SHORT COMMUNICATION

A. Tagliabracci · L. Buscemi · N. Cerri · N. Cucurachi
R. Lombardi · R. Mignola · T. M. Neri · C. Vecchiotti
F. De Ferrari · G. Masotti · D. Rodriguez
G. Umani Ronchi

Italian population data on the loci LDLR, GYPA, HBGG, D7S8 and GC

Abstract Allele and genotype frequencies for the five loci LDLR, GYPA, HBGG, D7S8 and GC were determined for 374 unrelated Italians using a multiplex PCR-amplification and typing commercial kit. The distribution of the genotype frequencies showed no deviations from Hardy-Weinberg expectations. The combined power of discrimination and chance of exclusion for all five loci were 0.999 and 0.702, respectively. A test for homogeneity was performed and no significant differences were observed among the Caucasian population samples.

Key words PM loci (LDLR, GYPA, HBGG, D7S8, GC) · PCR · DNA polymorphism · Population genetics

Introduction

The Amplitype PM PCR amplification and typing kit (Perkin Elmer) coamplifies and simultaneously types the five loci LDLR [1], GYPA [2], HBGG [3], D7S8 [4] and Gc [5]. For routine use of these genetic markers in forensic casework and paternity testing, population data are needed. The purpose of this study was to establish a data-base of Italian allele frequencies and to compare the data with other population studies for homogeneity.

Materials and methods

Blood was obtained from 374 unrelated donors living in Northern (Brescia $n = 100$; Parma $n = 46$) and Central Italy (Ancona $n = 52$; Rome $n = 176$). DNA extraction, amplification and typing were performed independently by the four laboratories following the manufacturer's protocol. An R × C contingency table program, kindly provided by G. Carmody (Carleton University, Ottawa, Canada), was used to test homogeneity between the different population samples. The Hardy-Weinberg expectation was tested by the chi-square test between observed and expected genotype frequencies. Independence among the five tested loci was defined by the mean of the observed variance (S_k^2) of the number of the heterozygous loci (under the assumption of independence) and its 95% confidence intervals [6, 7]. The power of discrimination (PD) was calculated using Fischer's equation [8] and the chance of exclusion (CE) was calculated from allele frequencies [9].

Results and discussion

No deviations were found when the four Italian population samples were tested for homogeneity, so they were pooled together as a single Italian population sample (Table 1). The distribution of genotype frequencies in this Italian sample showed no deviations from Hardy-Weinberg equilibrium. The S_k^2 criterion showed independence among the five loci ($S_k^2 = 1.24$; 95% confidence intervals of variance 1.402–1.078). The combined PD was 0.999 and the combined CE was 0.702. Our data was in agreement with other Caucasian population studies (Table 2), except for the HBGG and D7S8 loci, where the p-values for the G-statistic were 0.025 and 0.032, respectively. The pairwise comparisons showed that non-homogeneity was due to the Dutch sample at the HBGG locus and to the Spanish sample at the D7S8 locus, with a large increase of the p-values (0.577 and 0.891, respectively) when the two samples were, alternatively, excluded from the test.

A. Tagliabracci (✉) · L. Buscemi · D. Rodriguez
Istituto di Medicina Legale dell'Università di Ancona,
Policlinico, I-60020 Torrette, Ancona, Italy

N. Cerri · R. Mignola · F. De Ferrari
Istituto di Medicina Legale dell'Università di Brescia,
Spedali Civili, I-25100 Brescia, Italy

T. M. Neri
Cattedra di Genetica Medica dell'Università di Parma,
Ospedale Maggiore, I-43100 Parma, Italy

N. Cucurachi · G. Masotti
Istituto di Medicina Legale dell'Università di Parma,
Ospedale Maggiore, I-43100 Parma, Italy

C. Vecchiotti · R. Lombardi · G. Umani Ronchi
Istituto di Medicina Legale dell'Università di Roma
„La Sapienza", Largo Regina Elena, I-00161 Roma, Italy

Table 1 Genotype and allele frequencies in an Italian population sample ($n = 374$)

Geno-types	LDLR	GYPA	HBGG	D7S8	Gc
AA	0.158	0.318	0.233	0.364	0.080
AB	0.513	0.457	0.503	0.476	0.091
BB	0.329	0.225	0.240	0.160	0.027
AC	–	–	0.016	–	0.329
BC	–	–	0.008	–	0.158
CC	–	–	0.000	–	0.315
	$\chi^2 = 1.245$ $0.2 < p < 0.3$ df = 1	$\chi^2 = 2.245$ $0.1 < p < 0.2$ df = 1	$\chi^2 = 1.438$ $0.2 < p < 0.3$ df = 1	$\chi^2 = 0.019$ $0.8 < p < 0.9$ df = 1	$\chi^2 = 0.675$ $0.8 < p < 0.9$ df = 3
Alleles					
A	0.414	0.547	0.492	0.602	0.290
B	0.586	0.453	0.496	0.398	0.151
C	–	–	0.012	–	0.559

Table 2 Homogeneity test between Italians ($n = 374$) (this study), American Caucasians ($n = 148$) [10], Swiss ($n = 100$) [11], Dutch ($n = 155$) [12], Spanish ($n = 207$) (Herrera M 1995 personal communication)

Systems	χ^2	P	G statistic	P
LDLR	3.6822	0.4410 ± 0.0157	3.6732	0.4420 ± 0.0157
GYPA	4.2681	0.3810 ± 0.0154	4.2801	0.3800 ± 0.0153
HBGG	13.9031	0.0680 ± 0.0080	17.0672	0.0250 ± 0.0049
D7S8	10.4687	0.0300 ± 0.0054	10.3836	0.0320 ± 0.0056
Gc	8.1619	0.4220 ± 0.0156	8.1345	0.4210 ± 0.0156

References

1. Yamamoto T, Davis CG, Brown MS, Schneider WJ, Casey ML, Goldstein JL, Russel DW (1984) The human LDL receptor: a cystein-rich protein with multiple alu sequences in its mRNA. Cell 39:27–38
2. Siebert PD, Fukuda M (1987) Molecular cloning of a human glycophorin B cDNA: nucleotide sequence and genomic relationship to glycophorin A. Proc Natl Acad Sci USA 84:6735–6739
3. Slightom JL, Blechl AE, Smithies O (1980) Human fetal Gγ- and Aγ- globin genes: complete nucleotide sequences suggest that DNA can be exchanged between these duplicated genes. Cell 21:627–638
4. Horn GT, Richards B, Merrill JJ, Klinger KW (1990) Characterization and rapid diagnostic analysis of DNA polymorphisms closely linked to the cystic fibrosis locus. Clin Chem 36:1614–1619
5. Yang F, Brune JL, Naylor SL, Cupples RL, Naberhaus KH (1985) Human group specific component (Gc) is a member of the albumin family. Proc Natl Acad Sci USA 82:7994–7998
6. Brown AHD, Feldman MW, Nevo E (1980) Multilocus structure of natural populations of Hordeum Spontaneum. Genetics 96:523–536
7. Chakraborty R (1984) Detection of nonrandom association of alleles from the distribution of the number of heterozygous loci in sample. Genetics 108:719–731
8. Fischer RA (1951) Standard calculations for evaluating a blood group system. Heredity 5:95–102
9. Garber RA, Morris JW (1983) General equations for the average power of exclusion for genetic system of n codominant alleles in one-parent and no-parent cases of disputed parentage. In: Walker RH (ed) Inclusion probabilities in parentage testing. AABB, Arlington Va, pp 277–280
10. Budowle B, Lindsey JA, DeCou JA, Koons BW, Giusti AM, Comey CT (1995) Validation and population studies of the loci LDLR, GYPA, HBGG, D7S8, and Gc (PM loci), and HLA-DQα using a multiplex amplification and typing procedure. J Forensic Sci 40:45–54
11. Hochmeister MN, Budowle B, Borer UV, Dirnhofer R (1994) Swiss population data on the loci HLA- DQα, LDLR, GYPA, HBGG, D7S8, Gc and D1S80. Forensic Sci Int 67:175–184
12. Kloosterman AD, Sjerps M, Wust D (1995) Dutch Caucasian population data on the loci LDLR, GYPA, HBGG, D7S8, and GC. Int J Legal Med 108:36–38