

STUDY OF THE Y CHROMOSOME SNPs IN POPULATION OF THE STATE OF ESPIRITO SANTO, BRAZIL

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Introduction: Human identification through DNA analysis uses the genetic profile of an individual studying a combination of markers inherited from their parents. The genetic markers most widely used in routine forensic are present in the autosomes, however, the markers present in sex chromosomes (X and Y) and mitochondrial DNA helps analysis efficiently. Thus, the Y chromosome markers have been widely studied because they have many applications in forensics and population field, being used two main categories: biallelic loci (single nucleotide polymorphisms - SNPs - and Alu insertion) and multiallelic loci (minisatellites and microsatellites - STRs). SNPs have several advantages over STRs, especially with degraded or in small quantities samples, due to its high frequency, simplicity, small size and low mutation rate.

Objectives: To identify the major haplogroups existing in the population of Espirito Santo and evaluate the contribution of Africans, Amerindians and Europeans in their formation, since this state has received immigrants from various origins.

Methodology: We studied 15 Y-SNPs in 155 blood samples from male individuals living in the State of Espirito Santo to classify the major haplogroups. Genotyping was performed by PCR followed by minisequencing reaction (Snapshot Multiplex) and detection by capillary electrophoresis on ABI3500 genetic analyzer (Applied Biosystems).

Results and Discussion: The analysis allowed the discrimination of nine different haplogroups, featuring 76.77% of European origin - haplogroups R1b1 (47.10%), I(xI2a2) (9.68%), G and J2 each with 7.10%, J(xJ1a, J2) (1.29%), R1(xR1a, R1b1) (3.23%) and I2a2 with 1.29% - 20.65% of African and/or Asian origin (haplogroup BCDE) and 2.58% of Amerindian and/or Asian origin (haplogroup P(xR1)). The haplogroup diversity was 71.87%. These data reveal a major European contribution, consistent with the standard mixture of Brazilian people, characterized by the admixture among men of European origin and women of African and Amerindian origin. These results will allow the expansion of the genetic data of the studied population in relation to these markers, which then could feed a database of the Brazilian population, in addition to the international, allowing its use in national forensic context.