

Analysis of maternal and paternal lineages of the Azores Islands population

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Abstract. The aim of this study was the analysis of the origin of maternal lineage (mitochondrial DNA) and paternal lineage (Y Short Tandem Repeats) of Azores Islands population comparing our data with other populations from Europe and Africa. The comparison of maternal and paternal lineages from Azores Islands with other lineages from Europe and Africa was performed using the Arlequin software version 2.000. © 2006 Elsevier B.V. All rights reserved.

Keywords: Mitochondrial DNA; Y STR haplotype; Maternal and paternal lineage; Azores

1. Introduction

Historically, the Azores Islands, deserted at the time of their discovery, were populated mainly by the Portuguese, but French, Spaniards, Flemish, Jews, Moorish prisoners and African slaves also contributed to the initial settlement [1]. However, the specific contribution of each one of these settlers to the origin of the Azores Islands population was not clear, due to the lack of genetic analysis from both lineages.

To improve the genetic characterization of this population we typed the human mitochondrial DNA for HVI and HVII control regions and 17 STRs from the Y chromosome. The comparison of our data was made with the European (Central and North Portugal, NE Spain and SW Spain, France, Austria, Germany, England, Albania, Greece and Tunisia) and African (Mozambique, Angola, Guinea Bissau, Sao Tome and Prince and Cape Verde) [2–8] populations.

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Table 1
Mit DNA sequences (HVI and HVII) of Azores population

Population	Azores		
	HVI	HVII	HVI+HVII
<i>N</i> =64			
Number of different sequences	49	43	54
Number of different positions	58	28	86

2. Materials and methods

Polymorphisms of the two hypervariable segments (HVI and HVII) of mtDNA control region were analyzed in unrelated individuals from Azores Islands (*N*=64), using primers referred by Wilson et al. [9] in the PCR. Sequences were obtained using *ABI PRISM Cycle Sequencing Ready Reaction Kits*. Sequences were aligned and compared with the reference sequence using *SeqScape Software* (ABI version 2.0). Molecular diversity and analysis of variance were calculated in *Arlequin software 2.000*.

Y-chromosomal haplotypes were defined by 17 Y-STRs (DYS19, DYS385, DYS389 I, DYS389 II, DYS390, DYS391, DYS392, DYS393, DYS437, DYS438, DYS439, DYS460, DYS461, GATA A10, GATA C4 and GATA H4) in unrelated individuals of Azores Islands (*N*=62). The minimal haplotype was defined using *PowerPlexY PCR Amplification Kit* (Promega), with primers and amplification conditions described in manual; the other YSTRs were amplified with two tetraplex reactions (GEPY I and GEPY II), described by Sanchez-Diz et al. [10]. Haplotype diversity was calculated according to Nei (1987). Molecular Variance analysis (AMOVA) was performed with Markov Test, *Arlequin software 2.000*. Genetic distance matrix between populations was obtained by using the pairwise difference genetic distance (RST values), and DYS385 was not considered. The phylogenetic Neighbour-Joining tree was performed using the *TreeView software (1.5.2)*.

3. Results

3.1. Mitochondrial DNA

In the study of maternal lineages we found 54 different sequences for HVI and HVII regions in the population data from Azores and 50 are unique sequences. The other sequences were found in at least two individuals. From these 54 different sequences, 49 are defined for HVI region by 58 polymorphic positions and 43 for HVII region by 28 polymorphic positions (Table 1).

3.2. Y STRs chromosome

The analysis of haplotypes with 17 Y-STRs (DYS19, DYS389 I, DYS389 II, DYS390, DYS391, DYS392, DYS393, DYS385, DYS461, DYS437, DYS438, GATA C4, DYS460,

Table 2
Y haplotypes (17 YSTRs) of Azores population

Population	Azores		
	Number of different haplotypes	Number of unique haplotypes	Haplotype diversity
<i>N</i> =62			
Minimal haplotype	51	32	0.9797
Extended haplotype	57	53	0.9809

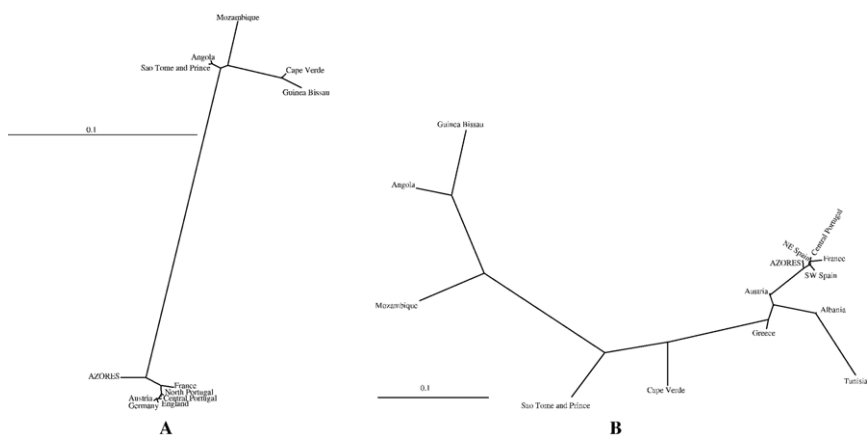


Fig. 1. Phylogenetic Neighbour-Joining trees of mtDNA (A) and Y-STRs (B) genetic distances between Azores Islands population and other European and African populations.

GATA A10, DYS439, and GATA H4) in population from Azores showed that the most common extended haplotype is (15, 14, 30, 23, 10, 11, 12, 14–14, 13, 15, 10, 23, 10, 17, 12, 27) and 57 different haplotypes were observed, 53 of them being unique. To minimal haplotype, a total of 51 different haplotypes were observed with 32 of them being unique. The combination of the minimal haplotype with the two tetraplexes reactions (GEPY I and GEPY II) reveals an increase of the haplotype diversity (0.9809) when compared only with minimal haplotype (0.9797) (Table 2).

4. Discussion and conclusions

The AMOVA analysis used to compare the maternal and paternal lineages of Azores Islands with European and African lineages revealed that the maternal lineage in Azores is more similar to European than to African lineage (Fig. 1A), which is in concordance with other preceding studies and the paternal lineage showed that the differences from the European and African lineages are not so clear, which means that the contribution of African slaves and Moorish prisoners was more relevant (Fig. 1B).

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