INNULS, A NEW STRATEGY FOR HUMAN IDENTIFICATION BASED ON RETROTRANSPOSABLE ELEMENTS

Bobby L LaRue[1,*], Sudhir K. Sinha[2], Anne H. Montgomery[2], Robyn Thompson[2], Lauren Klaskala[2], Gina Pineda[2], Jianye Ge[1], Jonathan King[1], Meredith Turnbough[1], Bruce Budowle[1]
[1]Institute of Applied Genetics; Department of Forensic and Investigative Genetics; University of North Texas Health Science Center; 3500 Camp Bowie Blvd., Fort Worth, Texas 76107, USA
[2] InnoGenomics Technologies, LLC, 1441 Canal Street, New Orleans, LA 70112, USA

Retrotransposable elements (REs) consisting of long interspersed nuclear elements (LINEs) and short interspersed nuclear elements (SINEs) are a group of markers that can be useful for human identity testing. Until now however, due to the inherent size difference (up to 6kb in some instances) associated with insertion and null alleles (or INNULs), the use of REs for forensic human identification testing has not been practical. Using a novel primer strategy, referred to as Mini-Primers, the overall amplicon size and difference in allele amplicon size for INNULs in a single-tube reaction has been reduced to allow for the broad application of these markers to human identity testing. Of the 12 preliminary markers tested (both LINES and Alus), the amplicon sizes ranged between 50 and 179bp. The size differences between the amplicons for alleles of the same locus have been reduced to as little as 1bp in a multiplex reaction. In addition to developing a practical method for using SINEs for genotyping individuals, this is the first presentation demonstrating that LINEs can be a potential multiplex marker for human identification. When tested in 3 major North American and 4 Asian populations, the markers generally met Hardy-Weinberg expectations and there was little evidence of detectable levels of linkage disequilibrium among the markers. Some of the markers in the panel were shown to be better suited for individual identification and others would be more suited for ancestral studies. Additionally, we have multiplexed several of the loci (~10) into a prototype single-tube panel as a “proof of concept” for their utility in a forensic setting. This initial study has demonstrated that bi-allelic INNULs can be a viable alternative to INDELs and SNPs for genotyping of individuals and provide a sensitivity of detection that would make them useful for forensic applications. ⌘