

INDEL MARKERS FOR HUMAN IDENTIFICATION: VALIDATION OF THE INVESTIGATOR DIPPLEX HUMAN IDENTIFICATION KIT

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STR based markers are the current standard in DNA based human identity testing. While assays based on this technology work quite well for many situations, STRs have some limitations. Mainly the amplicon sizes limit analysis of degraded samples. Additionally, the nature of the polymorphisms themselves creates “stutter” artifacts during the PCR which can confound low copy number analyses, and STRs have relatively high mutation rates which impacts kinship analyses. Single-nucleotide polymorphisms (SNPs) are more amenable to analysis of degraded samples because the genetic variation can be contained within smaller amplicon sizes, and due to the nature of the variation SNPs have substantially lower mutation rates compared to STR based markers. A drawback of SNP-based markers is the requirement for complicated detection strategies to detect the different base states. Insertion/deletion (Indel) based identity markers, which can be considered a subclass of SNPs, have the positive features of SNPs and have the added attribute of having a discernible size difference between alleles. Thus alleles can be detected by electrophoretic separation in a similar fashion as is done for STR typing. A validation study was performed on the Investigator DIPplex kit (Qiagen, Hilden, Germany) which is a single-tube assay containing 30 stable Indels and an amelogenin marker. The Indel loci are distributed over 19 autosomes and are at least 10Mbp from any commercially available STR or SNP marker, allowing them to be used as an adjunct with other human identity marker kits. Using this assay we typed sample populations of African Americans, Asian Americans, Caucasians and Hispanics. The power of discrimination for the 30 Indels reaches 10^{-13} . Validation studies included those described by SWGDAM, such as limits of detection, mixtures and mock-casework analyses. Electrophoretic artifacts were detected and overcome. Based on these results, the Investigator DIPplex assay holds promise as an additional tool for human identity and kinship analyses.