

Analysis of Y-chromosome and mtDNA variability in the Madeira Archipelago population

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Abstract. The Atlantic archipelago of Madeira is made up of two islands (Madeira and Porto Santo) with 250,000 inhabitants. These islands were discovered and settled by the Portuguese in the 15th century and played an important role in the complex Atlantic trade network in the following centuries. The genetic composition of the Madeira Islands' population was investigated by analyzing Y-chromosomal bi-allelic and STR markers in three different regions of the main island plus Porto Santo. We compared the results with mtDNA data and used the Y-chromosome STRs to determine the variability within each haplogroup. A sample of 142 unrelated males divided into four groups (Funchal City, West Madeira, North and East Madeira and Porto Santo) were analyzed. Significant genetic differences between these regions and the population of Funchal were found. The population of Funchal had lower gene diversity than expected. © 2006 Elsevier B.V. All rights reserved.

Keywords: Madeira Island; Y-chromosome; mtDNA

1. Introduction

The Madeira archipelago is made up of two inhabited islands, Madeira and Porto Santo, and has a population of about 250,000 inhabitants, with more than half living in Funchal. The Portuguese colonized the Madeira Archipelago in the 15th century and, in the beginning of the colonization, the archipelago was divided into three parts (Southwest and Northeast in the Madeira Island, and Porto Santo) and given to three administrators [1].

Many noblemen from Europe, namely Flanders, France and Italy came with the development of the sugar culture. Slaves from Guiné came to work the fields and, in the middle of the 15th century, they were more than 10% of the population. In the 16th

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century, sugar culture decreased and most of the slaves came to Funchal where they worked in people’s homes [2].

The aim of this work is to determine the genetic background of Madeira archipelago population; that is, if there are differences between the different parts of the islands and in which way these influences determined the genetic background of the present-day population of the archipelago.

2. Material and methods

A total of 142 DNA samples were used from unrelated males from Madeira archipelago whose ancestors were known to be originally from the same part of the archipelago, for at least three generations. The Y-chromosome haplogroups’ definition was done according to the YCC chromosome consortium [3] and we used the results of seven STRs from Y-chromosome [4] to see the variability within each haplogroup and to age the different haplogroups. We used the Arlequin software [5] to compare the four groups of Madeira archipelago and to calculate the Y-chromosome and mtDNA haplogroup diversity.

The age of STR variation within each haplogroup was estimated using mutation rate $w = 6.9 \times 10^{-4}$ per 25 years [6].

3. Results and conclusions

Funchal had the lower gene diversity to Y-chromosome and one of higher values to mtDNA (Fig. 1 and Table 1). The high percentage (almost 25% in Funchal) of African sub-Saharan haplotypes in mtDNA (like L and M1) can be explained by the fact that, through the 18th century, the number of illegitimate children represents about 50% of the newborn in Funchal [7].

A founder effect to the Y-chromosome haplogroup J2 in Porto Santo is suggested. There is a high frequency of Y-chromosome haplogroup J2 in Porto Santo (30%) but the age of J2 is 10.3 ± 2.8 kyr, $n = 7$, to the samples of Porto Santo against 21.8 ± 7.2 kyr,

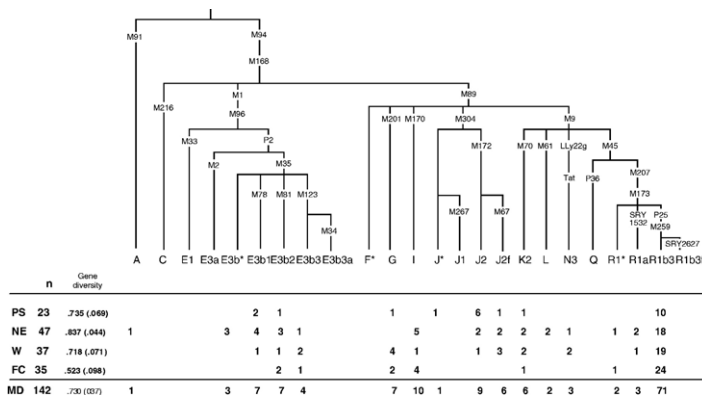


Fig. 1. Phylogenetic tree of the Y-chromosome haplogroups. Populations and gene diversity: Madeira archipelago (MD) $gd = 0.7275$ (0.0376), Porto Santo (PS) $gd = 0.7352$ (0.0685), Madeira Northeast (NE) $gd = 0.8227$ (0.0484), Madeira West (W) $gd = 0.7207$ (0.0741) and Funchal City (FC) $gd = 0.5227$ (0.0976).

Table 1

Frequencies (%) of the main mtDNA haplogroups [8] found in the Madeira archipelago (MD), Porto Santo (PS), Madeira Northeast (NE), Madeira West (W) and Funchal City (FC)

mtDNA haplogroup	PS (N=34)	NE (N=64)	W (N=39)	FC (N=16)	MD (N=153)
hH	47.2	45.3	20.5	19.0	36.6
hI		1.6		6.2	1.3
hJ	2.9	1.6	2.6	6.2	2.6
hK	14.7	3.1	5.1	6.2	6.5
hT	2.9	10.9	10.2		7.8
hL1		6.2	2.6	12.5	4.6
hL2		1.6	7.7		2.6
hL3	5.9	6.2	2.6	6.2	5.2
hM1		3.1		6.2	2.0
hPreV		3.1	2.6		2.0
hHV		3.1	2.6	12.5	3.3
hU/U6	14.7	9.4	17.9	25.0	14.4
hU6	2.9	1.6	7.7		3.3
hV	5.9	1.6	12.8		5.2
hX		1.6			0.7
hW	2.9		5.1		2.0
Haplogroup diversity	0.7469	0.7728	0.9015	0.9083	0.8279
S.E.	(0.0663)	(0.0496)	(0.0235)	(0.0479)	(0.0240)

$n=9$, to the other samples of Madeira archipelago. One cannot verify, using J2 haplogroup, the reflex of the multiple invasions of the island by North African pirates [1] because if this happened the frequency of haplogroup E3b2 (4.3%) would have been higher than this.

There are significant genetic differences ($P<0.05$) between the populations of Funchal and Porto Santo Island, and Funchal and the Northeast of Madeira, that could be the reflex of the differences in the colonization process of each part of the Madeira archipelago.

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