SHORT COMMUNICATION

A. Piccinini · K. Möller · P. Wiegand

HumFES/FPS and HumF13B: population genetic data from North Italy

Received: 17 July 1995 / Accepted: 31 October 1995

Abstract DNA extracted from 119 unrelated individuals was analysed by the polymerase chain reaction at the polymorphic microsatellite loci HumFES/FPS (n=115 individuals) and HumF13B (n=119 individuals). The samples were collected from Caucasians living in the area of Milano (northern Italy). After horizontal polyacrylamide electrophoresis, 8 alleles were observed for HumFES/FPS, and 5 for HumF13B. Testing for Hardy-Weinberg equilibrium showed no significant deviation. The allele frequency data were compared with a German and a Turkish population sample.

Key words Short tandem repeats · HumFES/FPS · HumF13B · Population studies · Northern Italy

Introduction

Microsatellite polymorphisms represent a widely used method for linkage studies and human identification (Edwards et al. 1991), even on highly degraded DNA (Brinkmann 1992).

This study was carried out with the systems FES/FPS (Polymeropoulos et al. 1991) and F13B (Nishimura and Murray 1992), on a northern Italian population to increase the population genetic database for different Caucasian groups.

Materials and methods

DNA was extracted from 200 μ l blood stains air-dried on sterile cotton fabric, from unrelated Caucasian individuals residing in the Milano area. The total number of tested samples for the two systems FES/FPS and F13B was 115 and 119 respectively.

A. Piccinini (國) Istituto di Medicina Legale, Università degli Studi di Milano, Via L. Mangiagalli 37, I-20133 Milano, Italy

K. Möller · P. Wiegand Institute of Legal Medicine, Westfälische-Wilhelms-Universität, Von-Esmarch-Strasse 86, D-48149 Münster, Germany The extraction procedure was carried out using 150 μ l Chelex 100 (5%) (Biorad, Germany) with the addition of 50 μ l proteinase K (2 mg/ml) as previously described (Wiegand et al. 1993).

The reaction assay amplification and electrophoresis conditions were carried out as previously described (Möller et al. 1994; Alper et al. 1995). Alleles were designated according to the number of repeats (Alper et al. 1995).

Isolation of fragments from the gel, Taq-Cycle sequencing and analysis of the sequence data were carried out as previously described (Möller and Brinkmann 1994).

The Hardy-Weinberg analysis was performed with standard Chi-square analysis and the logarithmic likelihood ratio (G) test using the exact test by randomly shuffling the observed alleles 5000 times (HWE analysis, Version 3.0, C. Puers, Münster, Germany). The comparison of observed with expected numbers of heterozygotes (gene diversity) was calculated according to Nei (1978), the mean exclusion chance was calculated according to Brenner and Morris (1990), the polymorphic information content according to Botstein et al. (1980), and the probability of match and the discrimination power according to Jones (1972). The frequency profile comparisons were performed using a test for genetic heterogeneity (2-way R × C contingency table; Chi-square and G statistic; Carmody, Ottawa, Canada).

Results and discussion

A total of 8 alleles was observed for the system Hum-FES/-FPS and 5 for HumF13B (Table 1). The use of high resolution non-denaturing gels, combined with the use of specific allelic ladders allowed the distinction of microheterogeneities caused by base substitutions (Möller et al. 1994; Alper et al. 1995). The typing results for Hum-FES/FPS in the Italian population showed no significant differences in comparison with German data (R × C contingency table; Chi-square: $P = 0.21 \pm 0.0129$ SE; G statistic: $P = 0.136 \pm 0.0108$ SE) and with Turkish data (Chi-square: $P = 0.465 \pm 0.0158$ SE; G statistic: $P = 0.439 \pm 0.0157$ SE) (Table 1). Additionally, an allele with 7 repeats was found (confirmed by sequencing), which has not been observed previously in the German population but had been typed in a Turkish survey (Alper et al. 1995).

HumF13B population data showed slight frequency differences when compared with a larger German survey. In particular, a clearly higher frequency was observed for

Table 1 Alleles observed for systems HumFES/FPS and HumF13B in the present study, with distributions in a German and a Turkish study for comparison. System of Alper et al. (1995) is used for numeration of alleles (*C* cathodal, *A* anodal)

Hum FES/FPS ^a Allele	Allele frequencies		
	Italians	Germans	Turks
7	0.0043	_	0.003
8	0.0087	0.0144	0.007
9 .	_	0.0054	0.005
10A	0.2391	0.2370	0.209
10	0.0566	0.0494	0.037
11A	0.0130	0.0332	0.017
11	0.40	0.4165	0.372
12	0.2261	0.2002	0.299
13A	_	0.0009	_
13	0.0522	0.0413	0.044
14	_	0.0017	0.007
HumF13B ^b			
6	0.0378	0.0982	0.075
7	0.0084	0.0099	0.030
8	0.2311	0.2378	0.315
9	0.2227	0.2234	0.243
9C	_	0.0009	-
10	0.50	0.4261	0.335
10C	_	0.0018	_
11	_	0.0009	0.003
11C	_	0.0009	_

 ^a Population sizes: Italians 115, Germans 557, Turks 203
^b Population sizes: Italians 119, Germans 555, Turks 200

Table 2 Statistical data for HumFES and HumF13 B (*D* discrimination power, *MEC* mean exclusion chance, *MEP* mean exclusion probability, *PIC* polymorphic information content, *pM* match probability)

Heterozygosity	FES	F13B
Observed Expected	0.687 0.734 ± 0.08 (SE)	0.588 0.648 ± 0.08 (SE)
MEC	0.5024	0.3842
MEP	0.4827	0.3528
PIC	0.6892	0.5876
pM	0.1155	0.1787
D	0.8845	0.8213

the allele 10 in the Italian population. Furthermore, no "intermediate" alleles were observed in the present population analysis (Table 1). Nevertheless, no significant differences between these two populations were observed (Chi-square: $P = 0.18 \pm 0.0121$ SE; G statistic: $P = 0.059 \pm 0.0075$ SE), while a comparison between Italian and Turkish allele frequency data showed significant differences (Chi-square: P = 0.0; G statistic: $P = 0.001 \pm 0.001$ SE).

No significant deviation from Hardy-Weinberg equilibrium could be detected (P > 0.05). Owing to allele number and distribution, FES had higher forensic efficiency values than F13B (Table 2).

References

Alper B, Meyer E, Schürenkamp M, Brinkmann B (1995) Hum-FES/FPS and HumF13B: Turkish and German population data. Int J Legal Med 108:93–95

Botstein D, White RL, Skolnick M, Davis RW (1980) Construction of a genetic linkage map in man using restriction fragment length polymorphisms. Am J Hum Genet 32:314–331

Brenner C, Morris J (1990) Paternity index calculations in single locus hypervariable DNA probes: validation and other studies. In: Proceedings of the international symposium on human identification 1989. Promega Corporation, Madison, pp 21–53

Brinkmann B (1992) The use of STRs in stain analysis. In: Proceedings from the third international symposium on human identification. Promega Corporation, Madison, pp 357-373

Edwards A, Civitello A, Hammond HA, Caskey CT (1991) DNA typing and genetic mapping with trimeric and tetrameric tandem repeats. Am J Hum Genet 49:746–756

Jones DA (1972) Blood samples: probabilities of discriminations. J Forensic Sci Soc 12:355–359

Möller A, Brinkmann B (1994) Locus ACTBP2 (SE33): sequencing data reveal considerable polymorphism. Int J Legal Med 106:262–267

Möller A, Meyer E, Brinkmann B (1994) Different types of structural variation in STRs: HumFES/FPS, HumVWA and Hum-D21S11. Int J Legal Med 106:319–323

Nei M (1978) Estimation of average heterozygosity and genetic distance from a small number of individuals. Genetics 89:583—590

Nishimura DY, Murray JC (1992) A tetranucleotide repeat for the F13B locus. Nucleic Acids Res 20:1167

Polymeropoulos MH, Roth DS, Xiao H, Merril RM (1991) Tetranucleotide repeat polymorphism at the human c-fes/fps proto-oncogene (FES). Nucleic Acids Res 19:4018

Wiegand P, Bajanowski T, Brinkmann B (1993) PCR typing of debris from fingernails. Int J Legal Med 106:81–84