EFFICIENCY AND ACCURACY OF THE AFFYMETRIX GENECHIP® HUMAN MITOCHONDRIAL RESEQUENCING ARRAY 2.0 AS COMPARED TO STANDARD SEQUENCING

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Although the hypervariable regions of human mitochondrial DNA (mtDNA) have the highest number of polymorphic sites per given length of sequence in the mtgenome, there are also a number of SNP sites found throughout the coding region that are valuable for identification. Analyzing all of the coding region SNPs would undoubtedly increase the power of discrimination that could be obtained using mtDNA. The Affymetrix Resequencing MitoChip 2.0® is a relatively quick, yet still cost-effective method of sequencing the entire mtgenome in one reaction. To investigate the efficiency and accuracy of microarray resequencing, ten DNA samples of various ethnic backgrounds and geographic origins were selected to compare standard sequencing results to the results of microarray resequencing. Initial analysis involved only the HV regions, and the microarray was able to achieve a no-call rate of approximately 2.8% and a miscall rate of 0.13% when compared to standard HV1/HV2 sequencing. Additionally, both the no-call and miscall rates were comprised mostly of two unusual haplotypes in the group of ten. With optimization and validation, the Affymetrix Resequencing MitoChip 2.0® could be a very useful tool in forensic DNA identification.