STUDY OF HUMACTBP2 STR POLYMORPHISM, PERFORMED BY PCR AND AUTOMATED LASER FLUORESCENCE (ALF) SEQUENCER IN A POPULATION SAMPLE OF CATALONIA

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System and Locus: HUMACTBP2 (also named SE33). Located in the 5' flanking region of the human beta-actin related pseudogene H-beta-Ac-psi-2 (Moos 1983) on chromosome 5 (Warne 1991) or 6 (Polymeropoulos 1992). $(AAAG)_n$ tandem repeat.

Population and sample sizes: Catalonia (NE Spain). Western Mediterranean caucasoid population. N:154

Methods: Standard PCR amplifications were accomplished with fluorescein labelled primers described by Polymeropoulos (1992). Electrophoretic methods: 6% polyacrylamide denaturing gel electrophoresis. The gels were run for 4h. at 1450 V, 38 mA, 45W, 50° C and laser power at 3 mW on the ALF DNA Sequencer. Cocktail allelic ladder of sequenced known alleles and nomenclature proposed by Möller (1994) were used.

Results: Table 1. Observed ACTBP2 genotypes in a sample population of Catalonia

	7-1	13-2	13	14	15	16	17	18	19	20	21-2	21	22-4	22-2	22	23	24	25	26	27	28	29	30	31	32	33-1	33	34-2	34	35	42
7-1			1																												
13-2						1																									
13					1	1															1					1					
14				1		1	1			2					1		1			2	1	1	1				1				
15					1	1		2		1					1		2		1		1	1									
16							3	3	1	2					1	1					1	2	2		1	1		1			
17								1	2		2		1	1	1	3	2	1				3	1	1	1						
18								1	5	3			1	1	1		1		1		3	1	2	2	1						
19									1	3									2	2			1	1			1	1			
20																		1		1	1	2		1							
21-2													1									1									
21												1			1					1		1		1					1		1
22-4													1			1		2	1		1	1									
22-2																			1												
22																	1		1												
23																															
24																			1												
25																					1		1								
26																			1	2	1		1			1					
27																				2	2			1	1						
28																					1	1									
29																							3						1	1	
30																															
31																															
32																															
33-1																															
33																															
34-2																															
34																															
35																															
42																															

Table 2. Allele frequencies

	Allele	Frequency		Allele	Frequency
1.-	07-1 =	0.0032	17.-	24 =	0.0260
2.-	13-2 =	0.0032	18.-	25 =	0.0195
3.-	13 =	0.0162	19.-	26 =	0.0519
4.-	14 =	0.0454	20.-	27 =	0.0584
5.-	15 =	0.0422	21.-	28 =	0.0519
6.-	16 =	0.0747	22.-	29 =	0.0649
7.-	17 =	0.0844	23.-	30 =	0.0390
8.-	18 =	0.0974	24.-	31 =	0.0227
9.-	19 =	0.0682	25.-	32 =	0.0130
10.-	20 =	0.0552	26.-	33-1 =	0.0097
11.-	21-2 =	0.0130	27.-	33 =	0.0065
12.-	21 =	0.0260	28.-	34-2 =	0.0065
13.-	22-4 =	0.0357	29.-	34 =	0.0065
14.-	22-2 =	0.0130	30.-	35 =	0.0032
15.-	22 =	0.0260	31.-	42 =	0.0032
16.-	23 =	0.0130			

Table 3. Forensic diagnosis suitability results.

	HUMACTBP2
Heterozygosity Index (HI)	93.50
Power discrimination (PD)	0.99
Chance Exclusion (CE)	0.89
Essen-Möller mean value (EM)	8.94

Comments: HUMACTBP2 has a polemic length and sequence polymorphism [4] because a high number of alleles have been described, some of which may vary by as little as 1 base, moreover its AT-rich sequence may have anomalous migration rates in different electrophoretic systems. Nevertheless as it is one of the most powerful PCR markers, we describe the experience in our laboratory. A high degree of variability observed in a population sample of Catalonia, could makes it an extremely useful marker in forensic genetics diagnosis. In general, HUMACTBP2 is an interesting polymorphism that needs standardization of the experimental conditions, in order to obtain allele identification that is reproducible in all forensic laboratories. In this sense, the use of sequenced allelic ladder is very important.

References

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