TYPING FOR THE HUMFES/FPS SHORT TANDEM REPEAT POLYMORPHISM IN AN AUSTRIAN CAUCASOID POPULATION SAMPLE

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SYSTEM AND LOCUS

HUMFES/FPS (intron 5 of the c-fes/fps proto-oncogene, 15q25-qter; Genbank Acession No.X06292 [Polymeropoulos 1991])

POPULATION AND SAMPLE SIZE: Vienna region (Austria). N=451

METHODS

primers [Moeller 1994]

A: 5'-ggg att tcc cta tgg att gg-3'
B: 5'-gcg aaa gaa tga gac tac at-3'

PCR

Amplification was carried out according to the protocol published by Moeller [1994] with some modifications:
reaction volume of 50µl
additional first denaturing step (5min - 98°C)
Hybaid Omnigene thermocycler

Electrophoresis

PAGE and silver staining was carried out on 6% native polyacrylamide gels in 112mM Tris/Acetic Acid and 200mM Tris/Tricine/0.55% SDS in horizontal mode as previously described [Schwartz 1994].

AMPFLP Typing

Typing was performed by side-to-side comparison with a sequenced allelic ladder, containing alleles 8 to 14, kindly provided by B. Brinkmann.

Sequencing

Automated single strand sequence determination of alleles corresponding to the ladder alleles was performed on an automatic DNA-Sequencer (ALF™, Pharmacia LKB Technology) according to the protocol of the Pharmacia AutoRead™ Sequencing Kit (dye primers, T7 polymerase) on a 6% sequencing gel. For each sample, the sense and the antisense strand were sequenced.

RESULTS AND COMMENTS

AMPFLP typing

AMPFLP Typing for the HumFES/FPS polymorphism in 451 healthy, unrelated Austrian Caucasoid individuals revealed 9 distinct alleles, corresponding to alleles 8-13 (including 10a and 11a; [Moeller 1994]) and an additional low frequency allele corresponding to 14 core repeats that has also been previously described [Hammond 1994]. Allele frequencies, together with the forensic statistical efficiency values are listed in Tab.1. The phenotype distribution showed no significant deviation from Hardy-Weinberg expectations.

Table 1: HUMFES/FPS allele frequencies (n=451)

allele designation	frequency (%)
8	1.7
9	0.3
10a	23.4
10	3.3
11a	2.3
11	43.5
12	22.5
13	2.9
14	0.1

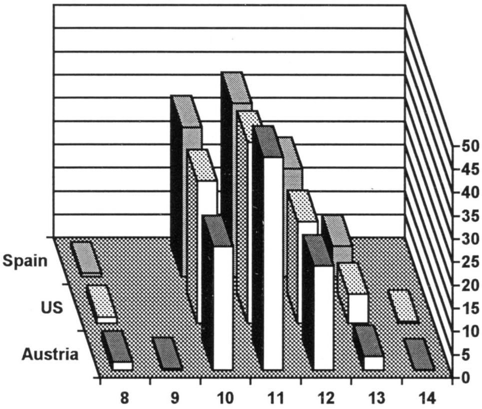
χ^2=9.76; df=10; 0.4<p<0.5

Mean exclusion chance (MEC) 0.48

Power of discrimination (PD) 0.86

Discrimination index (DI) 0.15

Fig. 1: Comparison of HUMFES/FPS allele frequencies



Austrian versus

US Caucasians: χ^2= 15.88; df=6; 0.01<p<0.025

Spanish Caucasians: χ^2= 22.21; df=6; 0.001<p<0.005

(to allow the comparison alleles 10a and 11a were counted with 10 and 11, respectively

When comparing the allelic distribution of our population to other Caucasian samples [Cabrero 1994, Hammond 1994], a statistically significant deviation in the distribution of alleles 10, 11 13 was observed (Fig.1).

Sequencing:

Sequence determination of 13 samples including all alleles was in perfect agreement with the already published data [Moeller 1994]. The allele 14 (sequence not published so far) showed just another reiteration of the core repeat and no transversion at position 34 of the 5' flanking region.

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