

RECENTLY INTEGRATED HUMAN ALU REPEATS: FINDING NEEDLES IN THE HAYSTACK

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Alu elements undergo amplification through retroposition and integration into new locations throughout primate genomes. Over 500,000 Alu elements reside in the human genome, making the identification of newly inserted Alu repeats the genomic equivalent of finding needles in the haystack. Here, we present two complementary methods for rapid detection of newly integrated Alu elements. The second method is based on an anchor-PCR technique we term Allele-Specific Alu PCR (ASAP). In this approach, Alu elements are selectively amplified from anchored DNA generating a display or “fingerprint” of recently integrated Alu elements. Alu insertion polymorphisms are then detected by comparison of the DNA fingerprints generated from different samples. Here, we demonstrate the utility of these methods by applying them to the identification of members of the smallest previously identified subfamily of Alu repeats in the human genome termed Ya8. This subfamily of Alu repeats is composed of about seventy elements within the human genome. Approximately fifty percent of the Ya8 Alu family members have inserted in the human genome so recently that they are polymorphic, making them useful markers for the study of human evolution.