Identification and characterization of a hypervariable region [D18S27] on chromosome 18

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SOURCE DESCRIPTION: An 8 Kb genomic fragment was isolated from a partial EcoRI genomic library in Charon 30. This random clone was further subcloned as a 1.6 Kb HaeIII fragment into Bluescript, and was designated pAC 404.

POLVMORPHISM: When hybridized to PstI - digested genomic DNA, pAC 404 detected polymorphism at a single locus hypervariable region. The size of the alleles detected ranged from about 4 to 9 Kb. Analysis of allele frequency distribution was carried out in 615 unrelated individuals of four ethnic groups (i.e. Blacks, Caucasians, Hispanics, Chinese Orientals). Results show similar range in allele sizes with some differences in allele frequency distributions.

POLYMORPHIC WITH: PstI, HaeIII, HinfI, EcoRI.

<u>HETEROZYGOSITY:</u> With PstI, 86% heterozygosity was observed in 150 unrelated Caucasians.

<u>CHROMOSOMAL LOCALIZATION</u>: Chromosomal localization studies were done by hybridization to a panel of humanmouse cell hybrids. The results show that pAC 404 recognizes sequences located on chromosome 18.

MENDELIAN INHERITANCE: Co-dominant segregation was observed in 15 families of three generations.

PROBE AVAILABILITY: Available in future.



1 2 3 4 5

Fig. 1. PstI-digested genomic DNA from five unrelated individuals (lanes 1-5) were electrophoresed, transferred onto nylon filter, and hybridized to pAC 404 at 65° C. The filter was washed at 65° C, 0.1xSSC (15mM NaCl, 1.5mM sodium citrate), pH 7.0. The numbers on the left side of the figure are sizes in Kbp.