Allele frequency distribution of two VNTR polymorphisms (YNH24/ D2S44; Alpha globin 3'HVR/D16) in Italy

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INTRODUCTION

Variable number of tandem repeats (VNTRs) are a class of polymorphisms whose degree of variability is still unrivaled among human genetic markers. Forensic applications of VNTRs require detailed studies on the population genetics of these systems. Since most hypervariable profiles exhibit arrays of continuous Kb measures, a standardized protocol and a computerized repository of laboratory data are higly desirable in order to achieve reliable gene frequencies.
The creation of an Italian repository of VNTR frequencies is presently under way, under the auspices of the Italian working section (GEFI) of the Society of Forensic Hemogenetics. A computer algorithm for storage and management of continuous data has been developed (Pascali et al, 1991) and will be used for storage, retrieval and analysis of VNTRs data. Data are presently being collected by a a network of 12 Italian laboratories, which are expected to investigate individuals from several regions of Italy. Participating laboratories will follow a standard experimental procedure and share the same archive.
This project is presently being developed. Here we present a first collection of data generated in our laboratory following the model proposed to the collaborative group. These data are liable to represent the distribution of two common VNTRs (D2S44; alpha globin 3'HVR) in Central-Southern Italy and will be enlarged as long as results from other laboratories will be available.

MATERIALS AND METHODS

Genomic DNA was extracted, enzymatically restricted (HinfI) and analysed according to a standard protocol of Southern blot hybridization (Schneider et al, 1991).
Submarine gels for YNH24 were electrophoresed to meet the best resolution available in a range from 1 Kb to 5 Kb. Separate sets of experiments with shorter electrophoretic runs were required for 3'HVR, because of the high frequency of small fragments (down to 0.38 Kb) generated in this system.
Hybridization of the resulting filter blots was carried out

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with YNH24 (D2S44) (Wong et al, 1987) and alpha globin 3'HVR (D16) (Jarman et al, 1986) specific probes, under high stringency conditions. Length measures were assumed from the autoradiograms by a digitizing tablet. Predictions of Kb values were based on the reciprocal method by Elder and Southern (1987). Lengths and migrational distances of a commercial size marker (1kbladder, Boehringer) were used for measuring unknown sizes of bands.

RESULTS AND DISCUSSION

The distribution of the allelomorphic fragments in the two VNTR systems is shown in Fig.1 and Fig.2. The following features are worthwhile mentioning for each system.

YNH24/D2S44

Data on this system were drawn from 518 chromosomes, screened from 259 unrelated individuals and 83 families. Fragments spanning from 0.5 Kb to 7.84 Kb were identified, with frequencies arranged in a multimodal shape, and a major peak at 2.8 Kb. In this system, only 5 homozygous were detected. This leads to calculate a mean heterozygosity level of 98.5%. Segregation in families did not show any apparent mutation at this locus.

Alpha globin 3'HVR

We screened 360 chromosomes from unrelated individuals. A group of 64 families was also studied. Fragments had a major peak at 0.59 Kb and a plurimodal profile encompassing .38 Kb to 5.46 Kb. The average heterozygosity amounted to 93.3% (12 homozygous out of 180 individuals). No mutational events were seen in the family groups.

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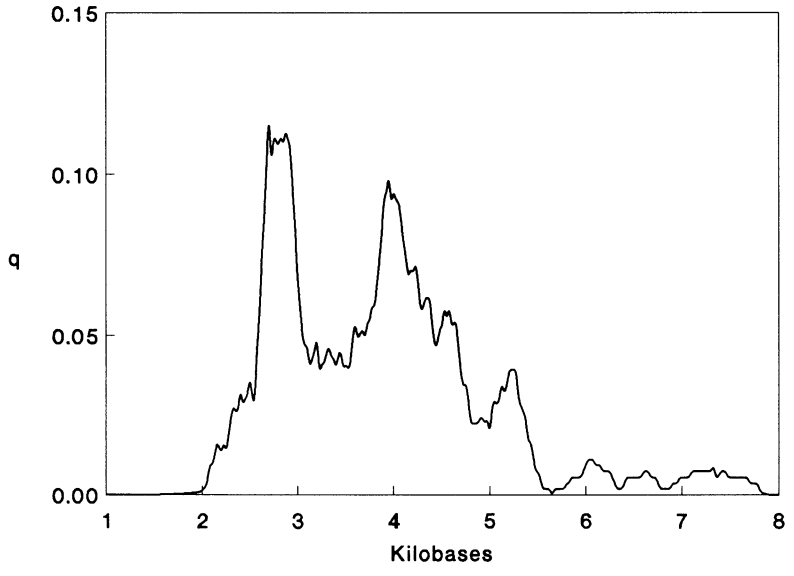

Fig 1. Allele frequency distribution of YNH24

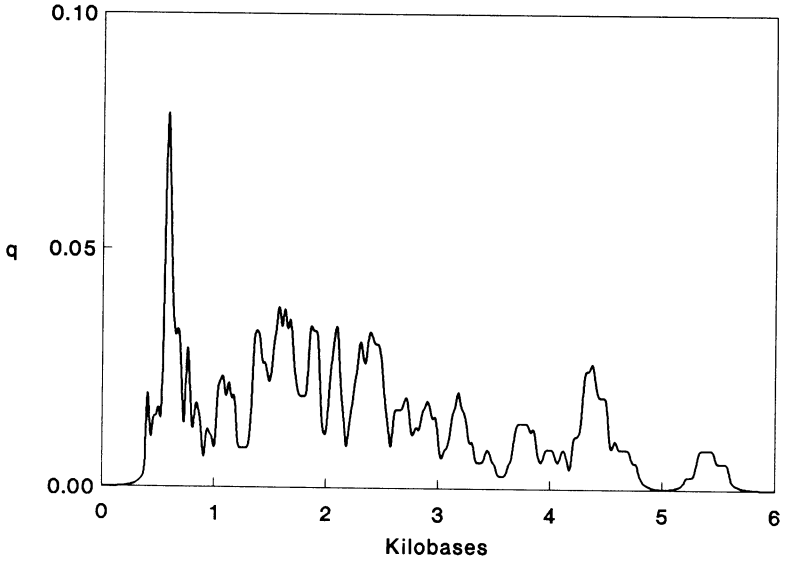

Fig 2. Allele frequency distribution of alpha globin 3'HVR