BRIEF COMMUNICATION

Richard Zehner,[1] *Ph.D.; Dietrich Mebs,*[1] *Ph.D.; and Hansjürgen Bratzke,*[1] *Ph.D.*

Population Genetic Study of the Simultaneously Amplified Loci HLA DQA1, LDLR, GYPA, HBGG, D7S8, and GC in a German Population Sample

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ABSTRACT: A population genetic study of the HLA DQA1 and the ''Polymarker'' loci LDLR, GYPA, HBG, D7S8, and GC has been performed in a German Caucasian population (Frankfurt am Main area).

All loci are in Hardy-Weinberg equilibrium and no unexpected association of loci has been observed. The data of the allele distributions are similar to those of other Caucasian populations. All six loci together have a power of discrimination (PD) of 0.9996 and an exclusion chance in paternity testing of 0.81.

KEYWORDS: forensic science, DNA typing population genetics, polymerase chain reaction polymarker, HLA DQA1, LDLR, GYPA, HBGG, D7S8, GC, Germany, Frankfurt am Main.

The commercially available ''Polymarker'' Typing Kit (Amplitype™ PM PCR Amplification and Typing Kit, Perkin Elmer, Norwalk, CT) is a well-characterized and validated PCR-Typing Kit (1). Simultaneous amplification of the polymorphic loci HLA DQA1, LDLR, GYPA, HBGG, the locus D7S8, and GC is possible, leading to a high combined power of discrimination (PD). This paper presents population data of a German population sample from the area of Frankfurt am Main.

Materials and Methods

Sample Preparation

Blood samples from 100 Caucasians originating in the Frankfurt am Main area (Germany) were used. Genomic DNA was isolated using standard proteinase K-digestion, phenol/chloroform extraction, and ethanol precipitation (2).

[1] Zentrum der Rechtmedizin der J. W. Goethe-Universitat, Frankfurt, Germany.

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Amplification and Typing

Amplification and typing was performed using the Amplitype PM PCR Amplification and Typing Kit (Perkin Elmer, Norwalk, CT) according to the manufacturer's recommendations. HLA DQA1 was typed with an aliquot of the generated PCR products on separate strips (HLA DQA1 Amplification and Typing Kit, Perkin Elmer).

Statistical Analysis

The typing data were analyzed with the exact test to establish Hardy-Weinberg equilibrium (3) and independence of the loci (4). Furthermore, the heterozygosity rate, the power of discrimination (PD) and the average exclusion rate in paternity cases was examined. All statistical analyses were performed using the software included in the computer assisted fragment evaluation system ''DNA View'' (Ch. Brenner, Berkeley, CA.)

Results

The allele frequencies in the present study are shown in comparison with other population studies in Table 1. The results of statistical analysis are shown in Table 2.

Independence testing of loci gave a value of $P > 0.05$ when comparing the typed loci with each other, except for the comparison of GC and HLA DQA1 where a P value of 0.04 was calculated.

For all six loci, a combined power of discrimination of 0.9996 and a combined exclusion chance in paternity cases of 0.81 were calculated.

Discussion

In the present study the distribution of the genotypes was found to be in accordance with the expected values ($P_{\text{exact-test}} > 0.05$). The independence test between the loci revealed P values > 0.05 each, except when the loci GC and HLA DQA1 are compared, where the value of P is 0.04. However, this observation does not necessarily mean non-independence, because in 15 statistical tests this observation is not completely unexpected.

The present data revealed no remarkable differences in the allele

TABLE 1—*Comparison of the obtained allele–frequencies of each locus with other databases.*

Locus (Alleles)	German (Frankfurt area) $N = 100$	German (5,6)	Swiss (7)	US Caucasians (1)
LDLR				
A	0.40	0.45	0.44	0.45
B	0.60	0.55	0.56	0.55
GYPA				
A	0.56	0.53	0.52	0.58
B	0.44	0.47	0.48	0.42
HBGγ				
A	0.54	0.52	0.48	0.47
B	0.455	0.47	0.52	0.52
C	0.005	0.006	0	0.007
D7S8				
A	0.67	0.59	0.58	0.59
B	0.33	0.41	0.42	0.41
GC				
A	0.26	0.32	0.28	0.26
B	0.15	0.11	0.18	0.17
C	0.59	0.57	0.54	0.57
HLA DQα				
1.1	0.12	0.16	0.15	0.12
1.2	0.19	0.21	0.19	0.18
1.3	0.08	0.09	0.10	0.04
2	0.16	0.10	0.15	0.12
3	0.15	0.14	0.15	0.22
4	0.29	0.31	0.27	0.33

frequencies investigated when compared with data from other geographical areas (1,5,6,7), suggesting genetic homogenity of the Caucasian race.

Since a combined power of discrimination of 0.9996 and an average exclusion chance in paternity cases of 0.81 was calculated, the study confirms that the multiplex system is a powerful tool in forensic examination. Data can be obtained that are important in calculating the probability of a random match between a stain and a suspect such as in forensic casework or in calculating a PI in paternity cases.

TABLE 2—*Results of the statistical tests for Hardy–Weinberg equilibrium (HWE), heterozygosity, power of discrimination (PD) and exclusion-chance in paternity cases (PE) for the investigated loci in the Frankfurt population sample.*

	LDLR	GYPA	HBGG	D7S8	GC	HLA DQA1
HWE: $p =$	0.08	0.28	0.80	0.32	0.83	0.96
Heterozygosity	0.48	0.50	0.50	0.45	0.56	0.81
PD	0.61	0.62	0.63	0.59	0.74	0.94
PE	0.18	0.19	0.19	0.17	0.30	0.62

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Additional information and reprint requests:
Richard Zehner
Zentrum der Rechtsmedizin der J.W. Goethe–Universität
Kennedyallee 104
60596 Frankfurt
Germany