DEVELOPMENT OF INSERTION-DELETION (INDEL) MARKER PANELS FOR ANCESTRAL AND INDIVIDUAL IDENTITY GENOTYPING

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Insertions-Deletions (INDELs) are a type of polymorphism where small sequences of DNA have been inserted or deleted in relation to a known consensus reference sequence. The differences between the alleles are based on amplicon size, rather than detecting a nucleotide substitution as used for Single Nucleotide Polymorphism (SNP) typing. These size differences can be easily resolved using capillary electrophoresis (CE) with traditional chemistries for assaying fragment length. Thus, no new instrumentation is required for INDEL analyses in standard forensic laboratories. Analytically, INDELs perform similar to that of STRs and can be multiplexed together to achieve a high power of discrimination. Due to the small size of the insertion fragment, amplicons can be designed much smaller than is possible with larger repeat motifs such as STRs. This makes them ideal for genotyping degraded DNA samples. The uses of these markers may be tailored towards human identity (HID) or as an ancestral informative marker (AIM) depending on the population statistic used for selection. In the present study an HID panel of 49 markers was selected for a multiplex PCR reaction. The alleles of these HID markers are evenly distributed in major populations, are in Hardy-Weinberg equilibrium, do not display Linkage disequilibrium, and exhibit relatively low population substructure. A second panel of AIM INDELs was selected from the public data generated by the 1000 Genomes project for separate multiplex assays based primarily on population specific alleles. Together these panels will increase the capacity to type degraded DNA for identity and ancestral purposes.