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SHORT COMMUNICATION

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Suitability of the YNZ22 (D17S5) VNTR polymorphism for legal medicine investigations in the population of Catalonia (Spain)

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Abstract Allele and phenotype frequencies for the YNZ22 locus were determined in a population sample from Catalonia (Spain) using the polymerase chain reaction (PCR). In 311 unrelated individuals, 14 alleles and 56 phenotypes were observed. No deviation from Hardy-Weinberg equilibrium was found. The observed heterozygosity was 81.35%. The YNZ22 polymorphism is useful for paternity testing with a CE value of 70% and an Essen-Möller value of 9.35 (log.)

Key words VNTR · YNZ22 · D17S5 · Population genetics · Paternity testing

Zusammenfassung Allelfrequenzen und phänotypische Häufigkeiten des Locus YNZ22 wurden in einer katalanischen Bevölkerungsstichprobe (Spanien) mittels der Polymerase-Kettenreaktion bestimmt. 14 Allele und 56 Phänotypen wurden bei insgesamt 311 nicht verwandten Individuen beobachtet. Eine Abweichung vom Hardy-Weinberg Gleichgewicht wurde nicht festgestellt. Die Heterozygotenrate betrug 81,35%. Der Polymorphismus YNZ22 ist mit einem CE-Wert von 70% und einem EM (Essen-Möller)-Wert von 9,35 (log) zur Vaterschaftsuntersuchung geeignet.

Schlüsselwörter VNTR · YNZ22 · D17S5 · Polymerase-Kettenreaktion (PCR) · Populationsgenetik · Vaterschaftsuntersuchung

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Introduction

The VNTR locus detected by probe YNZ22 (D17S5) on chromosome 17p13.3 (Nakamura et al. 1987) is considered to be a useful marker. The YNZ22 polymorphism has been studied using restriction enzymes MspI, TaqI, RsaI, PstI and HindIII. The MspI restriction pattern shows 10 alleles ranging in size from 0.5 to 1.3 kb (Nakamura et al. 1988). The polymorphism detected using the polymerase chain reaction (PCR), consists of 14 alleles, ranging in length from 170 bp (allele B1) to 1080 bp (allele B14) (Gécz 1991) with a 70 bp core sequence (Wolff et al. 1988).

Since the YNZ22 polymorphism and the p53 gene are both located on chromosome 17p, this polymorphism may be associated with several types of cancer (Singh et al. 1993; Makos et al. 1993).

Here we analyze a population sample from Catalonia (Spain) and discuss the suitability of this polymorphism for medico-legal purposes.

Materials and methods

Population studies. DNA samples were collected from 311 unrelated, healthy individuals (both sexes) from Catalonia (blood samples and hairs).

Family studies. 78 paternity testing cases were performed using YNZ22 locus.

Techniques. For PCR amplification of YNZ22 the method of Batanian et al. (1990) and Walsh et al. (1992), was used with some modifications. PCR reactions were carried out in 25 µl usually containing 10–15 ng of genomic DNA template (minimum 1 ng), 10 pmol of each primer (Horn et al. 1989), 200 µM each dNTP, 1 mM $MgCl_2$, 1.25 units of Taq polymerase and Mg-free buffer. Samples were processed through 34 cycles consisting of: 1 min at 95° C, 1 min at 60° C, and 3 min at 72° C with an auto-extension of a further 10 seconds every cycle.

Detection of the different alleles from PCR amplified products was carried out by ethidium bromide agarose gel electrophoresis. Alleles were scored following the terminology based on the number of repeats (Batanian et al. 1990).

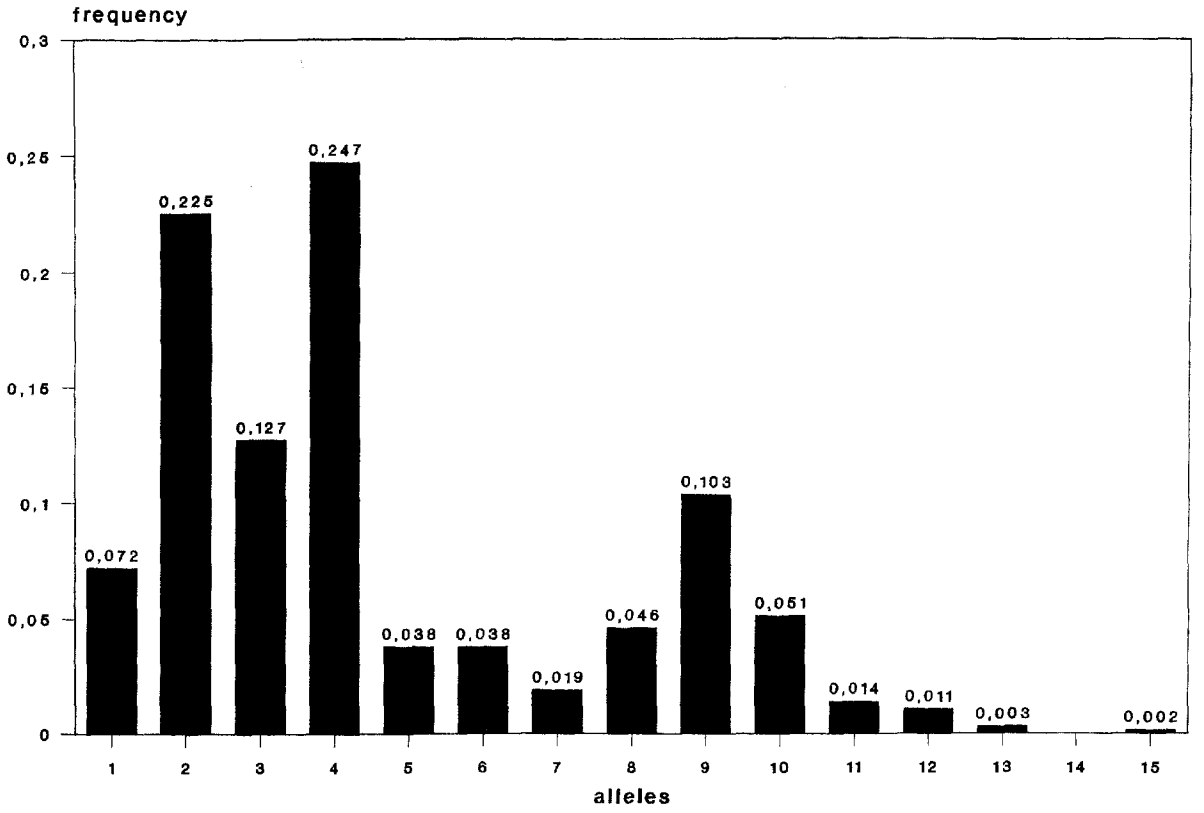

Fig. 1 Frequency distribution of the YNZ22 alleles in a population sample of 311 unrelated individuals

Results

The analysis demonstrated 56 phenotypes, which represent the products of 14 alleles. Allele frequencies are shown in Fig. 1. The distribution of the phenotypes is in Hardy-Weinberg equilibrium ($\chi^2 = 24.15$, df = 15, $0.1 > P > 0.05$). The Hardy-Weinberg formulation was calculated by comparing observed and expected phenotypes; all classes with less than 4 events were pooled (Budowle et al. 1991). The observed heterozygosity was 81.35%.

The YNZ22 polymorphism was also examined in 78 paternity testing cases. In 58 cases of "paternity practically proven" using other polymorphisms, the familial relationship was confirmed, supporting the assumed autosomal codominant mode of inheritance. In 20 cases of nonpaternity 13 incompatibilities were found (65%) supporting the "theoretical a priori chance exclusion value" CE = 70%, obtained from gene frequencies. The "Essen-Möller value" EM was 9.3548.

Discussion

To improve the amplification of large alleles relative to the small ones, we included a slight modification in the PCR amplification protocol and obtained better results on band intensity (Fig. 2).

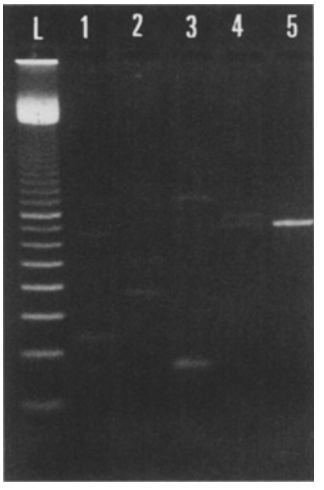

Fig. 2 YNZ22 amplification products of different individuals. Visualisation in ethidium bromide agarose gel electrophoresis. Allele designation corresponds to the number of 70 bp repeat units. The YNZ22 types from left to right are: 2–8, 4–6, 1–12, 9–10; 9. (L = 100 base-pair ladder)

We compared our results with the population data published so far which revealed that the Catalonia sample is very similar to that from Germany (Rand et al. 1992) and that from Italy (Tagliabracci et al. 1993), in which the frequencies for alleles 2, 3, 4, and 9 are very similar. Less similarity exists with the samples from USA (Batanian et al. 1990) and from Germany studied by Deka et al. (1992). Major differences were observed with other non-Caucasian populations (Deka et al. 1992). In view of these results and taking into account the scarcity of available data, it would be advisable to extend the study of this polymorphism to other population groups.

In the paternity testing field it can be inferred that YNZ22 is a suitable polymorphism, given the large num-

ber of alleles and their distribution. However caution should be exercised in analysing incompatibilities based on child-putative father homozygygotes (second order exclusion) since they could share a "rare" very long allele (24–25 repeats units), not amplified by PCR (Deka et al. 1992).

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