0.525
CC	22	27.56	1.2616	
Total	100	100	5.2429	0.1<p<0.2

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Systems and Locus:

LDLR	Cr. 19	214 bp
GYPA	Cr. 4	190 bp
HBGG	Cr. 11	172 bp
D7S8	Cr. 7	151 bp
GC	Cr. 4	138 bp

Population and sample size: 100 unrelated individuals living in Brescia area (Northern Italy)

Methods:

- DNA extraction: Phenol/ Chloform
- Amplification by using Amplitype PM Kit by Perkin Elmer
- Allele resolution: reverse Dot Blot technique.

Results:

LDLR
A = 0.95
B = 0.605

GYPA
A = 0.55
B = 0.45

D7S8
A = 0.655
B = 0.345

HBGG
A = 0.505
B = 0.485
C = 0.010

GC
A = 0.285
B = 0.190
C = 0.525

Comments:

Our allele distribution is similar to other Caucasian population except for HBGG system where the data obtained are different from Swiss population.