AMPLIFICATION DYNAMICS OF RECENTLY INTEGRATED L1 REPEATS

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The insertion of mobile elements into the genome represents a new class of nuclear markers for the study of human evolution. Long Interspersed Elements (LINEs) have amplified to a copy number of about 100,000 over the last 100 million years of mammalian evolution and comprise approximately 15% of the human genome. The majority of L1 elements within the human genome are 5' truncated copies of a few active L1 elements that are capable of retrotransposition. Some of the young L1 elements have inserted in the human genome so recently that populations are polymorphic for the presence of an L1 element at a particular chromosomal location. L1 insertion polymorphisms offer several advantages over other nuclear-based polymorphisms for human evolution studies. First, they are typed by rapid, simple, polymerase chain reaction (PCR) based assays. Second, they are stable polymorphisms that rarely undergo deletion. Third, the presence of an L1 element represents identity by descent, since the probability that two different young L1 repeats would integrate independently in the same chromosomal location is negligible. Fourth, the ancestral state of L1 insertion polymorphisms is known to be the absence of the L1 element, which can be used to root trees of population relationships. We have used computational biology to identify members of the L1 Ta subfamily and estimate that there are about 500 L1 Ta subfamily members in the human genome. We have also developed PCR based assays for the characterization of these elements. Phylogenetic analysis of nonhuman primate DNA samples showed that most of the recently integrated "young" Ta L1 elements were restricted to the human genome and absent from the genomes of nonhuman primates. Analysis of a diverse array of human populations showed that over half of the L1 Ta subfamily members are polymorphic. Polymorphic L1 elements represent a new source of identical by descent variation for the study of human evolution.