

Genetic Structure of Valencia (Spain) According to Alu Insertion Polymorphisms

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The Alu insertions are basically sequences of approximately 300 bp in length ancestrally originated from the 7SL RNA gene by retrotransposition. Alu insertion polymorphisms reflect both the maternal and paternal history of a population. The absence of the insertion gives the possibility of knowing the ancestral state, which constitutes an advantageous attribute in human evolutionary studies. Furthermore, alleles are identical by descent; consequently, it is highly unlikely that the same Alu insertion could occur more than once independently at the same locus. This fact means that polymorphic Alu insertions reflect unique evolutionary events. In addition, they are easily identifiable by Polymerase Chain Reaction (PCR) and agarose gels. In the present study, eight Alu insertions (TPA 25, PV92, APO, ACE, FXIIB, D1, A25 and B65) were typed in an autochthonous sample from Valencia province (Spain). Allele frequencies were estimated by direct counting and Hardy-Weinberg equilibrium was tested for each locus using the exact P method. The genetic information provided by the eight Alu loci analysed was stressed by means of Neighbour Joining and non-metric Multidimensional Scaling. Both methods were used to infer phylogenetic relationships at two different levels: i) among populations from the geographic scope of the Iberian Peninsula, ii) among Caucasoid populations independently of geographic origins.