

Study of allele frequencies distribution of the 3'-HVR marker
near α -globin gene in the french population

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The human genome contains some hypervariable regions (HVR), resulting from various combinations, between individuals in populations, of short portions of DNA tandemly repeated. The 5'-HVR sequence is located at 8 kb, in the indicated sense, of the α -globin gene on the short arm of chromosome 16. The corresponding probe was used in France in some cases of paternity disputes (1). We report here a study concerning polymorphisms of these sequences in a French control population.

Material and methods: DNA were extracted from venous blood of a french collection of unrelated individuals, by digestion with proteinase K and phenol-chloroform extraction ; 10 μ l of DNA from each sample was restricted with PvuII. Restriction fragments obtained were separated on 1% agarose gel, and transferred on nitrocellulose filters. After labelling with ³²P, the probe obtained was hybridized with filters, and

the filters washed in stringent baths; allelic classes are determined by at least 100 pb of electrophoretic resolution.

Results: Figure 1 shows allelic repartitions in frequencies of the 62 alleles obtained from a sample of $n = 120$ unrelated individuals ($2n$ chromosomes = 240). The most frequent heterozygous genotype is at 2.9-2.4 kb, with a frequency = 0.033. Calculations show that these diverse alleles are in Hardy-Weinberg equilibrium. Heterozygosity is of 90%. The discriminating power (2) of the HVR probe is $D_p = 1 - \sum p_j^2$, j representing allelic frequencies of each genotype = $1 - 0.0276 = 0.973$, an excellent performance.

REFERENCES

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3'HVR/ Pvu II

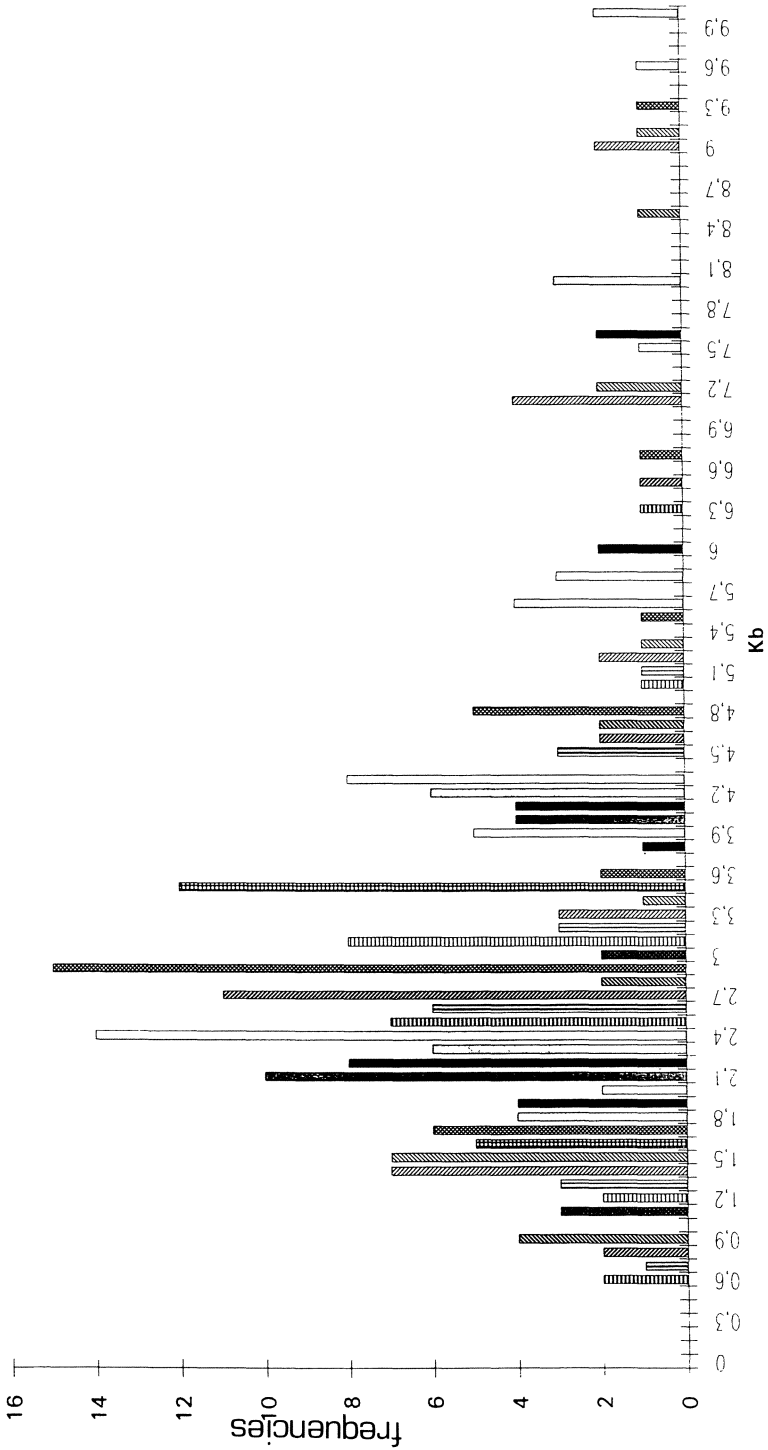


Figure 1. Allelic distribution in the population studied.