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SHORT COMMUNICATION

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Population studies of three AMPFLPs systems in a North Polish population

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Abstract Allele and phenotype frequencies for D1S80, D17S5 and ApoB were determined in a population sample of more than 200 unrelated persons from North Poland using the PCR method. For D1S80, D17S5 and ApoB 19, 13 and 21 alleles respectively were observed. No deviations from Hardy-Weinberg equilibrium were detected. All three systems have discrimination values above 92% and a cumulative discrimination index of 4.5×10^{-4}.

Key words VNTR · D1S80 · D17S5 · ApoB · Population genetics · North Poland

Introduction

Variable number of tandem repeat (VNTR) loci are important genetic markers used for precise gene mapping, construction of linkage maps or paternity and identification analyses in forensic haemogenetics. VNTR loci with a repeat core length between 10 and 100 bp are classified as minisatellites. The most commonly amplified minisatellite loci (AMPFLPs) are D1S80, ApoB, D17S5 and COL2A1 (Rand et al. 1992). Although microsatellite short tandem repeat (STR) loci offer some advantages over AMPFLPs systems, the latter can still be used as forensic or clinical markers. We describe here the results of three AMPFLPs systems investigated in a population sample from North Poland (Gdańsk area).

The aims of this investigation were to obtain allele frequencies for a Polish population, to test whether the allele frequencies conform to Hardy-Weinberg expectations and to calculate some parameters characterizing forensic and genetic usefulness.

Dedicated to Prof. Dr. J. Gerchow on the occasion of his 75th anniversary

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Materials and methods

Blood samples were taken from unrelated persons of both sexes living in North Poland (Gdańsk area) and DNA was isolated using a non-organic and non-enzymatic method (Lahiri et al. 1992). The VNTR systems analysed and PCR conditions are shown in Table 1. Electrophoretic separation of PCR products was performed by high resolution polyacrylamide gel electrophoresis according to Allen et al. (1989). Gels were silver stained using the modified method of Bassam et al. (1991). Allelic ladders for ApoB (alleles 29–41 and 45–55) and D17S5 (alleles 1–13) were kindly supplied by Prof. Brinkmann. The D1S80 ladder was purchased from Perkin Elmer.

Results and discussion

The distribution of allele frequencies for the systems investigated are shown in Tables 2–4. In our population sample we observed 19 alleles and 59 genotypes for D1S80, 13 alleles and 53 genotypes for D17S5 and 21 alleles and 62 genotypes for Apo B. In the Apo B system we observed two different alleles with a higher electrophoretical mobility than allele 29 (the number of repeats was estimated to be between 27 and 25) and some variant alleles (Table 4). In the D1S80 system we observed very few interalleles which were counted with the nearest allele. We did not observe any interalleles for the D17S5 locus. The AMPFLP allele frequencies found in

Table 1 VNTR loci studied

Locus	Chromosome location	Chromosomes tested (*n*)	Primer sequences	Amplification conditions
D1S80	1p36–p35	414	Kasai et al. (1990)	Kloosterman et al. (1993)
D17S5	17p13.3	408	Horn et al. (1989)	Rand et al.(1992)
ApoB	2p24	402	Boerwinkle et al. (1989)	Rand et al. (1992)

Table 2 Allele frequencies of D1S80 in the Polish population ($n = 207$)

Allele	Frequency	Allele	Freq-quency	Allele	Frequency	Allele	Frequency
18	0.2005	23	0.0386	28	0.0507	33	0.0024
19	0.0024	24	0.3623	29	0.0459	36	0.0072
20	0.0266	25	0.0628	30	0.0121	37	0.0121
21	0.0072	26	0.0290	31	0.0604	41	0.0024
22	0.0459	27	0.0169	32	0.0145		

Table 3 Allele frequencies of D17S5 in the Polish population ($n = 204$)

Allele	Frequency	Allele	Freq-quency	Allele	Frequency	Allele	Frequency
1	0.0515	5	0.0613	9	0.0539	13	0.0074
2	0.1324	6	0.0490	10	0.0759		
3	0.1961	7	0.0171	11	0.0098		
4	0.2672	8	0.0490	12	0.0294		

Table 4 Allele frequencies of Apo B in the Polish population ($n = 201$)

Allele	Frequency	Allele	Frequency	Allele	Frequency
< 29 a	0.0050	33	0.0348	43	0.0050
< 29 c	0.0025	35	0.1865	45	0.0249
29	0.0373	37	0.3980	47	0.0547
29 a*	0.0050	37 a	0.0124	49	0.0746
29 c	0.0149	37 c	0.0423	49 c	0.0050
31	0.0224	39	0.0224	51	0.0100
31 a	0.0100	41	0.0299	53 a	0.0025

Table 5 χ^2-test for Hardy-Weinberg calculations (allele binning method according to Rand et al. 1992)

Groups of allele	D1S80	D17S5	ApoB
I	18	1– 2	53–47
II	19–23	3	45–37 a
III	24	4	37
IV	25–28	5– 8	35
V	29–41	9–13	33–< 29 a
χ^2	17.18	15.75	13.99
P (df = 10)	0.1–0.05	0.2–0.1	0.2–0.1

the Polish population are similar to other European population data (Boerwinkle et al. 1989; Rand et al. 1992; Deka et al 1992, 1994; Woller et al. 1995).

The χ^2 test was carried out to estimate if the population sample conforms to Hardy-Weinberg equilibrium (Table 5). The Polish population meets H-W expectations for D1S80, D17S5, and ApoB systems. Table 6 shows some statistical parameters of forensic interest.

The observed frequency of heterozygotes in D17S5 (0.804) is significantly lower compared to the expected heterozygosity ± standard error for H (0.853 ± 0.025). One of the possible explanations for homozygote excess could be the phenomenon of preferential amplification of smaller alleles as observed in AMPLFP systems.

The cumulative discrimination index (Wong et al. 1987) for the three systems was calculated as 4.5×10^{-4}.

Table 6 Forensic values for the three systems ihn a Polish population

System	H[a] obs.	H exp ± SE	PD[b]	DI[c]	PIC[d]	pM[e]
D1S80	0.802	0.812 ± 0.027	0.935	0.071	0.793	0.065
D17S5	0.804	0.853 ± 0.025	0.950	0.069	0.836	0.050
ApoB	0.771	0.793 ± 0.029	0.927	0.093	0.773	0.073

[a] H – heterozygosity (Kloosterman et al. 1993)
[b] power of discrimination PD = $1\text{-}\Sigma(P_i)^2$, where P_i represent the frequency of each genotype
[c] discrimination index (Wong et al. 1987)
[d] polymorphic information content (Botstein et al. 1980)
[e] matching probability (Jones 1972)

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