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A multicentric study of SE33 allele frequencies in the Italian population

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Abstract

Allele and genotype frequencies for STR SE33 were obtained for a sample of 419 Italians in view of application in personal identification and paternity.

Keywords: SE33; Population study; Forensic applications

1. Introduction

The STR locus SE33 is a complex tetranucleotide and it is one of the most informative and polymorphic genetic markers. The aim of the present work was to establish the haplotype frequencies in an Italian population sample, to evaluate their effectiveness in forensic identification and paternity. For this purpose, collaborative research on the polymorphism of system was carried out by the Institutes of Legal Medicine in four Italian Provinces (Ancona, Padova, Pavia and Pisa).

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Table 1
Genotype frequency spectra in the four subsamples

No. of individuals per genotype	AN	PD	PI	PV
4	3	1	0	0
3	1	1	7	8
2	15	20	12	13
1	63	61	55	53
No. of different genotypes	82	83	74	74
No. of individuals	108	108	100	103
% of unique genotypes	58.3	56.5	55.0	51.5

Table 2
Distribution of the allele frequencies by subsample

Allele	AN	PD	PI	PV	Total
18	31	20	23	18	92
19	16	20	20	24	80
17	13	19	20	18	70
29.2	17	15	15	20	67
27.2	17	16	9	12	54
20	11	10	16	16	53
16	15	13	11	11	50
28.2	11	14	11	12	48
30.2	8	13	7	15	43
21	8	9	10	6	33
26.2	9	10	6	4	29
24.2	13	8	2	4	27
15	6	4	10	5	25
25.2	8	7	5	4	24
23.2	9	2	6	6	23
22.2	6	5	4	7	22
14	2	7	8	3	20
31.2	4	3	3	5	15
32.2	2	4	2	3	11
22		5	1	3	9
20.2		2	4	1	7
13		3	1	2	6
21.2	2		1	2	5
15.2		3			3
33.2	1	1		1	3
12	2				2
12.2	1		1		2
14.2		2			2
23	1			1	2
33			1	1	2
34		1		1	2
11	1				1
13.2			1		1
25			1		1
26				1	1
28	1				1
29	1				1
32			1		1
Total	216	216	200	206	838

2. Materials and methods

The database was drawn from 419 fresh blood samples collected from unrelated donors. Analysis was carried out using the amplification conditions proposed by Wiegand et al. [1], with slight modifications. Haplotypes were identified by an automated system (ABI Prism 310-ABD/PE) and allele assignment was performed by comparison with allelic ladder kindly provide by the Institute of Legal Medicine in Lausanne. The nomenclature used is in accordance with the recommendations of GEDNAP [2]. The observed alleles not present in the allelic ladder were all sequenced (data not shown).

3. Results and discussion

3.1. Variability at the genotype level

The total sample (419 individuals) included 180 different genotypes, the most frequent being type 18–19 ($n = 11/419$, $p_i = 0.026$). This genotype was also among the most frequent observed genotypes in each center. Table 1 shows the genotype frequency spectra in the four centers. The percentage of unique genotypes (last row) was included between 52% and 58%.

3.2. Variability at the gene level

A total of 38 different alleles were observed, 19 of which (50%) were common to all four subsamples, 7 alleles were present in only one subsample. Table 2 shows the distribution of the allele frequencies by subsample, ordered by decreasing total frequency. The heterozygosity was 0.939, making this marker one of the most discriminating loci

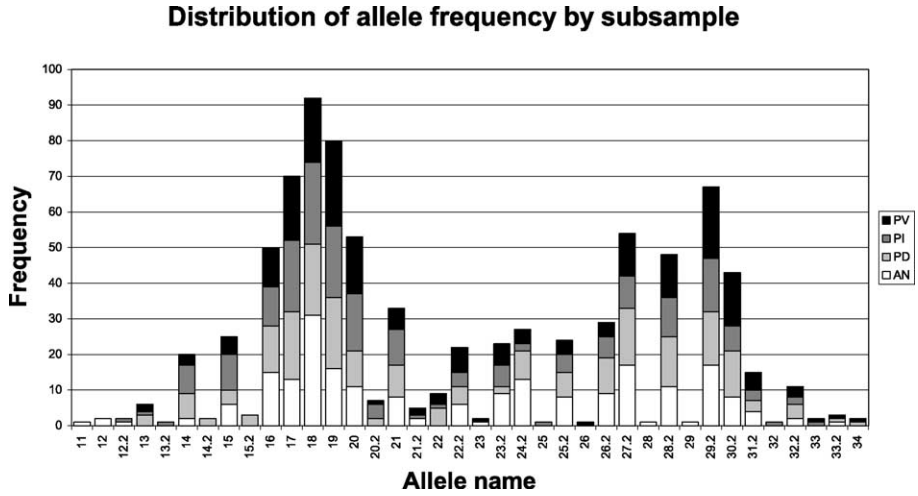

Fig. 1. Distribution of allele frequency in the total sample.

Table 3
Observed and expected number of homozygous and heterozygous genotypes

Genotypes	AN	PD	PI	PV	Total
Observed					
Heterozygous	101	100	93	98	392
Homozygous	7	8	7	5	27
Total	108	108	100	103	419
Expected					
Heterozygous	101.5	101.5	93.9	96.8	393.6
Homozygous	6.5	6.5	6.1	6.2	25.4
Total	108	108	100	103	419

currently available in forensic science. Fig. 1 shows the distribution of allele frequency in the total sample, as a stack column chart of the four subsamples. The homogeneity of the four subsamples is apparent.

3.3. Homozygosity test

Table 3 shows the total number of observed and expected number of homozygous and heterozygous genotypes in each subsample and in the total sample. The agreement with the HWE is excellent.

References

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