

Microgeographic substructure of Northern Portuguese mitochondrial DNA lineages: the female perspective of this region history

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Abstract. Mitochondrial DNA (mtDNA) lineages of 354 male samples from the five districts of Northern Portugal were defined. The aim was to obtain the female demographic history of this region and to compare it with the one obtained from Y chromosome haplotypes. The mtDNA landscape of North Portugal is in agreement with the one obtained from Y chromosome analysis, suggesting an East–West gradient in haplotype diversity. However, contrary to what happens with the Y chromosome, the easternmost district was the most diverse. Several explanations for this contrasting pattern are advanced. © 2003 Elsevier B.V. All rights reserved.

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1. Introduction

In a previous study of Y-chromosome STR markers in the five districts from Northern Portugal and two from Southern Galicia, we could detect signs of a slight microdifferentiation of the Easternmost district of North Portugal (Bragança), contrasting with the generally homogeneous genetic background of the region. In the present work, we analysed the mitochondrial DNA (mtDNA) variability in the same Northern Portuguese districts. The aim was to obtain the female genetic perspective of the North Portugal demographic history and to compare it with the one obtained with the Y chromosome haplotypes.

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2. Materials and methods

A total of 354 unrelated individuals from the five districts of Northern Portugal were analysed, distributed as follows: 117 from Porto, 69 from Braga, 68 from Viana do Castelo, 49 from Vila Real, and 51 from Bragança. HVS-I was amplified as described by Pereira et al. [1].

RFLP analysis of selected coding region positions was performed, whenever the assigned HVS-I sequence information alone to their corresponding haplogroups was insufficient. The typing was done with the use of standard protocols [2–5].

A stretch of 350 bp belonging to HVS-I segment (running from position 16050 to 16400; nomenclature according to Anderson et al. [6]) was analysed. Phylogenetic classification of the sequences was done according to Macaulay et al. [3], Richards et al. [4], Richards et al. [7], Quintana-Murci et al. [8], and Helgason et al. [9].

Haplotypes and haplogroups frequencies were determined by direct counting. Diversity indices, AMOVA, and pairwise F_{ST} values and associated p -values were calculated in Arlequin 2.0 [10].

Principal component (PC) analyses were performed using POPSTR (H. Harpending, personal communication) on the haplogroup composition (relative frequencies) of Northern Portuguese populations.

3. Results

Principal Component analysis (PC) was performed on the basis of the relative frequencies of mtDNA haplogroups (Fig. 1).

The first two PCs accounted for $\approx 70\%$ of the variance, from which 43.7% corresponds to the first one and displays an East–West gradient, separating the interior (Bragança and Vila Real) from the coastal districts (Porto, Braga and Viana do Castelo);

When AMOVA was computed without any hierarchical grouping, the amount of variation among populations was not significant (0.17%, $p=0.1896$). Performing

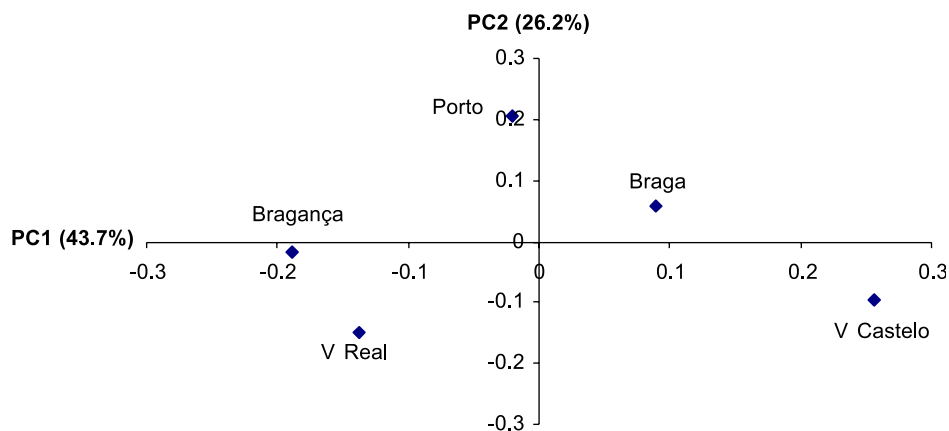


Fig. 1. Two-dimensional plot of the principal components of mtDNA haplogroup frequencies for Northern Portuguese samples.

AMOVA considering two groups, one composed by the easternmost districts and the other by the westernmost coastal districts, the percentage of variation among groups was slightly higher (0.18%, $p=0.1017$).

Nevertheless, pairwise F_{ST} values were significantly different when comparing Bragança and Viana do Castelo. Therefore, based on the pattern observed in the PC analysis as well as the pairwise F_{ST} values obtained (data not shown), AMOVA was repeated comparing Bragança with a single group composed by the other four districts. In this case, there was a two-fold increase in the proportion of genetic variation among groups (although statistically not significant). Other AMOVA designs considering one district (different from Bragança) against the others yielded lower among groups values (the next being 0.22% for Viana do Castelo).

Several diversity indices were calculated with HVS-I sequences for the five populations (data not shown). Bragança exhibited higher diversity values than the other districts regarding all diversity indices estimated. In contrast, Viana do Castelo, the most NW district is the one showing the most reduced diversity.

4. Discussion

The mtDNA landscape of North Portugal seems relatively homogenous, although suggesting an East–West gradient in haplotype diversity. This picture is in agreement with that obtained from Y chromosome analysis. However, in this case, contrary to what happens with the Y chromosome, the easternmost district is the most diverse.

Several (and not mutually exclusive) explanations for this contrasting pattern can be advanced: (a) different male/female effective population sizes; (b) differential migration patterns (coastal longer range migrations for males, shorter range continental movements for females); (c) different mating patterns and cultural practices between males and females. Since all the districts are closely related culturally and geographically, the first and the second options arise as the most plausible explanations.

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