

A 125-PLEX MICROHAPLOTYPE ASSAY BASED ON NEXT GENERATION SEQUENCING DEVELOPED FOR FORENSIC APPLICATIONS

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Microhaplotypes, defined as the combination of >2 SNPs within a segment of DNA up to 200 base pairs, are a new type of molecular markers in the field of forensic genetics. Microhaplotype possesses the advantages of both STR and SNP markers and has important application prospects in forensics including mixed DNA genotyping, ancestry inference and complicated kinship analysis. Next generation sequencing, capable of genotyping microhaplotype markers and directly yielding the phase, has been expected to become the major method for microhaplotype detection. Here we developed an in-house multiplex assay of 125 microhaplotypes based on next generation sequencing. The Illumina Truseq strategy was employed for library preparation, and the 125 microhaplotype markers were simultaneously sequenced on a Miseq machine. FastQ Files were exported and a software was developed for subsequent data analysis, genotype reporting and plotting. Over 300 Chinese Hans were sequenced using the 125-plex assay. Primers sequences and amplicon sizes of the assay were reported. Detailed sequencing information and population data were presented in the hope of providing a reference for related research and applications.