

Autosomal microsatellite analysis of the Azorean population

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Abstract. The knowledge of population history, demography and genetic structure has proven to be fundamental to address research in human genetics. Here, we describe the genetic diversity of the Azorean population and its affinity with other populations by the analysis of 13 microsatellite loci (TPOX, D3S1358, FGA, CSF1PO, D5S818, D7S820, D8S1179, TH01, vWA, D13S317, D16S539, D18S51 and D21S11) in 222 unrelated blood donors. These short tandem repeat (STR) markers were typed by Polymerase Chain Reaction (PCR) with fluorescently labelled primers. Statistical analysis was performed using Arlequin v.2.0, and Nei's genetic distance was calculated with DISPAN software and trees were constructed by Neighbor-Joining (NJ) using PHYLIP 3.63. To quantify the genetic contribution of Portuguese, African and European populations we calculated the admixture coefficient (mY) using Admix v. 2.0. The analysis of microsatellite loci shows that the Azorean population presents an average gene diversity of 0.776. For each marker, gene diversity ranges between 0.624 for TPOX and 0.882 for D18S51. Heterozygosity values calculated for each STR varies from 63.9% for TPOX to 89.2% for D18S51, although the majority of markers show values superior to 80%. In addition, the admixture coefficient reveals North Portuguese as the major contributor to the genetic background of the Azoreans. These results are corroborated by the dendrogram, in which Azores is closer to Belgians, Portuguese and Spanish, apart from Moroccans and Cabo Verdeans. Taken together, these data indicate that the gene pool of the Azorean population is very diverse and are consistent with our previous results on Y-chromosome. Moreover, no genetic differentiation between Azores and Portugal is observed. © 2005 Elsevier B.V. All rights reserved.

Keywords: Azores; Short tandem repeat; Diversity; Population genetics

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1. Introduction

Microsatellites, also named Short Tandem Repeats (STRs), have been particularly informative for human evolutionary inference. Their high variability and rapid mutation rate have produced sufficient allele frequency differences between population groups allowing phylogenetic inferences using relatively few loci [1]. Here, we present the autosomal genetic diversity of the Azorean population based on the analysis of 13 STRs.

2. Material and methods

The sample set comprised 222 Azorean unrelated blood donors, obtained from the anonymous DNA bank of São Miguel population, which was built according to the international ethical guidelines for sample collection, processing and storage [2].

Thirteen microsatellite loci (TPOX, D3S1358, FGA, CSF1PO, D5S818, D7S820, D8S1179, TH01, vWA, D13S317, D16S539, D18S51 and D21S11) were typed using fluorescently labelled primers described previously in Human Databases (STRBase, www.cstl.nist.gov/div831/strbase) and run on a CEQ™ 8000 Genetic Analysis System (Beckman Coulter).

Allele frequencies were calculated by direct counting; the Hardy-Weinberg equilibrium was assessed by an exact test provided by the Arlequin program. Heterozygosity and gene diversity values were also calculated with this software. F_{ST} related genetic distances were computed between pairs of populations by means of DISPAN and the distance matrix was used to construct a Neighbor-Joining (NJ) tree using PHYLIP 3.63. We used TreeView 1.6.6 to display tree phylogenies obtained from Neighbor-Joining. Population admixture was estimated by Admix2_0 software.

3. Results and discussion

The analysis of microsatellite loci shows that the Azorean population presents an average gene diversity of 0.776. For each marker, gene diversity ranges between 0.624 for TPOX and 0.882 for D18S51. Heterozygosity values calculated for each STR varies from 63.9% for TPOX to 89.2% for

Table 1
Heterozygosity, gene diversity and inbreeding coefficient for Azores

Azores	TPOX	D3S1358	FGA	CSF1PO	D5S818	D7S820	D8S1179
Heterozygosity	0.6390	0.7930	0.8190	0.7480	0.6580	0.7480	0.8150
HW (<i>p</i> value)	0.3130	0.2060	0.4000	0.2650	0.7510	0.0000	0.2550
Gene diversity	0.6240	0.7920	0.8560	0.7140	0.6950	0.8270	0.8160
Fis	-0.0327	-0.0006	0.0425	-0.0462	0.0546	0.0955	0.0009
	TH01	vWA	D13S317	D16S539	D18S51	D21S11	
Heterozygosity	0.8420	0.8010	0.7480	0.7380	0.8920	0.8060	
HW (<i>p</i> value)	0.1900	0.2590	0.1000	0.2780	0.0460	0.0002	
Gene diversity	0.8020	0.7960	0.7890	0.7660	0.8820	0.8420	
Fis	-0.0500	-0.0069	0.0526	0.0365	-0.0106	0.0422	
Av. gene diversity				0.7760			
Total Fis				0.0531			

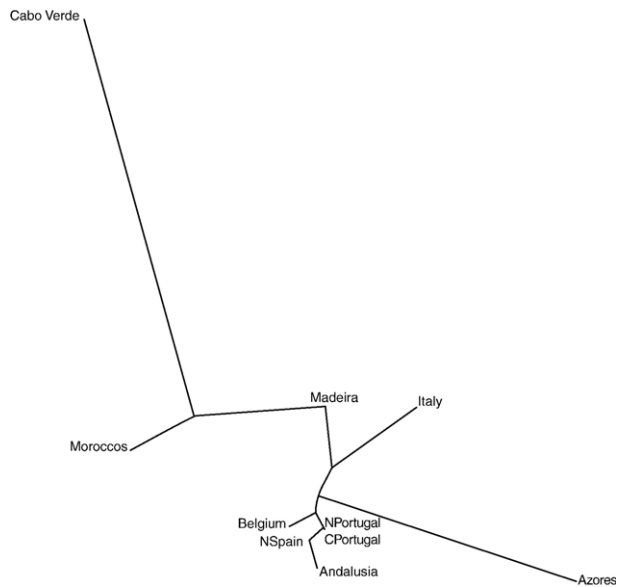


Fig. 1. Neighbor-joining tree based on Nei's genetic distances and constructed using information on 12 STRs.

D18S51, although the majority of markers show values superior to 80% (Table 1). In addition, the admixture coefficient reveals North Portuguese as the major contributor to the genetic background of the Azoreans. These results are corroborated by the dendrogram (Fig. 1), in which Azores is closer to Belgians, Portuguese and Spanish, apart from Moroccans and Cabo Verdeans.

Taken together, these data indicate that the gene pool of the Azorean population is very diverse and are consistent with our previous results on Y-chromosome [3]. Moreover, no genetic differentiation between Azores and Portugal is observed.

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