

## ALLELIC FREQUENCY IN THE POPULATION OF SPAIN USING THREE HYPERVARIABLE LOCI

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### INTRODUCTION

Tandemly repeated sequences represent a rich source of highly polymorphic markers useful in genetic linkage analysis, genome mapping and personal identification. The interpretation of the results from the analysis of these genetic markers, utilized in forensic and paternity testing, requires the existence of a database for the appropriate population.

Studies to determine the allele frequencies of single locus have been carried out for some world populations. The substructure of populations and their influence in forensic cases has generated a great deal of controversy (Lewontin and Hartl, 1991; Chakraborty and Kidd, 1991). We have constructed spanish population databases for the SLPs, YNH24, TBQ7, MS31 and MS43 (Valverde et al, 1993). In order to complete the spanish databases, we have studied other polymorphic loci. In the present paper we report the databases for five different loci in the spanish population. One of these markers is the D14S13 locus, detected by the CMM101 probe (Nakamura et al., 1988a). The other four loci are detected by PCR amplification followed by electrophoretic separation. The four analyzed loci are: D1S80 (Nakamura et al., 1988b; Budowle, 1991), HUMTH01 (Polymeropoulos et al., 1991a), HUMFES/FPS (Polymeropoulos et al., 1991b) and ACTBP2 (Polymeropoulos, M.H. et al., 1992)

### MATERIALS AND METHODS

**SAMPLES:** DNA was extracted from 190 unrelated volunteer donors, randomly selected from Spain. The high molecular weight DNA was extracted following the perchlorate protocol as described (Valverde et al., 1992; Díez et al., 1992).

**D14S13 LOCUS:** 5-10 µg of DNA were HaeIII digested, and the resultant fragments electrophoresed and blotted following the EDNAP protocol recommendations. The CMM101 probe was <sup>32</sup>P labelled by random priming.

**LOCI DETECTED BY PCR (D1S80, HUMFES, HUMTH01, ACTBP2):** All the amplification were performed in a final volume of 50 µl in a Perkin-Elmer thermocycler. The different cycle temperatures, primers sequences and electrophoresis conditions were as described (Budowle, 1991; Polymeropoulos et al., 1991a,b, 1992)

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## RESULTS

Figures 1 and 2 shows the allele frequency distribution for the D14S13 and D1S80 loci. The allele frequency distribution in the spanish population is similar to other already published for white-caucasian individuals. A total of 24 different D1S80 alleles were observed in 156 individuals. Two alleles (#18 and 24) were found to be relatively common in the Spanish sample having frequencies of 0.175 and 0.305, respectively. Similarly, for STR's locus HUMTH01 (Fig 3), there were 6 alleles observed in 190 individuals with 4 alleles occurring at a frequency greater than 0.100. We have found a mutation in the allele number 5 of the HUMTH01 locus produced by the addition of a single nucleotide. There were 6 alleles for HUMFES locus (Fig 4) with 3 of them relatively common. Allele number 2 was not found in any case for this locus. HUMFES and HUMTH01 loci have a similar allele distribution to other caucasian population groups. ACTBP2 locus (Fig 5) shows a variation in the alleles mobility depending the electrophoretic conditions, as it has been reported for other AT-rich VNTR (Sullivan et al., 1992). This fact can represent a potential problem for the interlaboratory data comparison and protocol standardization.

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FIGURE 1

**CMM101**

SPANISH POPULATION (n = 162)

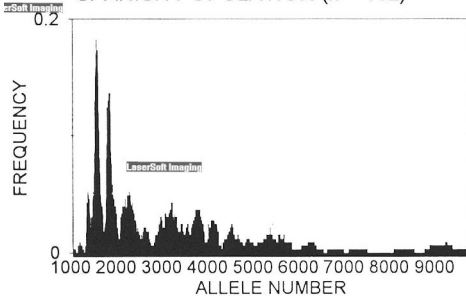


FIGURE 2

**D1S80**

SPANISH POPULATION (n = 156)

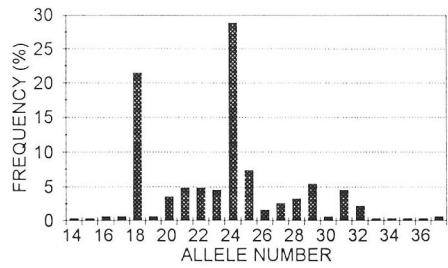


FIGURE 3

**HUMTH01**

SPANISH POPULATION (n = 192)

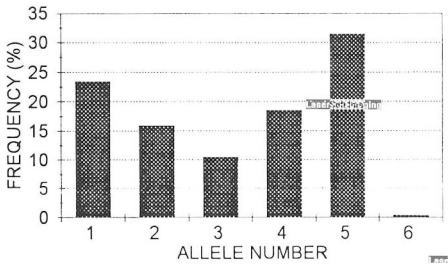


FIGURE 4

**HUMFES/FPS**

SPANISH POPULATION (n = 190)

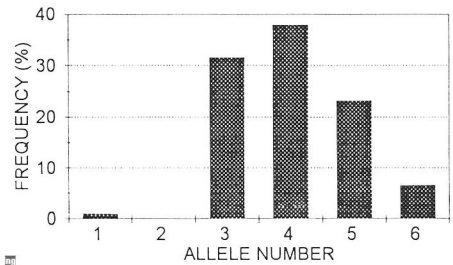


FIGURE 5

**ACTBP2**

SPANISH POPULATION (n = 116)

