

Sequence Polymorphism and Haplogroup Distribution of Mitochondrial DNA Control Regions HVS1 and HVS2 in a Southwestern Nigerian Population

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Abstract : The human mitochondrial DNA (mtDNA) is about 17 kbp circular DNA fragments found within the mitochondria together with smaller fragments of 1200 bp known as the control region. Knowledge of variation within populations has been employed in forensic and molecular anthropology studies. The study was aimed at investigating the polymorphic nature of the two hypervariable segments (HVS) of the mtDNA, i.e., HVS1 and HVS2, and to determine the haplogroup distribution among individuals resident in Lagos, Southwestern Nigeria. Peripheral blood samples were obtained from sixty individuals who are not related maternally, followed by DNA extraction and amplification of the extracted DNA using primers specific for the regions under investigation. DNA amplicons were sequenced, and sequenced data were aligned and compared to the revised Cambridge Reference Sequence (rCRS) GenBank Accession number: NC_012920.1) using BioEdit software. Results obtained showed 61 and 52 polymorphic nucleotide positions for HVS1 and HVS2, respectively. While a total of three indels mutation were recorded for HVS1, there were seven for HVS2. Also, transition mutations predominate nucleotide change observed in the study. Genetic diversity (GD) values for HVS1 and HVS2 were estimated to be 84.21 and 90.4%, respectively, while random match probability was 0.17% for HVS1 and 0.89% for HVS2. The study also revealed mixed haplogroups specific to the African (L1-L3) and the Eurasians (U and H) lineages. New polymorphic sites obtained from the study are promising for human identification purposes.

Keywords : hypervariable region, indels, mitochondrial DNA, polymorphism, random match probability

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