ANALYSIS OF THE COMPLETE mtDNA IN BRAZILIAN SAMPLES: HAPLOTYPES DIFFERENTIATION AND NEW VARIANTS IDENTIFICATION IN NATIVE AMERICAN HAPLOGROUPS

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The use of mtDNA typing in most forensic laboratories is based on the polymorphisms present in the nucleotide sequence of the hypervariable region (control region), comparing the questioned sample with the reference sequence (rCRS) for the polymorphisms annotation. However, it has been reported that about 75% of the total mtDNA variation occurs outside the control region (CR) and that whole mtDNA sequencing would increase the discrimination power and the value of the generated data. This increase in the genetic diversity obtained when analyzing the whole mtDNA has been observed in different world populations [Ma et al. 2016] and can make more efficient the classification of haplogroups according to ethnic origin [Park et al. 2017]. Brazilian territory was occupied only by Native Americans until about the year 1500 and colonized by European navigators that brought African slaves generating one of the most miscegenated (substructured) populations of the world [Freitas et al. 2019]. Initial data about the population of the Center-West Brazil region for mtDNA CR were published in 2019 [Simão et al. 2019] and, to date, there is no published data for complete mtDNA using massively parallel sequencing (MPS) for any Brazilian population. In this work, 74 samples were selected from individuals of the west central region of Brazil (Midwest Brazil) who shared haplotypes by the mtDNA CR. The libraries were performed for all samples and 22 samples were already sequenced and analyzed by Variant Analyzer software (Illumina system). When analyzed only the CR, 8 different haplotypes were identified in these 22 samples. The complete mtDNA analysis allowed the identification of 16 haplotypes, 6 of which were shared by two samples each, that is, the diversity of haplotypes in those samples were increased. In addition, some genetic variants still not reported in the EMPOP were identified: 16182del; 16178del, 16192.1T, 14881A; 15382A, 15716A. Each one of these variations was identified separately in 6 different mtDNAs, 4 of which were classified in haplogroups A2 or B2 from Native American ancestral origin; the other 2 mtDNAs were classified into haplogroups with African ancestral origin (L2a1f and L0a2a2a). The haplogroup L2a1f has been reported to occur in African Americans [Behar et al. 2008]. It is possible that these identified variations are related to Native American haplogroups or maybe occur in the Americas, which should be confirmed by the analysis of more samples.