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Microgeographic mitochondrial DNA patterns in the South of Iberia

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Abstract. Along history, Andalusia (South of the Iberian Peninsula) has been a territory occupied by many civilizations coming from Europe and North Africa. Here, we aim to identify its mitochondrial composition by analyzing the two hypervariable regions (HVS-I and HVS-II) and selected coding region SNPs of the mitochondrial DNA (mtDNA). A total of 419 individuals from 28 villages (belonging to different provinces and with more than 200 years of history) have been sampled. This sampling has been designed in order to uniformly cover the geographic area of South Iberia. Historical record indicates that these villages have experienced little recent migration. Preliminary results revealed that 94% of the haplotypes belong to typical European haplogroups, 2.1% are sub-Saharan lineages and only 1.6% North African. AMOVA analysis indicates that the main percent (97.6%) of the variability in these populations is found between individuals, 2.2% between villages of the same province and 0.25% between provinces. In addition, haplotype diversity is high (0.99) in Andalusia in comparison with other Iberian and European populations. The results point to a lack of significant demographic impact (at least in the maternal mtDNA side) of North Africa despite the close geographic proximity and eight centuries or Arabian colonization. © 2005 Published by Elsevier B.V.

Keywords: Mitochondrial DNA; Microgeographic; SNP; South Iberia

1. Introduction

Along history, Andalusia (South of Iberia) has been a territory occupied by several civilizations coming from Europe and the North of Africa. Tartesos, Phoenicians, Romans,

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Visigoths and Arabs were among the main settlers. Historical records have revealed that eight centuries of Arabian colonization caused more a cultural than a demographic impact. In the present work, we analysed the mitochondrial DNA (mtDNA) variability in an autochthonous Andalusian population sample. The main aim of this study is to determine the phylogeographic composition of the mtDNA lineages in Andalusia and the level of North African and sub-Saharan contribution to this region. Our sampling strategy by provinces also will allow us to assess the level of regional population substructure, which could have a significant impact to the forensic application of the mtDNA test in routine casework.

2. Materials and methods

Saliva samples were collected from 419 autochthonous individuals of Andalusia, distributed as follows in 28 villages (belonging to different provinces): 15 individuals from each Cortegana, Rociana, Minas de Río Tinto, Guillena, El Coronil, Casariche, Herrera, El Carpio, Hornachuelos, Benamejí, Iznájar, Alcalá del Valle, Alcalá de los Gazules, Peal de Becerro, Arjona, Vilches, Castillo de Locubín, Villanueva del Trabuco, Villanueva de Algaidas, Pizarra, Dúrcal, Órgiva, Albuñol, Iznalloz, Carboneras, Macael and 14 individuals from Algodonales.

The criteria for sampling selection of the villages were: (i) more than 200 years of history, (ii) no significant recent immigration rate (certain degree of isolation) and (iii) stable population size (4000–8000 inhabitants).

DNA extraction, quantification, amplification and sequencing were made in agreement with standard protocols (provided under request). When it was not possible to assign haplogroups alone using the hypervariable regions one and two (HVS-I and HVS-II) sequence information [1], RFLP analyses of selected coding region positions was performed.

Haplotypes and haplogroups frequencies were determined by direct counting. AMOVA and pairwise F_{st} values were calculated using Arlequin 2.0 [9] and DnaSp software.

3. Results and discussion

A total of 283 different haplotypes were observed in 419 individuals belonging to the autochthonous Andalusian population. This corresponds with a haplotype diversity value of 0.990, a value that is higher than the one found in other European and Iberian populations [2–5,8]. MtDNAs sequences were classified in 43 different haplogroups or

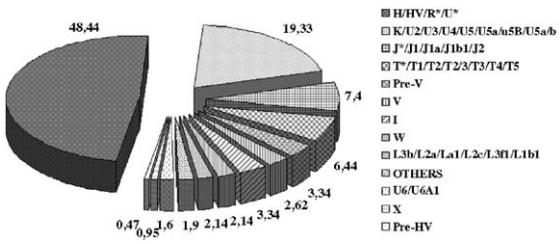

Fig. 1. General distribution of haplogroup frequency in Andalusia (values are expressed in percentage). The term "OTHER" includes haplogroups D, N*, N1a, N1b and M1.

Table 1
Results of analyses of molecular variance (AMOVA) in the HVS-I and HVS-II regions

	Source variation	Variation (%)
HVS-I	Among individuals from the same village	97.49
	Among villages within the same province	2.28
	Among provinces	0.23
HVS-II	Among individuals from the same village	97.64
	Among villages within the same province	2.08
	Among provinces	0.28
HVS-I and HVS-II	Among individuals from the same village	97.56
	Among villages within the same province	2.19
	Among provinces	0.25

lineages, an haplogroup pattern that does not show significant differences with regard to other European populations (Fig. 1).

With regard to the main geographical (continental or subcontinental) origin of the haplogroups, 94.36% of the haplotypes were typically European [6,7,10,11], 2.10% sub-Saharan lineages and 1.60% North African lineages. It is interesting to note that the frequency of maternal lineages coming from the North of Africa is low in spite of the long Moslem presence in the South of the Iberian Peninsula. Our data agree with historical documentation in that the Islamic invasion was of low demographic impact. Sub-Saharan lineages have also been detected at low frequency in Andalusia. Although an ancient introduction cannot be discarded, it is more probable that they originated from more recent genetic flow from the North of Africa or during the slave trade period.

The results of the analysis of molecular variance (AMOVA) show a higher variability among individuals (97.56%) than among provinces (0.25%) (Table 1). Therefore, to our level of resolution, Andalusia behaves like a homogeneous population.

References

[1] Anderson, et al., Nature 290 (1981) 457–465.
[2] Beleza, et al., International Congress Series 1261 (2004) 386–388.
[3] Bertranpetit, Cavalli-Sforza, Am. J. Human Genet. 55 (1991) 51–67.
[4] Comas, et al., Hum. Genet. 99 (4) (1997) 443–449.
[5] Macauly, et al., Am. J. Hum. Genet. 64 (1999) 232–249.
[6] Richards, et al., Am. J. Hum. Genet. (2000) 1251–1276.
[7] Richards, et al., Am. J. Hum. Genet. 71 (5) (2002) 1168–1174.
[8] Salas, et al., Eur. J. Hum. Genet. 6 (1998) 365–375.
[9] Schneider, et al., Arlequin, Genetics and Biometry Lab, 2000.
[10] Torroni, et al., Am. J. Hum. Genet. 55 (1994) 760–776.
[11] Torroni, et al., Genetics 144 (1996) 1835–1850.