

Mobile Elements and Genomic Variation

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Abstract

Primate mobile elements (SINEs, LINEs and SVA elements) belong to discrete subfamilies that can be differentiated from one another by diagnostic nucleotide substitutions. An analysis of several recently integrated mobile element lineages was undertaken to assess mobile element associated primate genomic diversity. Our screening of the mobile elements resulted in the recovery of a number of "young" Alu, L1 and SVA elements with different distributions throughout the primate lineage. Many of the mobile elements recovered from the human genome were restricted to the human lineage, with some elements that were polymorphic for insertion presence/absence in diverse human populations. The distribution of Alu, L1 and SVA elements throughout various primate genomes makes them useful tools for resolving population genetic relationships and non-human primate phylogenetic relationships. We have also characterized the structural genomic variation associated with the insertion of recently integrated mobile elements in primate genomes along with post insertion recombination based events. These genomic deletions are yet another source of mobile element associated genetic variation within the primate lineage.