Int J Legal Med (1996) 108:280–282

SHORT COMMUNICATION

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Hungarian population data on the loci HLA-DQα, LDLR, GYPA, HBGG, D7S8 and GC

Received: 28 April 1995 / Received in revised form: 27 December 1995

Abstract Population data studies for HLA-DQα and PM loci (LDLR, GYPA, HBGG, D7S8, GC) were carried out on a Hungarian Caucasian population sample of 163 unrelated individuals. Whereas the observed PM allele frequencies were similar to those reported for Caucasians, significant differences were found for HLA-DQα between the Hungarian and some Caucasian population data. All six loci meet Hardy-Weinberg expectations and there is no evidence for association between any pairs of loci.

Key words HLA-DQα · LDLR · GYPA · HBGG · D7S8 · GC · Allele frequencies · Population genetics · Polymerase chain reaction (PCR)

Introduction

The HLA-DQα, LDLR, GYPA, HBGG, D7S8 and GC loci are extensively characterised and validated PCR-based systems for forensic use [1, 2]. The amplification and typing of the six sequence polymorphic loci can be performed using commercially available kits (AmpliType HLA-DQα Forensic DNA Amplification and Typing Kit, AmpliType PM PCR Amplification and Typing Kit, Perkin Elmer). This paper presents the allele and genotype frequencies for the HLA-DQα, LDLR, GYPA, HBGG, D7S8 and GC loci in a Hungarian population sample.

Materials and methods

DNA for PCR analysis was extracted from whole liquid blood samples from 163 unrelated individuals by a previously described method [3]. The amplification and typing conditions used were those recommended by the manufacturer (Perkin Elmer).

Possible divergence from Hardy-Weinberg expectations (HWE) was determined by calculating the unbiased estimate of the expected homozygote/heterozygote frequencies [4–6], the likelihood ratio test [7–9] and the exact test [10]. An inter-class correlation criterion [11] was used for detecting disequilibrium between loci pairs. Independence among the six PCR-based loci was determined by examining whether or not the observed variance of the number of heterozygous loci in the population sample is within it's confidence interval under the assumption of independence [12–13]. Population homogeneity was tested using a computer program (RXC contingency table) kindly provided by G. Carmody (Carleton University, Canada).

Results and discussion

The distributions of observed genotypes and allele frequencies for HLA-DQα and PM loci are shown in Tables 1 and 2, respectively. There was no detectable deviation from HWE for these loci based on the homozygosity test,

Table 1 Observed genotype and allele frequency distributions for HLA-DQα in 163 unrelated Hungarian Caucasians

Genotype	Frequency	Genotype	Frequency
1.1–1.1	0.037	1.3–1.3	–
1.1–1.2	0.074	1.3–2	0.043
1.1–1.3	0.055	1.3–3	0.025
1.1–2	0.037	1.3–4	0.055
1.1–3	0.037	2–2	0.012
1.1–4	0.098	2–3	0.006
1.2–1.2	0.037	2–4	0.037
1.2–1.3	0.037	3–3	0.006
1.2–2	0.031	3–4	0.104
1.2–3	0.043	4–4	0.110
1.2–4	0.117		
Allele	**Frequency**	**Allele**	**Frequency**
1.1	0.187	2	0.089
1.2	0.187	3	0.114
1.3	0.107	4	0.316

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Table 2 Observed genotype and allele frequency distributions for PM loci in 163 unrelated Hungarian Caucasians

Genotype	Frequency	Genotype	Frequency
LDLR AA	0.166	D7S8 AA	0.350
LDLR AB	0.515	D7S8 AB	0.509
LDLR BB	0.319	D7S8 BB	0.141
GYPA AA	0.362	GC AA	0.061
GYPA AB	0.460	GC AB	0.055
GYPA BB	0.178	GC BB	0.018
HBGG AA	0.239	GC AC	0.337
HBGG AB	0.521	GC BC	0.166
HBGG BB	0.233	GC CC	0.362
HBGG AC	0.006		
HBGG BC	–		
HBGG CC	–		
Allele	**Frequency**	**Allele**	**Frequency**
LDLR A	0.423	D7S8 A	0.604
LDLR B	0.577	D7S8 B	0.396
GYPA A	0.592	GC A	0.258
GYPA B	0.408	GC B	0.129
HBGG A	0.503	GC C	0.614
HBGG B	0.494		
HBGG C	0.003		

Table 3 Tests for independence and discrimination powers for HLA-DQα and PM loci for 163 unrelated Hungarian Caucasians

	HLA-DQα	LDLR	GYPA	HBGG	D7S8	GC
Observed homozygosity	20.3%	48.5%	54.0%	47.2%	49.1%	44.2%
Expected homozygosity[a]	20.0%	51.0%	51.6%	49.5%	52.0%	45.8%
Homozygosity test[b]	0.930	0.513	0.533	0.556	0.451	0.682
Likelihood Ratio test[b]	0.446	0.540	0.617	0.512	0.404	0.876
Exact test[b]	0.522	0.540	0.513	0.566	0.496	0.867
Power of discrimination	0.93	0.61	0.63	0.62	0.60	0.72

[a] Expected homozygosity is an unbiased estimate
[b] These values are probability values. Test considered significant if $P < 0.05$

Table 4 Two locus inter-class correlation test for HLA-DQα and PM loci for 163 unrelated Hungarian Caucasians

	LDLR	GYPA	HBGG	D7S8	GC
GYPA	0.668				
HBGG	0.442	0.833			
D7S8	0.591	0.671	0.380		
GC	0.287	0.098	0.763	0.749	
HLA-DQα	0.557	0.737	0.092	0.819	0.194

Values are two-sided probability values. Test considered significant if $P < 0.05$

likelihood ratio test and the exact test (Table 3). An inter-class correlation test analysis demonstrated that there is no evidence for correlation between the alleles at any of the pairs of loci (Table 4). Furthermore, there is no evidence of association for the six loci described in our Hungarian sample population using the s_k^2 criterion (s_k^2 = 1.567; 95% confidence interval of variance 1.125–1.686). Pairwise testing for population homogeneity revealed no significant differences for all the six loci between the Hungarians and other Caucasians (Swiss [14], Dutch [15, 16], Germans [17, 18]). However, the Hungarian HLA-DQα allele frequencies differed significantly ($P < 0.05$) from some Caucasian population data (US Caucasian [2], UK Caucasian [19], Italian [20]). The combined power of discrimination (PD) value for the six loci in the Hungarian population is 0.9996.

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BOOK REVIEW

Ferner RE (1996) Forensic Pharmacology. Oxford University Press, Oxford, 249 pages. ISBN 0-19-854826-5

This book written by a non-forensic scientist constitutes an original but useful contribution in the vast domain of the legal medicine. While homicide by poisoning has become for several decades a subject of rarity due to the improvement of analytical toxicology, chemicals in general and medicines in particular are more and more involved as interfering or determinant factors in forensic cases and other related matters. In this respect, Doctor Ferner's book is an essential guide through the data of this new common branch of law and pharmacology defined as "the point at which medicines, mayhem and malpractice meet".

The text is divided into 3 parts including 15 chapters to which 3 appendices are added. Part 1 refers firstly to general considerations on drugs, pharmacokinetics, analytical determination and interpretation, and secondly, to legal considerations about admissibility of the proof and the role of the expert. Part 2 is devoted to specific problems concerning the effects of drugs on human behaviour and especially on the victims of crime and different aspects of the so up-to-date matter "negligence and medicines". Part 3 deals with the major drugs of concern in forensic practice such as ethanol, benzodiazepines, insulin, potassium salts, neuroleptics, opiates, paracetamol, anabolic steroids and vaccines. More technical aspects of pharmacology are developed in the appendices. There are related to pharmacokinetic calculations, detailed aspect of measurement of ethanol and calculations based on ethanol concentration as well as brief details on drugs of forensic importance.

Due to the importance of the subjects, the clarity of the text illustrated by numerous examples from the literature and the author's experience, the book will without any doubt meet the expectations of a wide range of readers more or less involved in this complex matter such as general and clinical practitioners, forensic pathologists and toxicologists, pharmacologists, students of clinical medicine, lawyers and police officers. An extensive bibliography and an index are included, which make this book a very helpful guide in the field of pharmacology applied to forensic medicine.

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