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Microgeographic substructure of NW Iberian Y chromosome STR haplotypes

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Abstract. In order to assess the microgeographic heterogeneity of Y chromosome lineage's distribution in the north-western part of Iberian Peninsula, 409 male samples from the five districts of Northern Portugal and two districts of Southern Galicia were typed for 15 Y chromosome microsatellite markers. The results suggest that, in NW Iberia, barriers other than political were more important in shaping present Y chromosome lineage substructuring.

Keywords: Y-STRs; NW Iberia; Microgeography; Substructure

1. Introduction

Because of their abundant polymorphism, selective neutrality and ease of assessment, microsatellite loci seem to be ideal markers for population genetic monitoring in short evolutionary time scales, and thus many studies comparing closely related populations have been published [1,2]. The aim of this work was to assess the microgeographic heterogeneity of Y chromosome lineages in the North-Western part of Iberian Peninsula, through the analysis of 15 Y chromosome microsatellite markers in 409 male samples. The region analysed includes the Southern area of Galicia and the neighbouring Northern Portugal. The border between the two countries does not follow in many tracts important geographic barriers, but mountain ranges clearly differentiate interior and coastal parts of both political regions. In the framework of this scenario, the main goal of this research was to assess if geographical barriers present in the NW of Iberia were more important than political and linguistic ones for Y-STR substructuring.

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2. Materials and methods

Four-hundred nine blood samples were collected in the five districts of North Portugal (located to the North of Douro River) and two districts of Southern Galicia, from healthy unrelated individuals born in each area, after informed consent. The samples are distributed as follows: 23 from Montes Baixo Miño and 31 from Ourense, 157 from Porto, 54 from Braga, 60 from Viana do Castelo, 45 from Bragança and 39 from Vila Real.

Samples were typed with 15 Y-STRs (DYS19/389I/389II/390/391/392/393/437/438/439/460/461 and GATA A10/C4/H4) in two multiplex reactions as described by Beleza et al. [3].

RST with associated probability values and AMOVA were estimated using the software package ARLEQUIN 2.0 [4].

Microsatellite variance per locus and the average microsatellite variance across loci were calculated according to Goldstein et al. [5] for each population. For the estimation of DYS389II variance, alleles at this locus were considered excluding variation at DYS389I. A variance ratio was also estimated according to Jorde et al. [6], dividing the variance of each microsatellite in each population by the average variance of the microsatellite across the seven populations.

3. Results and discussion

It was possible to identify 378 different haplotypes from the typing of 409 non-related males from the NW of Iberian Peninsula with 15 Y-specific microsatellites, 20 of them being shared by different populations (4 haplotypes were shared by three of them and the remaining 16 by only two).

Analysis of molecular variance (AMOVA) was performed in different ways (Table 1). Considering a political approach, in which the two countries (Portugal and Galicia) were separated in two groups, the percentage of variation explained by differences among groups was slightly smaller than when considering no grouping, reflecting the common ancestry of Northern Portugal and Galicia.

RST pairwise comparison values (data not shown) were low, as it could be predicted from a microgeographic study of neighbouring populations, revealing the homogeneity existent between the samples, but showed a significant difference when comparing the

Table 1
Results from analysis of molecular variance (AMOVA)

Grouping	Source of variation	Variation (%)
No grouping	Among populations	0.29
	Within populations	99.71
Portugal vs. Galicia	Among groups	0.26
	Among populations within groups	0.22
	Within populations	99.52
Bragança vs. others	Among groups	1.07
	Among populations within groups	0.02
	Within populations	98.91

interior Northern Portuguese district Bragança with the coastal Northern Portuguese district Braga. Moreover, the highest pairwise RST values always involved Bragança and the other coastal districts.

In this context, performing AMOVA with Bragança versus all other samples together resulted in a ≈ 4-fold increase of the proportion of molecular variation observed between groups (Table 1).

Considerably high haplotype diversities were found in all populations studied (>99%). However, this standard diversity measure does not take variation in microsatellite repeat number (allele size) into consideration. Knowing that microsatellite variance of allele repeat size also offers a good measure of microsatellite diversity [5] a variance ratio was estimated [6] to remove the possible error arising from the contribution of highly mutating microsatellites. The average variance ratios across loci from Vila Real, Bragança and Montes Baixo Miño were significantly lower than the ones from Viana do Castelo, Braga and Ourense. The lower diversity found for Vila Real and Bragança (interior districts) may reflect some genetic drift imposed by important mountains ranges that separate these two districts from the other coastal districts. However, modern demographic migrations may have eroded the genetic substructure that could have existed in the past, and now it is only possible to observe some differentiation of Bragança in comparison to the other populations.

These results suggest anyway that, in NW Iberia, barriers other than political are more important in shaping Y chromosome lineage substructuring.

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