Allele frequencies of the HUMFES/FPS system in northern and central Italy

ALLELE FREQUENCIES OF THE HUMFES/FPS SYSTEM IN NORTHERN AND CENTRAL ITALY.

Tagliabracci A., Paoli M., Rodriguez D. (*)
Cucurachi N., Buscemi L., Ferrara S.D. (**)
Previderè C., Peloso G., Riva A., Pierucci G. (°)
Domenici R., Fornaciari S., Spinetti I., Nardone M., Bargagna M. (°°)

Universities of Ancona (*), Parma (**), Pavia (°), Pisa (°°)

INTRODUCTION

The tetrameric (ATTT) short tandem repeat HUMFES/FPS system, located on chromosome 15 (15q25-qter), displays a polymorphism (Polymeropoulos et al. 1991) which has been investigated for forensic purposes (Hammond et al. 1994). The aim of the present work was to study the polymorphism of this system in a large Italian population sample: 1) to verify allele frequency distribution, 2) to check whether allele frequencies show inter-regional differences, 3) to evaluate the effectiveness of this system for paternity testing and personal identification. For this purpose, collaborative research was performed on subjects from four Italian regions in laboratories of the Institutes of Legal Medicine of the Universities of Pavia (Lombardy), Parma (Emilia), Pisa (Tuscany) and Ancona (Marches).

MATERIALS AND METHODS

The study was carried out on fresh blood samples collected from healthy unrelated donors living in Pavia (n= 120), Parma (n=150), Pisa (n=162) and Ancona (n=159). The four laboratories conducted analyses according to the method described by Möller *et al.* (1994) with minor modifications; electrophoresis was carried out on a non-denaturing polyacryamide gel and the bands were visualized by silver staining (Budowle et al. 1991). Alleles were identified by side-to-side comparison with home-made ladders consisting of a cocktail of amplified products (fig. 1).

The 4 Italian population samples were tested for heterogeneity with an R x C contingency table using a computer program kindly supplied by G. Carmody (Carleton University, Ottawa, Canada). The results were verified by the chi-square test between observed and expected genotypes according to the Hardy-Weinberg law and by comparison of the observed and expected heterozygosity frequencies, the latter calculated as allele diversity (Nei et al. 1974). $\sqrt{[h(1-h)/N]}$ was the formula used to compute the standard error for H, where h was the expected heterozygote frequency and N the number of subjects examined.

The power of discrimination (PD) was calculated using Fischer's (1951) equation. The exclusion chance was calculated from allele frequencies (Garber and Morris 1983).

Table 1. HUMFES/FPS observed genotypes in 591 Italians from four regions.

Genotypes	Observed
8-10	2
8-11	2
8-12	2
9-11	1
10-10	45
10-11	122
10-12	92
10-13	20
10-14	2
11-11	89
11-12	115
11-13	28
11-14	5
12-12	38
12-13	20
12-14	3
13-13	4
13-14	1

Table 2. HUMFES/FPS allele frequency distribution from 591 Italians.

HUMFES/FPS Allele	Lombardy 120 subjects	Emilia 150 subjects	Tuscany 162 subjects	Marches 159 subjects	Combined data
8	0.013	0.007	_	0.003	0.005
9	_	0.003	_	_	0.001
10	0.27	0.26	0.287	0.289	0.277
11	0.387	0.387	0.358	0.396	0.382
12	0.25	0.277	0.278	0.236	0.261
13	0.058	0.06	0.068	0.072	0.065
14	0.020	0.007	0.009	0.003	0.009

Heterogeneity test: $\chi^2 = 15.9478$, $P = 0.6110 \pm 0.0154$;
G statistic = 16.2560 , $P = 0.6380 \pm 0.0152$

a) H-W equilibrium expectation

$\chi^2 = 4.887$ $P > 0.975$ $df = 21$

b) Obs. Heterozy. = 0.70 Exp. Heterozy. = 0.70 ±0.018

c) PD = 0.86 Mean excl. chance = 0.45

RESULTS AND DISCUSSION

591 subjects living in 4 Italian regions were typed for STR system HUMFES/FPS. A total of 18 genotypes (Table 1) and the 7 common alleles (Table 2) identified by Hammond et al. (1994) were found and classified according to the repeat number. The two anodal variant alleles 10a and 11a, recently described by Möller et al. (1994), were pooled together with alleles 10 and 11, respectively.

The 4 samples showed similar distribution of allele frequencies when tested for heterogeneity (Table 2), so that they can be considered as a single Italian population sample.

The PD and mean esclusion chance were, respectively, 0.86 and 0.45, suggesting that the HUMFES/FPS system is a useful and powerful tool for forensic purposes.

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Fig 1. Silver-stained HUMFES/FPS system polyacrylamide gel of amplified fragments. Left to right: ladder (alleles 8,10,11,12,13,14): lane 1,4,7. HUMFES/FPS phenotypes: lane 2: 10-11; lane 3: 10-12; lane 5: 10-11; lane 6: 10-11.