

Diversity of maternal and paternal lineages in the geographic extremes of the Azores (Santa Maria and Flores Islands): Insights from mtDNA, Y-chromosome and surname data

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Abstract. The mitochondrial DNA (mtDNA) and Y-chromosome (NRY) haplogroup composition, as well as the surname distribution in Santa Maria and Flores Islands (Azores Archipelago, Portugal), were analysed. Both mtDNA and NRY haplogroup distribution display significant differences between the two islands. For mtDNA, gene diversity was higher for Flores. NRY diversity derived from haplogroups, as well as the heteronymy values were statistically lower for Santa Maria. R_{st} values indicated reduced levels of microdifferentiation within both islands. Integration of previous results with those obtained in the present study further reinforced the fact that the different Azorean islands cannot be considered as a whole homogeneous population. © 2005 Elsevier B.V. All rights reserved.

Keywords: Azores Islands; mtDNA; NRY; Heteronymy; Demographic history

1. Introduction

Santa Maria (SMA) and Flores (FL) Islands occupy, respectively, the eastern and western limits of the Azores. Azores is formed by 9 islands, clustered in 3 geographical groups (Eastern, Central and Western). The peopling process was initiated in 1439 by SMA [1], proceeding gradually to the remaining islands and finishing in 1503 by FL [2]. Thus, SMA (57.6 inhabitants/km²) and FL (28.3 inhabitants/km²) [3] are not only

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chronologically distant in terms of settlement history but also represent the geographic extremes of the Archipelago, constituting interesting case studies in terms of comparative genetic structure. In this work, we analysed mtDNA and NRY haplogroup composition, as well as the surname distribution in both populations. The main goal was to investigate the influence that geographic location, as well as historical and demographical backgrounds have in the genetic structure of these two islands.

2. Materials and methods

Mitochondrial DNA (mtDNA) haplogroups were determined for 26 unrelated individuals born in SMA, following Santos et al. [4]. Previously published mtDNA data for 36 individuals born in FL were used [4]. Data on markers of the non-recombining region of the Y-chromosome (NRY), for 23 individuals born in SMA and 22 born in FL, were extracted from Montiel et al. [5]. Surnames of all individuals with more than 18 years of age, resident of SMA and FL Islands during the electoral census of 2004, were collected. Values of heteronymy and average within-group conditional kinship — R_{st} were calculated (data grouped by parish for R_{st}).

3. Results

The mtDNA and NRY haplogroup composition for SMA and FL are shown in Table 1. On what concerns mtDNA, the most frequent haplogroup in SMA is H (57.69%) whilst in FL it is V (33.33%). Relatively to the NRY composition, haplogroup R1(xR1b3f) presents the highest frequency in both islands, although values are markedly distinct (82.61% in SMA and 27.27% in FL). Both mtDNA and NRY haplogroup distribution display significant differences between the two populations ($p=0.001$ and $p=0.002$, respectively).

Nei's gene diversity (Table 2) of mtDNA is higher for FL, although the difference is not significant. Concerning the NRY, SMA exhibits a rather low haplogroup diversity (Table 2) compared to FL, as well as to the other Azorean islands [6]. The difference in diversity between SMA and FL is statistically significant (t -test, $p=0.0002$).

The values of R_{st} (Table 2) are low for both islands, indicating reduced levels of micro-differentiation within the two populations. Heteronymy values (Table 2) reveal a high level of

Table 1
mtDNA and NRY haplogroup (Hg) frequencies for Santa Maria and Flores

mtDNA Hg	SMA ($n=26$)	FL ($n=36$) ^a	NRY Hg	SMA ($n=23$) ^b	FL ($n=22$) ^b
H	57.69 (15)	22.22 (8)	E(xE3a)	4.35 (1)	22.73 (5)
V	3.85 (1)	33.33 (12)	F(xJ,K)	0.00 (0)	4.55 (1)
Pre*V	3.85 (1)	5.56 (2)	G	8.70 (2)	9.09 (2)
U	11.54 (3)	2.78 (1)	I(xI1b2)	0.00 (0)	4.55 (1)
U6b	11.54 (3)	5.56 (2)	I1b2	0.00 (0)	13.64 (3)
K	0.00 (0)	2.78 (1)	J	4.35 (1)	9.09 (2)
T	3.85 (1)	19.44 (7)	P*,Q	0.00 (0)	4.55 (1)
J	7.69 (2)	0.00 (0)	R1(xR1b3f)	82.61 (19)	27.27 (6)
M	0.00 (0)	5.56 (2)	R1b3f	0.00 (0)	4.55 (1)
X	0.00 (0)	2.78 (1)			

^a Data extracted from Santos et al. [4].

^b Data extracted from Montiel et al. [5].

Table 2

Nei's gene diversity (\hat{H}), average within-group conditional kinship (R_{st}) and heteronymy (Het) values for Santa Maria and Flores Islands

Population	mtDNA (\hat{H})	NRY (\hat{H})	Surnames	
			R_{st}	Het
Santa Maria	0.6554	0.3202	0.0026	0.9714
Flores	0.8127	0.8701	0.0027	0.9816

surname diversity in both populations. Nevertheless, in accordance with NRY results, heteronymy is significantly higher for FL Island (t -test, $p < 0.0001$).

4. Discussion and conclusions

In addition to a distinct mtDNA and NRY haplogroup composition, all the diversity indexes calculated are lower for SMA. The demographic history patterns, in terms of population size, denotes important differences for the two islands, which could have contributed for the results obtained in the present study. Previous mtDNA [4] and NRY results [5] had denoted the presence of a distinct haplogroup distribution in the Western group. Results now obtained also raise the possibility that the two islands of the Eastern group (S. Miguel and SMA) do not constitute a homogeneous cluster, with SMA presenting significantly lower diversity indexes than S. Miguel, for both mtDNA ($p = 0.0272$) and NRY ($p = 0.0242$). These results reinforce the idea that, from the haploid genome perspective, the Azores cannot be considered as an unstructured population, exhibiting not only inter but probably also intra-group heterogeneity. Thus, a representative sampling of each island is required to adequately describe the variability of the Azorean gene pool.

Acknowledgments

This work was supported by CIRN (POCTI/ARRA Açores/172). R.M. (SFRH/BPD/13256/2003) and C.S. (SFRH/BPD/20944/2004) are postdoctoral fellows of the FCT.

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