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SHORT COMMUNICATION

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A study of the short tandem repeat systems HUMVWA and HUMTH01 in an Austrian population sample

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Abstract The genotype distributions for the short tandem repeat systems (STRs) HUMVWA and HUMTH01 have been studied in 128 unrelated Caucasians fromêAustria. The allelic distributions were in accordance with Hardy-Weinberg expectations. The heterozygosities were 0.82 and 0.81, whereas the mean exclusion chance was 0.62 and 0.55, respectively. In one person, a VWA-allele consisting of 11 repeats was found.

Key words PCR · HUMVWA · HUMTH01 · Short tandem repeat · Population study

Introduction

In recent years, short tandem repeat systems (STRs) have gained importance in forensic analysis of biological specimens as well as in paternity testing [1]. Among the most often used are HUMVWA [2] and HUMTH01 [3], both tetrameric repeat systems. This study was performed to obtain allele frequencies in an Austrian population sample.

Materials and methods

Blood from 128 unrelated Austrian caucasians was extracted using the SuperQuickGene kit (AGTC, Denver, Colo). PCR and electrophoresis was performed according to [4] for HUMTH01 and [5] for HUMVWA. Ladders were purchased from Serac (Bad Homburg, FRG). Sequencing of the small HUMVWA-allele found in this study was performed as previously described [6].

Statistics

The mean exclusion chance (ME, AVACH) was calculated according to Krüger et al. [7], The discriminating power was calculated as 1–Σ (expected phenotype frequencies)^2 [8]. For checking the Hardy-Weinberg expectations, chi^2-tests were performed. Also the comparisons of the allele frequencies between different populations were done by chi^2 tests of R*C contingency tables. For the chi^2 tests alleles with less than 5% were pooled.

Results and discussion

The allelic frequencies of both STRs for Austrians (this study), Germans [4, 5] and US-Blacks [9, 10], are presented in Table 1.

For HUMVWA 24 genotypes were found corresponding to 9 alleles (Table 2). The heterozygosity rate was 0.82, the mean exclusion chance (ME) was 0.62 and the discriminating power (DP) was 0.93. No deviation from Hardy-Weinberg expectations was found (chi^2: 13.52, df=19, p=0.8–0.9).

Table 1 Allele frequencies for HUMTH01 and HUMVWA in Austrians, Germans [4, 5] and US-Blacks [9, 10]

HumTh01	Austrians (*n* = 256)	Germans (*n* = 220)	US Blacks (*n* = 740)
6	0.207	0.207	0.135
7	0.219	0.180	0.370
8	0.129	0.126	0.211
9	0.113	0.171	0.146
9,3	0.32	0.302	0.138
10	0.012	0.014	–
HumVWA	(*n* = 250)	(*n* = 642)	(*n* = 202)
11	0.004	–	–
13	0.004	0.005	0.005
14	0.102	0.110	0.069
15	0.122	0.098	0.223
16	0.205	0.210	0.243
17	0.283	0.270	0.203
18	0.213	0.210	0.158
19	0.055	0.077	0.084
20	0.012	0.017	0.010
21	–	0.002	0.005

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Table 2 Phenotypes for HUMTH01 and HUMVWA and their frequencies in an Austrian population sample

HUMVWA			HUMTH01		
Allele	Observed	Frequency	Allele	Observed	Frequency
11//15	1	0.009	6//6	3	0.025
13//16	1	0.009	6//7	17	0.143
14//14	2	0.018	6//8	11	0.092
14//15	6	0.053	6//9	4	0.034
14//16	3	0.027	6//9.3	16	0.134
14//17	5	0.044	7//7	5	0.042
14//18	4	0.035	7//8	7	0.058
14//19	3	0.027	7//9	3	0.025
14//20	1	0.009	7//9.3	19	0.160
15//15	2	0.018	8//8	1	0.008
15//16	4	0.035	8//9	4	0.034
15//17	11	0.097	8//9.3	11	0.092
15//18	5	0.044	9//9	4	0.034
15//19	2	0.018	9//9.3	9	0.076
16//16	6	0.053	9//10	2	0.017
16//17	16	0.142	9.3//9.3	11	0.092
16//18	12	0.106	9.3//10	1	0.008
16//19	5	0.044	Total	128	
17//17	8	0.071			
17//18	16	0.142			
17//19	4	0.035			
17//20	2	0.018			
18//18	4	0.035			
18/19	2	0.018			
Total	125				

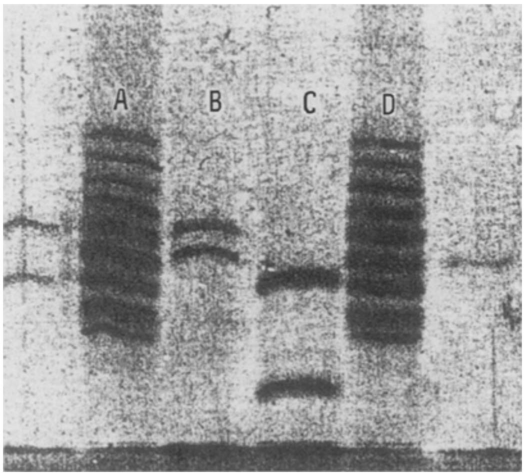

Fig. 1 Pherogramm of the small HUMVWA-allele. Lane A and D: Allelic ladders (including alleles 13 through 21). Lane B: Alleles 16 and 17. Lane C: alleles 11 and 15

Testing for population homogeneity revealed no differences between Germans and Austrians (chi^2: 2.3, df: 5, p = 0.8–0.9), but significant differences between US-Blacks and Austrians (chi^2: 16.94, df: 5, p = 0.001–0.005).

In one person an allele was found that migrated distinctly faster than the allele 13 (Fig. 1). Sequencing of this allele proved that it had a length of 126 bp and consisted of 11 repeats and regular flanking regions. The repeat structure was TCTA $(TCTG)_3(TCTA)_7$. Alleles with less than 13 repeats are uncommon in caucasians and have been found in non-caucasians (B. Brinkmann, pers. communication) and non-human primates only [11].

For HUMTH01 a total of 17 genotypes were found corresponding to 6 alleles (Table 2). The heterozygosity rate was 0.81, ME was 0.55 and DP 0.91. No deviation from Hardy-Weinberg expectations was found (chi^2: 15.55, df: 14, p = 0.4–0.3).

Testing for population homogeneity revealed no differences between Germans and Austrians (chi^2: 3.85, df: 4, p = 0.4–0.5), but significant differences between US-Blacks and Austrians (chi^2: 49.14, df: 4, $p < 0.0005$).

No mutations were found in 14 paternity cases confirmed by classical systems.

In conclusion both systems proved to be of value for paternity testing (combined ME: 0.83) in an Austrian population sample.

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