TWO NOVEL MULTIPLEX INDEL ASSAYS FOR DETERMINING ANCESTRAL AND INDIVIDUAL IDENTITY FOR USE WITH DEGRADED DNA SAMPLES

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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. Small amplicon markers are 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. Based on our previous work, we have selected 30 AIM INDELs and 49 HID INDELs and incorporated them into two separate panels for forensic genotyping. The AIM markers were selected from the public data generated by the 1000 Genomes project for separate multiplex assays based primarily on population specific alleles. The final markers in the panel were based not only on infomativeness, but also based on amenability to multiplex PCR. In a similar fashion, the panel of 49 HID markers was selected from a larger set of markers we have previously described with an emphasis not only on discriminatory power, but also on the capability of being included into a multiplex PCR reaction. Performance of the two multiplex assays was assessed. Together these panels will increase the capacity to type degraded DNA for identity and ancestral purposes.