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SHORT COMMUNICATION

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Short tandem repeat HumACTBP2 (SE33) and HumVWA: population genetic study on a north Italian population

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Abstract Allele frequencies at the short tandem repeat (STR) loci HumACTBP2 and HumVWA were determined in 118 unrelated individuals from Northern Italy (Milan area). For locus HumACTBP2 (SE33) a total of 39 alleles was observed. Furthermore, two interalleles (N18m + N19m) and one allele (> N35) were found which were not observed in a wider German population survey (n = 560). For the STR system HumVWA, 7 alleles could be detected. Both systems showed no significant deviation from Hardy-Weinberg equilibrium. A comparison of Italian and German population data revealed no significant differences for locus HumVWA, while significant differences were observed for locus HumACTBP2.

Key words Short tandem repeats · HumACTBP2 · HumVWA · Population studies · Northern Italy

Introduction

HumACTBP2 (Polymeropoulos et al. 1992) is known to be one of the most polymorphic STR systems so far validated for forensic purposes (Wiegand et al. 1993a) not only because of its high length polymorphism but also for its highly variable sequence polymorphism (Urquhart et al. 1993; Möller and Brinkmann 1994).

The allele frequency distributions for HumACTBP2 and a further STR system HumVWA (Kimpton et al. 1992) were investigated on a population sample from Northern Italy (Milan area) in order to collect population data suitable for forensic casework and population genetic studies.

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Materials and methods

DNA from 118 unrelated Caucasian individuals, residing in the Milan area was extracted from air dried blood on sterile cotton fabric as previously described (Wiegand et al. 1993b). The polymerase chain reaction assay and electrophoresis were carried out according to Möller et al. 1994 (VWA) and Wiegand et al. 1993a (ACTBP2), respectively. Bands were visualised by silver staining (Budowle et al. 1991).

Allele designation of HumACTBP2 alleles was performed by side-to-side comparison with a sequenced allelic ladder composed of 26 sequenced alleles (Möller et al. 1995) and for HumVWA alleles using a sequenced ladder described by Möller et al. (1994).

Hardy-Weinberg analysis was performed using the HWE-Analysis-Software, Version 3.0, provided by C. Puers (C. Puers, Münster, Germany). The comparison of observed and expected numbers of heterozygotes (gene diversity) was calculated according to Nei (1978), the mean exclusion chance according to Krüger et al. (1968), the mean paternity exclusion probability according to Brenner and Morris (1990), the polymorphic information content according to Botstein et al. (1980), the probability of match and the discrimination power according to Jones (1972) and the distinct heterozygous and homozygous genotypes according to Chakraborty (1993). The frequency profile comparison between Italian and German populations was carried out using a test for genetic heterogeneity ($R \times C$ contingency table; G. Carmody, Ottawa, Canada).

Results and discussion

For HumACTBP2 a total of 39 alleles was observed, none of which exceeded 10% in frequency (Table 1). The number of alleles observed at this locus confirmed the high degree of polymorphism and the resulting efficiency values (Table 2) demonstrate the usefulness of ACTBP2 for forensic casework. In contrast to a larger German population survey two alleles which did not match the ladder alleles (N18m + N19m) below N20 were observed in the Italian population sample. Their frequencies were assigned to the next anodal alleles (Möller et al. 1995). Furthermore, an allele > N35 was found. A test for heterogeneity revealed significant differences ($P < 0.05$) between the Italian an the German populations (Table 2).

Table 1 Allele frequency distribution for HumACTBP2 in an Italian population sample (Milan area) and comparison with a larger German (Münster area) survey. The prefix "N" indicates the application of a non-denaturing (native) gel system. Alleles showing the same electrophoretic mobility under denaturing conditions, but different positions in a non-denaturing gel system are marked by an asterisk. The "consensus nomenclature" introduced by the ACTBP2 working group of the German Society of Legal Medicine and published by Rolf et al. (1997) is given in brackets. n = number of individuals

Allele	Allele frequencies	
	Italians (n = 118)	Germans (n = 560)
N12 (12)	–	0.0054
N13 (13.2)	0.0183	0.0054
N14 (14)	0.0494	0.0321
N15 (15)	0.0183	0.0321
N16 (16)	0.0494	0.0536
N17 (17)	0.0763	0.0625
N18 (18)	0.0853	0.0643
N19 (19)	0.0986	0.0679
N20 (20)	0.0717	0.0643
N21 (21)	0.0361	0.0518
N22 (22.2)	0.0092	0.0304
N23 (22.2*)	0.0450	0.0161
N24 (22.2**)	0.0362	0.0375
N25 (23.2)	0.0317	0.0304
N26 (26.2)	0.0450	0.0375
N27 (25.2)	0.0540	0.0696
N28 (27.2)	0.0495	0.0643
N29 (27.2*)	0.0986	0.1071
N30 (28.2)	0.0718	0.1035
N31 (29.2)	0.0049	0.0269
N32 (30.2)	0.0273	0.0196
N33 (33.2)	0.0048	0.0142
N35 (34)	0.0093	0.0035
> N35	0.0093	0

Table 2 Statistical data for a) HumACTBP2 and b) HumVWA in the Italian population sample. The statistical evaluations were carried out as outlined in the text

HumACTBP2		HumVWA	
H_{obs}	0.9182	H_{obs}	0.7966
H_{exp}	0.9429 ± 0.0434 (± 1.96 SE)	H_{exp}	0.8158 ± 0.0700 (± 1.96 SE)
MEC	0.8564	MEC	0.6282
MEP	0.8836	MEP	0.6286
PIC	0.9352	PIC	0.7862
pM	0.0165	pM	0.0653
D	0.9835	D	0.9346

H_{obs} = observed heterozyosity, H_{exp} = expected heterozygosity, SE = standard error
MEC = mean exclusion chance, MEP = mean paternity exclusion probability
PIC = polymorphic information content; pM = match probability
D = discrimination power

Table 3 Allele frequency distribution for HumVWA in an Italian population and comparison with a larger German population sample. n = number of individuals

Allele	Allele frequencies	
	Italians (n = 118)	Germans (n = 1510)
13	–	0.0013
14	0.0974	0.0974
15	0.1229	0.0987
16	0.2246	0.2040
17	0.2670	0.2795
18	0.1822	0.2212
19	0.0890	0.0854
20	0.0169	0.0126
21	–	0.0020
22	–	0.0006

In contrast to a previous study on two different Italian population samples, where eight HumVWA alleles could be detected (Buscemi et al 1995) only seven alleles could be observed in our population survey (N = 118), due to the absence of allele 11 (Table 3). Nevertheless, no significant differences between the Italian populations could be observed applying the Carmody test.

The population data showed no significant deviations from Hardy-Weinberg equilibrium (Table 2).

Northern Italy and the Milan area have been subjected to migration from all parts of the country, especially from Southern Italy, for at least one century. Therefore our data could well be representative of the entire Italian population, however more data on other Italian populations are necessary to confirm this assumption.

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