

## LINE-1 PRETa ELEMENTS IN THE HUMAN GENOME

**J. S. Myers\*, A-H. Salem\*, A. C. Otieno\*, and M. A. Batzer**

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

\* Contributed equally to this work



The preTa subfamily of Long Interspersed Elements (LINEs) is characterized by a 3 base pair “ACG” sequence in the 3' untranslated region and contains approximately 378 members in the human genome. Here, we have extracted 342 L1 preTaelements from the draft human genomic sequence and screened individual elements using subfamily specific polymerase chain reaction (PCR) assays to determine their phylogenetic origin and levels of human genomic diversity associated with this subfamily. Twenty-nine percent of the elements amenable to complete sequence analysis were full-length (approximately 6 kb) and have apparently escaped any 5' truncation. Twenty-seven of these full-length elements have two intact open reading frames and may be capable of retrotransposition. Sequence analysis of the preTa L1 elements showed a low level of nucleotide divergence with an estimated age of 2.27 million years old suggesting that expansion of the L1 preTa subfamily occurred after the divergence of humans and African apes. A total of 197 preTaL1 elements were screened with PCR based assays to determine the level of human genomic variation associated with each element and the phylogenetic origin this subfamily. All of the preTaL1 elements analyzed by PCR were absent from the orthologous positions in non-human primate genomes with thirty (15%) of the preTaL1 elements being polymorphic with respect to insertion presence or absence in the human individuals tested. The polymorphic L1 preTa identified in this study provide useful identical by descent markers for human identification and the study of modern human evolution.