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SHORT COMMUNICATION

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Polymorphism analysis of the VNTR locus D17S5 in Central Spain

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Abstract The fragment length polymorphism YNZ22 (D17S5) was analysed for a sample of 207 unrelated individuals living in Madrid (Spanish Caucasians) using PCR-methodology and high resolution separation. Hardy-Weinberg expectations (HWE) were calculated after pooling alleles into four groups. No deviations from HWE were detectable using the conventional χ^2-test. The power of discrimination was estimated as 0.96 and the mean paternity exclusion chance as 0.7587. A comparison of the allele frequency distribution with those of other Caucasian groups revealed no major differences.

Key words Population genetics · PCR-FLP · D17S5 · Spanish population · YNZ22

Introduction

Locus D17S5 is a highly polymorphic VNTR marker which has been localized on chromosome 17 and linked to the Miller-Dieker syndrome locus (Batanian et al. 1990). Its heterozygosity index is higher than 80% in mixed Caucasians (Nakamura et al. 1987). Horn et al. (1989) designed primers for the unique flanking region to study the D17S5 polymorphism by PCR. Amplified fragments comprise repetitions of the core (70bp) plus the size of the primers (20 and 21 bp) and a 57 bp additional flanking region. In this study we present data for D17S5 (YNZ22) in a sample of 207 unrelated Caucasian individuals from Madrid in Central Spain.

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Materials and methods

DNA for AMP-FLP analysis was obtained from peripheral blood specimens from a random sample of 207 non-related blood donors of Spanish descent at the Regional Transfusion Centre in Madrid by standard procedures. Methods for amplification, fragment analysis, Southern-blotting and hybridization comprised minor modifications to previously described protocols (Batanian et al. 1990; Ugozzoli et al. 1991; Deka et al. 1992; Buscemi et al. 1994). Alleles were sequentially named according to the number of repeat units (1 for the smallest allele). Forensic parameters were calculated as recommended in previous studies (Fisher 1951; Nei and Roychoudhury 1974; Buscemi et al. 1994).

Results and discussion

The population studied contained 12 alleles plus a new extremely large one. Other authors have also reported the existence of very high molecular weight alleles (Deka et al. 1992; Rand et al. 1992; Buscemi et al. 1994). The lack of the 13 tandem repeat allele can be due to the size of the studied sample, given the low frequency of large alleles in general. Most of the alleles could be detected with ethidium bromide staining and ultraviolet transillumination of the gels. However, high molecular weight alleles and homozygous genotypes were re-examined by Southern blot hybridization to confirm weak bands as alleles and to look for the presence of high molecular weight alleles not detected by ethidium bromide staining.

Figure 1 shows the estimated allele frequencies for the D17S5 locus. A total fo 56 genotypes corresponding to 13 alleles were found in the 207 individuals studied. Based on the "allele binning strategy" (Brenner and Morris 1990), the χ^2 analysis for Hardy-Weinberg equilibrium

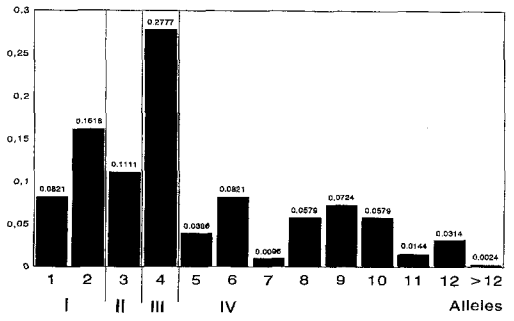

Fig. 1 Frequency distribution of YNZ22 system alleles in a population sample of 207 unrelated caucasians from Madrid, Spain. H-W expectations were calculated by using the four bin (I–IV) groups of alleles suggested by Rand et al. 1992

hypothesis comprised the four bin groups suggested by Rand et al. (1992). No deviation from Hardy-Weinberg equilibrium was observed ($\chi^2 = 7.962$, df = 6, $0.25 < p < 0.20$). The estimated heterozygosity was 83.09% and the allelic diversity (H) was calculated according to Nei and Roychoudhury (1974) as 0.9273 ± 0.0180. The paternity chance of exclusion (CE) was evaluated through a computer programme provided by Carracedo (Santiago de Compostela) as 0.7587 and the power of discrimination (PD) calculated according to Fisher (1951) was 0.96.

As already reported (Batanian et al. 1990; Deka et al. 1992; Rand et al. 1992; Buscemi et al. 1994) low molecular weight alleles comprise more than the 50% of the total. The most frequent alleles were, in decreasing order, YNZ22*4, YNZ22*2 and YNZ22*3, the rest being much less common. The YNZ22*7 frequency was below 1% as well as, in general, those alleles with more than 12 tandem repeats. Of the larger alleles, YNZ22*9 and YNZ22*10 were the most frequent and, in general, our population frequencies closely resemble those established for several Caucasian groups (Batanian et al. 1990; Rand et al. 1992; Deka et al. 1992; Buscemi et al. 1994; Gené et al. 1995).

In conclusion, the population sample analysed shows an allelic frequency distribution in line with those already studied. According to the medico-legal parameters (H, CE, PD), D17S5 is a highly polymorphic marker and represents a powerful tool for personal identification and paternity analysis.

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