SL801

The Y Alu Insertion Polymorphism in Korean Population

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The Alu family of short interspersed repeats(SINEs) is found in all primate species, with about 500,000 copies per haploid genome present in humans that have accumulated in their genomes via retrotransposition from a few master gene. Some of the 500,000 Alu element in the human genome have arisen so recently that not all chromosomes carry an Alu element at a particular locus. Once an Alu element insertion occurs, it appears not to be subject to loss or rearrangement, and the probability of independent insertion at the exact same position is negligible(Deininger, 1989). Thus, Alu insertion polymorphisms at specific loci have been shown to be valuable for human population studies. Based on the mapping data(Hammer, 1994), the Alu element, referred to as the Y Alu polymorphic (YAP) element, is present at a specific site on the non-recombining portion of the Y chromosome(Yq11) in some individuals and absent in others. The frequency of Y chromosomes containing the YAP element(YAP') is highest in sub-Saharan African population, followed by North African and European populations (Hammer, 1994). Most Asian populations(India, China, Papua New Guinea) reported so far lack the YAP^{+} element. An exception to this pattern was the finding of the YAP* element in Japanese populations(42%)(Hammer and Horai, 1995). We have examined the Y Alu insertion polymorphism using a PCR based assaying method in Korean population. A total of 180 unrelated males were analyzed to raise the possibility of tracing paternal lineages and male-mediated gene flow between Korea and Japan. The frequency of YAP^* element was found to be 1.1%(2/180). We also cloned these two YAP^* elements derived from PCR products by ligating into PCRTM II and subsequently sequenced to determine the length of the 3'-terminal poly(A) region of the sequence. The first clone, designated p[YAP*]KB-1 was found to have 41 dA-nucleotides and the second one, p[YAP*]KK-1 appeared to have 50 dA-nucleotides. Nucleotide sequence comparison with the 3'-terminal poly(A) region in other YAP elements revealed that they seem to be belong to the type 3a which is frequently found in Japanese population. It might be suggested that these YAP^* chromosomes provide an explaination for the admixtures recently from Japan. Therefore, it is that a large infusion of YAP chromosomes entered Japan with the Yayoi migration starting 2,300 years ago from Korea and a very few YAP chromosome admixed into Korean population so recently from modern Japanese originated from Jamon people who lived in Japan more than 10,000 years ago.