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Mitochondrial variation in the Bahia–Brazil population

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Abstract. The variability of the mtDNA in 100 individuals from Bahia–Brazil was studied for the purpose of forensic investigation and population genetics. The mtDNA was extracted and amplified to obtain sequences of HVS-I and HVS-II. Amplified products were purified and sequenced, and the sequences were analysed. The differences between DNA sequences in comparison with the Cambridge Sequence Reference (CRS) are shown.

Keywords: Bahia–Brazil population; mtDNA; Population genetic data

1. Introduction

The analysis of the HVS-I, HVS-II and HVS-III regions of the mitochondrial D-Loop has been studied because of its copy number per cell and polymorphism, which are important for forensic investigation and population genetics. The mtDNA information is passed from mother to child allowing the maternal line to be traced. The mutation rates are higher than those observed in the nuclear DNA. The mtDNA inheritance pattern shows limitation in the discrimination power, although the distinct female lineage can be determined by different haplogroups. The Bahian population has become very diversified due to the inclusion of a wide range of ethnic backgrounds. In this study, a database of the Bahian population was established with HVS-I and HVS-II regions. The frequency of the nucleotide sequence variations and match probability were estimated.

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2. Material and methods

The mtDNA was extracted [1] from blood samples of 100 unrelated (in three generations) healthy donors from Bahia–Brazil and amplified to obtain sequences of HVS-I and HVS-II according to Ref. [2] using a 9600 thermal cycler. Amplified products were purified and sequenced with BigDye™ v.3.0 Terminator Cycle Sequencing Ready Reaction Kit and primers described by Ref. [2] on the 9600 thermal cycler. The sequences were analysed on the ABI Prism 377 automatic sequencer or by capillary electrophoresis on the ABI PRISM 3100 Avant Genetic Analyzer employing ABI software [3]. The mtDNA results are provided in a DNA sequence analysis format and the intra- and inter-sequence differences are determined in comparison with the Cambridge Reference Sequence (CRS) [4]. The sequence variation from the Cambridge Reference Sequence was established by calculating nucleotide diversity and gene diversity [5]. The nomenclature and interpretation of the mtDNA profiles were according to Refs. [6,7].

3. Results and discussion

Sequence polymorphisms from the CRS sited between 16024 and 16390 in HVS-I and from 73 to 374 in HVS-II show a total of 82 different sequences for HVS-I and 74 for HVS-II. Considering both regions, 94 different sequences were observed and from these 86 sequences were found once, 4 were found twice and 2 were found 3 times. Table 1 summarizes the mutation events found in the Bahian population.

As regards HVS-I, 92 polymorphism sites were identified, of which the most frequent were 16223 (C/T = 85%), 16278 (C/T = 44%), 16311 (T/C = 39%), 16189 (T/C = 34%), 16294 (C/T = 22%), 16187 (C/T = 21%), 16362 (T/C = 19%), 16129 (G/A = 17%), 16270 (C/T = 16%), 16126 (T/C = 15%), 16390 (G/A = 14%), 16264 (C/T = 14%) and 16327 (C/T = 13%). One C insertion was found at 16188.1 (insC = 2%), 16192.1 (insC = 2%) and 16285.1 (insC = 1%), and one T deletion at 16325 (delT = 2%). The positions with more than one difference were 16114 (A = 6% and T = 1%), 16258 (T = 1% and G = 1%), 16259 (A = 1% and T = 2%), 16265 (T = 1% and C = 2%), 16325 (delT = 2% and C = 7%) and 16343 (T = 3% and G = 1%).

Instead, for HVS-II, 52 polymorphism sites were identified, of which the most frequent were 263 (G = 99%), 73 (G = 96%), 152 (C = 52%), 195 (C = 43%), 150 (T = 30%), 182 (T = 26%), 146 (C = 22%), 247 (A = 21%), 198 (T = 19%) and 357 (G = 11%). Lastly, one C insertion was found at 309.1 (insC = 40%) and 315.1 (insC = 90%), one A insertion was found at 316.1 (insA = 9%), and two C insertions at 309.1 and 309.2 (insC = 9%). The positions with more than one difference were 185 (A = 6% and T = 12%) and 189 (C = 8%

Table 1
Sequence polymorphisms and gene diversity in the Bahian population ($n = 100$)

	Pyrimidine transitions	Purine transitions	Transversions	Insertions	Deletions	Nucleotide diversity	Sequence diversity
HVS-I	450	85	29	5	2	0.01103	0.9940
HVS-II	220	304	45	157	15	0.01341	0.9917
HVS-I + HVS-II	669	390	73	163	17	0.01222	0.9979

and G = 23%). In addition, a singular pattern was observed in HVS-II in which five samples shared three A deletions at positions 249, 290 and 291.

A total of 144 polymorphism sites (nucleotide positions) were identified for HVS-I and HVS-II regions.

In comparison with other populations [9–16], the Bahian population shows higher values for number of different sequences (82 in HVS-I and 74 in HVS-II), number of variable nucleotide positions (92 in HVS-I and 52 in HVS-II) and sequence diversity (99.40% in HVS-I and 99.01% in HVS-II).

Length polymorphism or length heteroplasmy [8] occurred in nine different individuals in HVS-I and HVS-II.

HVS-II qualifies as a suitable tool for population genetics because of its polymorphism content, but this region shows a lower variability between individuals in comparison to HVS-I.

In accordance to Ref. [17], we found a higher mutation rate in HVS-II compared with HVS-I and this can be useful for practical applications in forensic genetics.

However, the differences permit us to understand the evolutionary process, but, in some differences, it was not possible to determine which mutation event occurred. In this work, these differences were considered as nucleotide substitutions.

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