Assessing Human Genetic Diversity by *Alu* Insertion Polymorphisms

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Alu insertion polymorphisms constitute approximately 5-10% of the human genome and exist in copy numbers of 500,000 to 1,000,000 per haploid genome. Recently, *Alu* insertion polymorphisms have been utilized in evolutionary studies.

Some of the *Alu* insertions are shown to be human specific. Thus, they are useful in studies of human population history. In this study we have focused on assessing human genetic diversity by these unique genetic markers. In order to do that, we have analyzed eight human specific *Alu* insertion-deletion polymorphisms (PV92, TPA25, APO, FXIIIB, D1, A25, B65) in 16 populations from Africa, in 12 populations from Europe, in 5 populations from the Middle East, in 17 populations from Asia, 3 populations from Oceania and 4 populations from the Americas. A total of 2308 individuals were typed.

The data shows that the *Alu* insertion polymorphisms show higher genetic diversity in Africa compared to other geographical regions. An AMOVA analysis was performed in order to assess the amount of variation within populations, among populations in distinct geographical regions and among geographical regions. According to our data the vast majority (86%) of variation is due to variation among individuals in particular populations. Only about 2% of the variation is due to differences among populations within geographical regions, and about 12% of the variation is detected among geographical regions. This finding is in accordance with other studies utilizing other genetic markers. The population relationships were further studied by constructing phylogenetic trees from genetic distances bases on distribution of *Alu* insertions.