

## GENOME WIDE ANALYSIS OF THE HUMAN ALU YB-LINEAGE

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The *Alu* Yb-lineage is a “young” largely human-specific group of SINE (short interspersed element) subfamilies that have integrated throughout the human genome. In this study, we have computationally analyzed the draft sequence of the human genome for *Alu* Yb-lineage subfamily members present on autosomal chromosomes. A total of 1733 Yb *Alu* subfamily members have integrated into human autosomes. The estimated average age of Yb-lineage subfamilies, Yb7, Yb8 and Yb9, was estimated as 4.81, 2.39 and 2.32 million years, respectively. In order to determine the contribution of the *Alu* Yb lineage to human genomic diversity, 1202 polymerase chain reaction (PCR)-based assays were performed using oligonucleotide primers that amplified the genomic regions containing individual Yb-lineage subfamily members. Approximately 20% of the *Alu* elements were polymorphic for insertion presence/absence in the human genome. Fewer than 0.5% of the Yb *Alus* were also present at orthologous positions in non-human primate genomes. Sequence analysis of these loci showed that each of the orthologous loci from non-human primate genomes contained Y, Sg and Sx *Alu* family members. Thus, these Yb *Alu* elements are thought to have integrated into the human genome quite recently and replaced the older *Alu* elements via one of three mechanisms: gene conversion, *Alu*-mediated deletion or parallel, independent insertion. The high copy number, level of insertion polymorphism, and estimated average age suggests that many members of the *Alu* Yb elements will be useful in a wide range of genetic analyses.