

Role of Alu Element in Detecting Population Diversity

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ABSTRACT Indel polymorphisms are becoming increasingly useful markers for population genetic studies. These are stable mutational events that are unlikely to undergo reverse or convergent mutations, making them useful as markers for distinguishing chromosomal lineages identical by descent and for using in population diversity studies. There has been a great deal of interest to use retrotransposones like Alu element, which displays indel polymorphism, in the population diversity studies. The polymorphic Alu insertions are particularly useful for such studies because the probability of independent retroposition at the same exact chromosomal site is virtually zero. All loci carrying a particular Alu insertion are derived from a unique event and hence are identical by descent. Further the ancestral state for polymorphic Alu insertions is the absence of the insertion hence the direction of mutational change is the gain of the Alu element at a particular locus. Knowing the ancestral state and the direction of mutational change greatly facilitates the analysis of population relationship but is generally not possible for other types of loci. These features make Alu insertion polymorphisms more attractive than other autosomal classical markers for analyses of population history and structure. This review is focused on Alu element and its role in detecting population diversity with few empirical examples from around the world. The phylogenetic studies based on Alu polymorphisms on Indian populations has also been presented.