Analysis of the human Alu Ya-lineage

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The Alu Ya-lineage is a group of related, Short Interspersed Elements (SINEs) found in primates. This lineage includes subfamilies Ya1-Ya5, Ya5a2 and others. Some of these subfamilies are still actively mobilizing in the human genome. We have analyzed 2482 elements that reside in the human genome draft sequence and focused our analyses on the 2318 human autosomal Ya Alu. A total of 1470 autosomal loci were subjected to polymerase chain reaction (PCR)- based assays that allow analysis of individual Ya-lineage Alu elements. About 22% (313/1452) of the Ya-lineage Alu elements were polymorphic for the insertion presence/absence on human autosomes. Less than 0.01% (5/1452) of the Ya-lineage loci analyzed displayed insertions in orthologous loci in non-human primate genomes. DNA sequence analysis of the orthologous inserts showed that the orthologous loci contained older pre-existing Y, Sc or Sq Alu subfamily elements that were the result of parallel forward insertions or involved in gene conversion events in the human lineage. This study is the largest analysis of a group of "young", evolutionarily-related human subfamilies. The size, evolutionary age and variable allele insertion frequencies of several of these subfamilies makes the Ya-lineage a useful tool for human population studies and primate phylogenetics.