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HVI and HVII sequence polymorphisms of the human mtDNA in the North of Portugal: Population data and maternal lineages

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Abstract. We have analysed the sequence polymorphisms of the hypervariable segments I and II (HVI and HVII) of the human mitochondrial DNA (mtDNA) in the population of North of Portugal. The aim of this study was to create a population database for HVI and HVII regions and to analyse these two segments in maternal relatives. For the population data, the HVI and HVII segments were analysed in 142 unrelated and healthy individuals, chosen randomly, from the North of Portugal. For the maternal lineages analysis, these two segments were studied in 24 sets of families from that region of Portugal. The sequence polymorphisms of the HVI and HVII were determined by polymerase chain reaction (PCR) and direct sequencing.

Keywords: Mitochondrial DNA (mtDNA); HVI–HVII; North Portugal; Population data; Maternal lineages

1. Introduction

The control region of the human mtDNA is highly polymorphic due to a rapid rate of evolution. The mtDNA does not undergo recombination and is present in high copy number per cell. For this reason, its analysis is an important tool for genetic identification. The haploid maternal inheritance of mtDNA is very useful for the identification of maternal relatives. The analysis of mtDNA polymorphisms by direct sequencing of PCR products is of great importance in forensic casework because it has been applied successfully to trace and degraded samples. The aim of this work was to create a

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population database of the variability of HVI and HVII segments in the population of North of Portugal and to analyse some maternal lineages from this geographical area.

2. Materials and methods

Blood and/or oral swab samples were collected from 142 unrelated individuals and 24 sets of maternal relatives from the North of Portugal. DNA was extracted using two different standard methods. The HVI and HVII segments were amplified by PCR using specific primers [1]. The two segments were directly sequenced by capillary electrophoresis on both strands. Nucleotide substitutions and insertions/deletions were found by comparison with Anderson's reference sequence [2]. The genetic structure of the population was analysed. The match probability and discrimination power were calculated. The classification into haplogroups [3–5] was made considering the HVI segment and some positions of the HVII segment.

3. Results and conclusions

3.1. Population data

The HVI and HVII sequences of the mtDNA were analysed between positions 16 033–16 391 and 57–408, respectively. Table 1 presents the diversity and genetic parameters. Fig. 1 shows the distribution of haplogroups in the population of the North of Portugal. The most frequent nucleotide substitutions found were 263A>G (99.30%), 315.1C (97.89%), 73A>G (56.34%), 16 126T>C (22.54%) and 16 189T>C (15.49%). In the HVI segment, one deletion occurred on position 16 189. In HVII, there were 5 insertions (57.1C, 60.1T, 309.1C, 309.2C and 315.1C) and one deletion on position 310. Position heteroplasmy was observed in three individuals. Length heteroplasmy occurred in 27 individuals, being more frequent in the HVII segment. A total of 113 different haplotypes (HVI+HVII) were found in the 142 mtDNA sequences analysed. The sequence diversity observed in HVII is higher than the one observed in HVI (see Table 1). The most frequent haplotype (HVI+HVII) found in the North of Portugal was 263G 315.1C. The discrimination power, taken together both segments, is 0.9871. This means that the probability of finding the same haplotype in two individuals of different maternal lineages is 0.0129. The principal European haplogroups were found in the North of Portugal population, being the haplogroup H the most frequent (see Fig. 1).

Table 1
Diversity and genetic parameters in the population of North of Portugal

	HVI	HVII	HVI+HVII
Sample size	142	142	142
No. of haplotypes	84	70	113
No. of polymorphic positions	71	54	125
Transitions	67	49	116
Transversions	6	4	10
Insertions	0	5	5
Deletions	1	1	2
Nucleotide diversity	0.012817 ± 0.007007	0.010664 ± 0.005980	0.011745 ± 0.006054
Sequence diversity	0.9638 ± 0.0106	0.9745 ± 0.0051	0.9941 ± 0.0022
Mean number of pairwise differences	4.601439 ± 2.272475	3.796224 ± 1.923065	8.397662 ± 3.910071
Match probability	0.04294	0.03234	0.01290
Discrimination power	0.95706	0.96766	0.98710

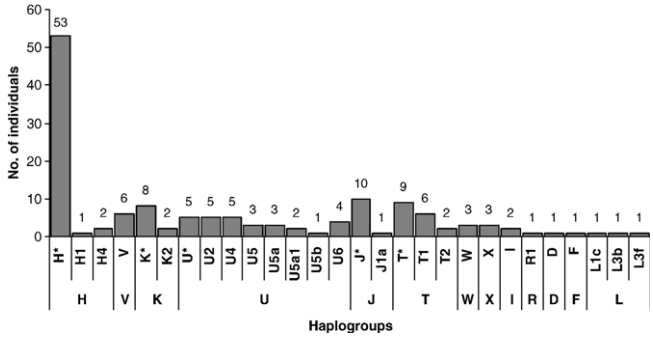

Fig. 1. mtDNA haplogroup distribution in the population of North of Portugal.

3.2. Maternal lineages

The sequences of HVI and HVII segments between positions 16033–16391 and 57–408, respectively, were analysed in 24 sets of maternal relatives (21 pairs mother/child, 1 set mother/two children, 1 pair grandmother/grandchild and 3 siblings). No haplotype differences were found between individuals belonging to the same maternal lineage. This suggests that no mutational events have occurred between the generations studied.

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