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Haplotype distribution of the mitochondrial control region in the native population of the Canary Islands

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Abstract. The hypervariable regions HV1 and HV2 of mitochondrial DNA (mtDNA) were sequenced in 164 native unrelated individuals from Canary Islands. A total of 116 different haplotypes were observed. Genetic diversity and discrimination power obtained were 0.9916 and 0.9856, respectively. Some haplogroups have been also identified from the HV1 data. The analysis of HV1 sequences of 56 populations allows us to locate the Canary Islands in the European/North African mitochondrial genetic landscape. © 2005 Published by Elsevier B.V.

Keywords: mtDNA haplotype; Canary Islands; Sequencing; Control region; mtDNA phylogeny

1. Introduction

The mitochondrial DNA analysis represents a powerful tool for both forensic and bioanthropology studies. For forensic purposes, the statistical interpretation of the results obtained in the casework depends on the population frequency of the particular sequences. Thus, it is essential to establish a mitochondrial reference database, which would be also helpful to ascertain some key aspects of the genetic composition and origin of the studied population.

The aim of this study was to analyse the hypervariable regions of the mitochondrial DNA (HV1 and HV2) in a total of 164 unrelated native individuals from the Canary

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Table 1
Summary of haplotypes frequencies

Unique haplotypes		91
Haplotypes repeated	2 times	18
	3 times	2
	4 times	1
	5 times	2
	7 times	1
	10 times	1
Total		116

Islands, and to calculate the statistical parameters needed for the genetic forensic practice.

2. Materials and methods

A total of 164 samples (blood or saliva) from unrelated individuals were studied. A consent form with ancestry data was voluntarily filled and signed by every individual, with at least two or three maternal ancestors born in Canary Islands.

DNA was isolated by a common organic method (proteinase-K digestion and phenol-chloroform separation), followed by purification with ultra filtration (Microcon-YM100®). Then, both HV1 and HV2 were simultaneously amplified (in one reaction) using the primer set described by Wilson et al. [1], and standard PCR reagents and conditions (Applied Biosystems). Amplified products were purified by Sephadex, and sequenced on both strands using the dRhodamine Dye Terminator Sequencing kit (Applied Biosystems). The analysis of the extension products was performed on an ABI Prism 310. Sequences were edited by the Sequencing Analysis software (Applied Biosystems) and the comparisons were performed with the BioEdit shareware (kindly provided by Prof. Tom Hall). Genetic distances between all populations were estimated as linearized F_{ST} statistics [2] by using the ARLEQUIN program version 2.000.

3. Results and discussion

Complete HV1 and HV2 region sequences were obtained for 164 individuals (data available upon request) and were aligned with the Cambridge Reference Sequence [3]. A total of 116 different

Table 2

HV1 and HV2 sequences data	
Number of different haplotypes	116
Number of polymorphic sites	115
Number of observed transitions	106
Number of observed transversions	12
Number of observed sites with indels	4
Parameters of forensic interest	
Genetic diversity	0.9916
Matching probability	0.0144
Discrimination power	0.9856

Table 3
Haplogroups identified from the HV1 data

Haplogroup	Number of individuals
H	20
I	4
J	14
K	7
L	3
T	13
U6a	3
U6b1	23
V	2

haplotypes was observed, what represents a high variability, being HV1 the most polymorphic region. A summary of the haplotype results is shown in Table 1.

The values obtained for genetic diversity and discrimination power (Table 2) are comparable to other European populations, and suggests a high usefulness of the mtDNA analysis for forensic identification in this population. The majority of the nucleotide substitutions were transitions C–T.

Although haplogroups are mainly determined by SNP positions in the coding region, some haplogroups have been identified from the HV1 data set (Table 3). The haplogroups U6a and U6b are characteristic of North African populations, reaching the highest frequencies in Berbers [4]. In this work, the highest frequency was for the sub-haplogroup U6b1, defined by an additional substitution at 16163, which reaches 88% of the total U6 sequences obtained. This feature suggests an unique introduction of the North African lineages into the Canary Islands in the past, and it is in agreement with other previously published data [5].

Control-region sequences from 9791 individuals grouped into 56 populations were assembled from the current and previous studies to examine broad affinities at the population level. The FST values were based on pairwise sequence differences between positions 16090 and 16365 to allow maximum comparability between all populations. The genetic distances between all 56 populations were summarized by a Principal Co-ordinate analysis (PC) (data not shown). A broadly north–southwest tendency is evident in the first dimension (89.9% of the variance, axis 1), with the sequences from Basque Country and Sao Tome/Bioko Islands occupying the respective poles. Both mtDNA sample sets from Canary Islands are centrally located in the PC plot mainly due to their strong mitochondrial gene flow from North Africa and Europe [5].

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