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Frequencies for five short tandem repeat (STR) systems in a population from North Poland

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Abstract A population study of unrelated individuals from North Poland (Gdansk area) was carried out to investigate the allele distributions of the five STR systems HUMCD4, HUMFES/FPS, HUMVWA31, HUMTH01 and ACTBP2. PCR products were separated on horizontal non-denaturing polyacrylamide gels followed by silver staining. For all STR systems analysed the distribution of observed phenotypes did not deviate from Hardy-Weinberg equlibrium. A comparison of allele distributions between Polish and other European Caucasian population samples is presented.

Key words Population genetics · Poland · Short tandem repeats · ACTBP2 · HUMFES/FPS · HUMCD4 · HUMVWA31 · HUMTH01 · PCR

Introduction

PCR amplification of minisatellite (AMPFLP) and microsatellite (STR) VNTR systems is widely used in forensic DNA analysis [1]. STR systems with relatively small fragment lengths, allele sizes differing by 3–7 bp and relative stability (resistance) to DNA degradation, are often used as markers for individualization of biological traces [2].

The present study investigates allele frequency distributions for five STR systems in the population of Northern Poland and a comparison of allele frequency distributions between Polish and other European Caucasian populations.

Materials and methods

Blood samples from 203 healthy, unrelated donors living in the Gdańsk area (North Poland). DNA was extracted using the method described by Lahiri et al. [3]. PCR amplifications was performed according to Wiegand et al. [4] (ACTBP2 and HUMTH01), Möller et al. [5] (HUMFES/FPS) and Wall et al. [6] (HUMCD4). Electrophoresis was carried out as described by Allen et al. [7]. Gels were stained with silver [8] and alleles identified by comparison with allelic ladders kindly supplied by Prof. B. Brinkmann (Institute of Forensic Medicine Münster, Germany).

The χ^2-test was used to calculate deviations from the Hardy-Weinberg equilibrium (HWE). To demonstrate homogeneity of the allele frequency distributions between different populations, RxC contingency tables were used (computer program kindly provided by G. Carmody, Carleton University, Ottawa, Canada).

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Results and discussion

The allele distributions for the five STR systems in the North Polish population sample are given in Table 1.

HUMTH01

For HUMTH01 system we found 7 different alleles giving 19 phenotypes. No deviations from HWE were found ($\chi^2 = 11.22$; $0.5 < P < 0.7$; d.f. = 12).

We have shown that there is no significant difference in the allele frequencies at the HUMTH01 locus between the Polish and the German [4], Swiss [9], Spanish [10] and Hungarian [11] populations (Table 2). Deviation from population homogeneity was however, observed when comparing the Polish population sample with French [12], Danish [13], Dutch [14] and Austrian populations [15]. The greatest discrepancies between the Polish and Austrian populations were observed for alleles 7 ($\chi^2 = 13.4500$; $P = 0.0000$) and 9 ($\chi^2 = 9.3303$; $P = 0.0020$) and between the Polish and French populations for allele 10 ($\chi^2 = 6.3400$; $P = 0.0150$).

HUMVWA31

For HUMVWA31 we observed 8 alleles and 27 phenotypes in the population sample of 185. Alleles 11, 12 and

Table 1 Allele frequencies for five STR systems in the Polish population

Alleles	ACTBP2 $n = 176$	HUMTH01 $n = 203$	HUMVWA31 $n = 185$	HUMFES $n = 106$	HUMCD4 $n = 129$
1					
2					
3					
4					
5		0.0025			0.3256
6		0.2488			0.3101
7		0.1256			0.0116
8		0.1207		0.0189	–
9		0.1847		0.0094	–
9.3		0.3054		–	–
10		0.0123		0.0708	0.3062
10a				0.1934	–
11				0.4057	0.0194
11a				0.0142	–
12	0.0170			0.2217	0.0194
13	0.0057		0.0054	0.0660	0.0078
14	0.0426		0.0648		
15	0.0540		0.0972		
16	0.0625		0.1945		
17	0.0739		0.2864		
18	0.0540		0.2324		
19	0.0852		0.0945		
20	0.0739		0.0243		
21	0.0369				
22	0.0483				
23	0.0227				
24	0.0341				
25	0.0227				
26	0.0370				
27	0.0653				
28	0.0653				
29	0.0454				
30	0.0682				
31	0.0426				
32	0.0227				
33	0.0085				
34	0.0057				
35	0.0028				
36	0.0028				
37					
39					

21 were not observed. The most frequent alleles were 16, 17 and 18 (Table 1), and the most frequent phenotypes 16,17 (12.97%) and 17,18 (11.89%). The population sample analysed meets HWE expectations ($\chi^2 = 15.8760$; $0.3 < P < 0.5$; d.f. = 14). Our allele frequency distribution is similar to those observed for Dutch [14], German [5], English [16, 17], Austrian [15] and Hungarian [11] populations. Statistically significant differences were observed between the Polish and Finnish [18] and the Polish and Spanish [19] populations.The greatest difference between the Polish and Spanish populations was found for allele 14 ($P < 0.001$).

HUMFES/FPS

In the population sample of 106 persons we observed 8 alleles and 20 phenotypes. Alleles 7 and 14 were not observed. The most frequent alleles were 11 and 10a and the most frequently occurring phenotypes were 11, 12 (20.75%) and 10a, 11 (17.92%). Statistical calculations showed that population sample meets HWE expectations with a high P-value ($\chi^2 = 0.5317$; $0.95 < P < 0.98$; d.f. = 4). Our allele frequency distribution is similar to those observed for German (Prof. B. Brinkmann - personal communication), French [12], Spanish [10, 20], Dutch [14] and Austrian [21] population samples (Table 2).

Table 2 Pairwise comparison of allele frequencies for five STRs between Caucasian population samples

Compared populations	χ^2	P	G-statistic	P
HUMCD4				
PL × D [PBB]	13.108	0.092	13.851	0.113
PL × A [22]	9.830	0.252	12.209	0.208
HUMFES/FPS				
PL × A [21]	8.380	0.285	8.864	0.318
PL × F [12]	7.583	0.268	7.923	0.306
PL × E (N) [20]	3.939	0.584	4.606	0.544
PL × E (NW) [10]	3.609	0.612	3.615	0.622
PL × D [PBB]	8.074	0.331	9.242	0.338
PL × NL [14]	8.315	0.121	7.984	0.172
HUMTH01				
PL × D [4]	7.034	0.314	7.452	0.317
PL × H [11]	4.245	0.640	4.253	0.662
PL × CH [9]	5.954	0.438	5.723	0.512
PL × E [10]	10.642	0.079	12.582	0.056
PL × F [12]	12.834	0.038	14.802	0.027
PL × NL [14]	11.583	0.033	11.691	0.044
PL × DK [13]	21.694	0.002	22.626	0.003
PL × A [15]	22.345	0.000	22.998	0.000
HUMVWA31				
PL × NL [14]	6.470	0.590	8.018	0.510
PL × D [5]	8.138	0.418	8.718	0.413
PL × GB [17]	7.574	0.362	8.242	0.349
PL × GB [16]	10.440	0.147	10.682	0.166
PL × A [15]	13.423	0.094	14.194	0.108
PL × H [11]	13.964	0.063	15.265	0.057
PL × SF [18]	18.984	0.006	19.552	0.011
PL × E [19]	26.619	0.000	30.171	0.000
ACTBP2				
PL(N) × PL(W) [DJJ]	27.101	0.298	27.279	0.371
PL × D [PBB]	39.922	0.038	39.677	0.043
PL × CRO [23]	19.540	0.018	19.090	0.021

PL – Poland, D – Germany, A – Austria, F – France, E – Spain, (N) – north, (NW) – north-west, NL – Holland, H – Hungary, DK – Denmark, GB – Great Britain, (W) – west, CRO – Croatia, PBB – Prof. B. Brinkmann, personal communication, DJJ – Dr. J. Jaroszewski, personal communication

HUMCD4

The distribution of HUMCD4 alleles was analysed in a sample of 129 individuals. We observed alleles 5, 6, 7, 10, 11, 12 and 13 and we found 15 out of 28 possible phenotypes. In the Polish population the most frequent alleles were 5, 6 and 10 and the most frequent phenotypes were 5, 6 (19.38%), 6, 10 (17.83%), and 5, 5 and 5, 10 with a frequency of 14.73%. The population sample was in HW equilibrium ($\chi^2 = 6.8315$; $0.2 < P < 0.3$; d.f. = 5). A comparison of our data with German (Prof. B. Brinkmann - personal communication) and Austrian [22] populations showed no deviation from population homogeneity (Table 2).

ACTBP2

ACTBP2 is one of the most informative STR systems introduced so far in our laboratory. A total of 25 alleles was observed in a population sample of 176 unrelated persons. The observed frequency of the most frequent allele was not higher than 9% (Table 1) and the frequency of most frequent phenotype below 4%. The statistical analysis of the population sample clearly showed that ACTBP2 meets HWE expectations ($\chi^2 = 2.45$; $0.95 < P < 0.98$; d.f. = 8). Possible heterogeneity of ACTBP2 allele frequencies between the North Polish, German (Prof. B. Brinkmann – personal communication) and Zagreb [23] population samples was assessed using an RxC contingency table. Deviation from population homogeneity was observed. The north Polish sample, however was not statistically different from a population of west Poland (Dr. J. Jaroszewski – personal communication) (Table 2).

Table 3 shows the forensic efficiency values of the systems expressed as various statistical parameters. The results of the statistical analysis were satisfactory and highly informative suggesting that the five such STR systems are powerful tools for forensic purposes. Some systems such as ACTBP2 and HUMTH01 are routinely used in our laboratory and in our opinion are very suitable markers for routine stain investigations. Preliminary tests performed on the other systems also demonstrate their potential efficency in stain analysis.

Table 3 Forensic value of the five STR systems analysed in the Polish population

System	PD[a]	H[b]	DI[c]	PIC[d]	Most frequent phenotype	(%)
ACTBP2	0.995	0.955	0.004	0.942	17,30	(3,40)
HUMVWA31	0.932	0.789	0.080	0.775	16,17	(12.97)
HUMTH01	0.916	0.778	0.087	0.746	6,9.3	(12.81)
HUMFES/FPS	0.883	0.736	0.121	0.702	11,12	(20.75)
HUMCD4	0.862	0.636	0.217	0.644	6,10	(17.83)

[a] PD indicates power of discrimination which is calculated using the formula $PD = 1-\Sigma(Pi)^2$, were Pi is the frequency of each genotype. [b] observed heterozygosity; [c] DI – discrimination index [24]; [d] PIC – indicates polymorphic information content [25]

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