SNP ANALYSIS OF A FORENSICALLY INFORMATIVE STR AND SNP MULTIPLEX USING NEXT GENERATION SEQUENCING

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Biological stains submitted as evidence to a forensic laboratory are typically characterized using short tandem repeat (STR) profiles generated using a PCR-based kit, in conjunction with capillary electrophoresis (CE) analysis. When these profiles cannot be matched to a reference profile or a profile found in the Combined DNA Index System (CODIS), single nucleotide polymorphism (SNP) analysis may be used to further characterize the stain. SNPs can be used to predict the ancestry, lineage, and phenotype of a stain source, as well as add strength to the individual identity of a stain in a fashion similar to STRs. However, the sequencing of SNPs has historically been a laborious process, discouraging the use of this method in forensic laboratories. With next-generation sequencing (NGS) of hDNA, characterized by its massively parallel sequencing technologies, the process of SNP sequencing has been simplified. Illumina has developed a multiplex called ForenSeq DNA Signature Prep Kit that allows the sequencing of several ancestry, phenotype, and identity SNPs, in addition to several STR loci, on Illumina's MiSeq Personal Sequencer. This would allow forensic laboratories to continue with traditional STR profiling, while also generating additional information provided by informative SNPs. All of these developments are a great achievement, but without a robust and straightforward method for data analysis this NGS approach cannot be implemented for routine casework. This poster demonstrates that the MiSeq (Illumina), platform can successfully determine both STRs and autosomal SNPs of interest using the "pre-release" version of the ForenSeq DNA Signature Prep Kit (Illumina). SNPs and STRs were sequenced from generated amplicons consisting of a multiplex with 62 STRs (29 autosomal (including 13 CODIS loci), 9 X-STRs, and 24 Y-STRs) along with 176 SNPs (56 ancestry-informative, 24 phenotypic-informative, and 96 identity-informative). Results will be presented that demonstrate the ForenSeq DNA Signature Prep Kit yields reproducible results from amounts of DNA ranging from 1ng to 100 pgs. Additionally, there was concordance of SNP genotypes generated from the ForenSeq DNA Signature Prep Kit (Illumina) with genotypes generated by 23andMe. Statistical analyses of the SNP genotypes were conducted using both Illumina analytical software and FROGdb (Yale) for identity, ancestry and phenotype predictions.