THE USE OF MASSIVELY PARALLEL SEQUENCING (MPS) TO ACCURATELY AND RAPIDLY SEQUENCE THE mtGENOME OF 283 INDIVIDUALS FROM 3 NORTH AMERICAN POPULATIONS

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Due to its high copy number per cell, mitochondrial DNA (mtDNA) can be used when trying to type biological samples that contain degraded DNA such as hair or human remains. Additionally, due to its inheritance patterns, mtDNA can be used to determine matrilineal relationships. Mitochondrial DNA typing in forensic genetics has been performed traditionally using Sanger-type sequencing. Consequently sequencing of a relatively-large target such as the entire mitochondrial genome (mtGenome) is laborious, time consuming, and impractical. Thus, sequencing typically focuses on the control region due to its high concentration of variation. Massively parallel sequencing (MPS) has become less expensive and more user friendly allowing for high-throughput processing of large target areas. In this study, Nextera XT DNA Sample Preparation Kit and the Illumina MiSeq™ were employed to generate high quality whole genome mitochondrial haplotypes from 283 individuals (African American, *n* = 87; Caucasian, *n* = 83; Southwest Hispanic, *n* = 113) in a cost-effective and rapid manner. Results showed that haplotypes can be generated generally at a high depth of coverage with limited strand bias. The distribution of variants across the mitochondrial genome demonstrated greater variation within the coding region than the non-coding region. Haplotype and haplogroup diversity were described with respect to whole mtGenome and HVI/HVII. An overall increase in haplotype or genetic diversity and random match probability and more accurate haplogroup assignment demonstrate that MPS of the mtGenome using the Illumina MiSeq system is a viable and reliable methodology.