

COMPREHENSIVE ANALYSIS OF TWO *ALU* Yd SUBFAMILIES

**J. Xing¹, A. Salem^{1,2}, D.J. Hedges¹, G.E.Kilroy¹, W. S. Watkins³, J. E. Schienman⁴, C. Stewart⁴,
J. Jurka⁵, L. B. Jorde³ and M. A. Batzer¹**

¹*Department of Biological Sciences, Biological Computation and Visualization Center,
Louisiana State University, Baton Rouge, Louisiana*

²*Department of Anatomy, Faculty of Medicine, Suez Canal University, Ismailia, Egypt.*

³*Department of Human Genetics, University of Utah Health Sciences Center, Salt Lake
City, Utah*

⁴*Department of Biological Sciences, University at Albany, State University of New York,
Albany, New York*

⁵*Genetic Information Research Institute, Mountain View, CA*



Alu elements have inserted in the human genome throughout primate evolution. A small number of *Alu* insertions have occurred after the divergence of humans from non-human primates and therefore should not be present in non-human primate genomes. Most of these recently integrated *Alu* elements are contained with a series of discrete *Alu* subfamilies that are related to each other based upon diagnostic nucleotide substitutions. We have extracted members of the *Alu* Yd subfamily that are derivatives of the *Alu* Y subfamily that share a common 12 bp deletion that defines the Yd lineage from the draft sequence of the human genome. Analysis of the Yd *Alu* elements resulted in the recovery of two new *Alu* subfamilies, Yd3 and Yd6, which contain a total of 295 members (198 Yd3 and 97 Yd6). DNA sequence analysis of each of the *Alu* Yd subfamilies yielded age estimates of 8.74 and 1.21 million years old for the *Alu* Yd3 and Yd6 subfamilies respectively. Two hundred *Alu* Yd3 and Yd6 loci were screened using polymerase chain reaction (PCR) assays to determine their phylogenetic origin and associated levels of human genomic diversity. The *Alu* Yd3 subfamily appears to have started amplifying relatively early in primate evolution and continued propagating albeit at a low level as many of its members are found in a variety of hominoid genomes. Only two of the elements are polymorphic in the human genome and absent from the genomes of non-human primates. By contrast all of the members of the *Alu* Yd6 subfamily are restricted to the human genome, with 13% of the elements polymorphic for insertion presence/absence in diverse human genomes. A single *Alu* Yd6 locus contained an independent parallel forward insertion of a paralogous *Alu* Sq sequence in the owl monkey genome. These *Alu* subfamilies are a source of genomic fossil relics for the study of hominoid phylogenetics and human population genetics.