ADVANCES OF MASSIVELY PARALLEL SEQUENCING FOR FORENSICS AND HUMAN IDENTIFICATION

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Analyses of Short Tandem Repeats (STRs) through Capillary Electrophoresis (CE) technologies are the traditional gold standard for forensics. However, with increasing global diversity, there is a demand for forensic applications that supply information for challenging sample types. For casework samples such as ancient remains, touch (low DNA copy number), mass disaster victims, and sexual assault cases, more information is needed for accurate human identification. Recent studies have demonstrated the use of Massively Parallel Sequencing (MPS) to interrogate simple di-allelic Single Nucleotide Polymorphisms (SNPs) as a new method for human identification. Thus, the HID-Ion AmpliSeq™ Identity and Ancestry Panels were developed to determine random match probabilities and infer biogeographical ancestries, respectively. CE STR designs were optimized for MPS, which detects STR sequence polymorphisms, thus increasing the discrimination potential of the widely used markers. Additional polymorphic STRs with sequence-based alleles, that can only be observed through sequencing, have been included. To innovate further, a set of microhaplotypes is currently being investigated to determine the number of contributors within a mixture sample, and potentially ancestry information. Microhaplotyes are loci of 3-7 SNPs that are in close genetic proximity, but are far enough to not be considered as Multi-Nucleotide Polymorphisms (MNPs). Microhaplotypes are multi-allelic in phase and have the low recombination rate of SNPs rather than the higher rate observed in STRs. Lastly, a Whole Mitochondrial (Mito) Genome Panel was developed to analyze samples where nuclear DNA is far too degraded to be quantified and amplified. The Mito genome is advantageous for determining maternal lineage in forensics, as well as studying metabolomics for the diagnostic and clinical world. The panels are designed through the Ion AmpliSeq™ pipeline and sequenced in a multiplex with STRs on the Ion S5™ Sequencers.