

GENETIC PROFILING USING THE ILLUMINA® ForenSeq™ DNA SIGNATURE PREP KIT ON THE MiSeq DESKTOP SEQUENCER

Sarah E. Schmedes¹, Jennifer Churchill¹, Jonathan King¹, and Bruce Budowle^{1,2}

¹Institute of Applied Genetics, Department of Molecular and Medical Genetics, University of North Texas Health Science Center

²Center of Excellence in Genomic Medicine Research (CEGMR), King Abdulaziz University

Massively parallel sequencing (MPS) provides the capability to produce gigabases of sequence data for hundreds of samples in one run, at a dramatically reduced cost per nucleotide than that of traditional Sanger sequencing. Current methods used in forensic laboratories primarily are multiplex short tandem repeat (STR) amplification assays coupled with capillary electrophoresis to genotype, typically, 13-27 loci per individual/sample. However, the high throughput achieved by MPS makes it possible to sequence hundreds to thousands of genetic markers per individual, while multiplexing multiple individuals per run. The Illumina® ForenSeq™ DNA Signature Prep Kit was used to sequence a large battery of genetic markers, including 29 autosomal STRs, 24 Y-STRs, 9 X-STRs, 86 identity-informative single nucleotide polymorphisms (SNPs), 56 ancestry-informative SNPs, and 22 phenotypic-informative SNPs from reference and casework-type samples on the MiSeq Desktop Sequencer (Illumina). Sensitivity, reproducibility, mixtures, concordance with previous methods and casework-type samples were evaluated. This all-inclusive forensic typing panel provides higher discriminatory power while still maintaining backwards compatibility with current databases. In addition, typing of many genetic markers in an effective manner can permit database expansion which may increase the number of investigative leads that can be developed. The ForenSeq™ DNA Signature Prep Kit provides a fast, easy-to-use, DNA preparation method with minimal DNA input requirements for reference and casework-type samples.