



Genetic variation at autosomal and Y-chromosome STRs, and HVRI mtDNA in Ibiza population

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Abstract. We have analyzed nine autosomal and eight Y-chromosome STR loci, as well as the sequencing of the HVRI mtDNA in a sample from the Ibizan population. Differences between Ibiza and the other islands of the Balearic Archipelago have been detected, especially in the origins of the founder settlements. The mtDNA and autosomal STRs diversities evidence the differentiation, but the Y-chromosome polymorphism did not show clear differences between Ibiza and the other islands.
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1. Introduction

The Balearic Archipelago is located in the western Mediterranean Sea, near the eastern coast of the Iberian Peninsula. It is composed of three major islands, Majorca, Minorca and Ibiza. Due to their geographic position in the Mediterranean Sea, the Balearic Islands have been settled by people of different populations throughout their history. This fact has moulded the genetic structure of the human population of the archipelago. Differences between Ibiza and the other two main islands, Majorca and Minorca, can be found, especially in the origins of the founder settlements [1]. Although there is evidence that Majorca and Minorca were first settled about 4000 B.C. (the “talaiotic”-stone builder-culture), there is no clear proofs of settlements on Ibiza until the arrival of the Carthaginians in 654 B.C. They remained in Ibiza for at least five centuries and were not restricted to coastal areas but also colonized the interior of the island. Ibiza was annexed, but never occupied by Romans. During its history, the human population of Ibiza has been remained isolated and, thus, received little gene flow from outside. This apparent reproductive isolation was disturbed by a 1970s tourist influx that doubled the total population of the island.

The aims of the present study were: to measure the genetic differentiation of the Ibiza population from the other Balearic islands based on the analysis of autosomal and

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Y-chromosome microsatellites, and the comparative analyses between Y-specific markers and mtDNA sequence data on the same individuals, that allow us to study possible differences in male versus female contribution patterns.

2. Materials and methods

We have obtained blood samples from 102 individuals of the autochthonous population from Ibiza Island. DNA was organically extracted following a standard protocol. Autosomal STRs: PCR amplification of the tetrameric STR systems D3S1358, vWA, FGA, D8S1179, D21S11, D18S51, D5S818, D13S317 and D7S820 was achieved in a GeneAmp PCR System 2400 (Perkin-Elmer). Y-chromosome markers: Eight microsatellites (DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385) and the *Y-Alu* polymorphism at the DYS287 locus were analyzed by means of manual analysis of the polyacrilamide gels and automatic analysis (ABI 310, Perkin Elmer). Mitochondrial DNA data were obtained from a previous work [2]. The statistical analyses were mainly performed using the Arlequin software [3].

3. Results

3.1. Autosomal polymorphism

It was described a total of 71 alleles for the 9 autosomal markers studied: 6 at D3S1358, 7 at vWA, D5S818 and D13S317, 9 at D8S1179, 10 at D21S11, 12 at FGA and 13 at D18S51. As indicated in Table 1, a high level of variability was found (79.9%). No single locus evidenced a departure from HWE. The most significant markers, in terms of genetic differentiation, were D18S51, FGA, D7S820, D21S11, D8S1179 and D13S317.

3.2. Y-chromosome variation

We analysed 45 paternally unrelated male samples from Ibiza for minimal Y-chromosome haplotype. We obtained 35 distinct Y-chromosome haplotype configurations. The most frequent haplotype 14-10-17-24-11-13-13-11.14 (DYS19-DYS2389I-DYS2389II-DYS390-DYS391-DYS392-DYS393) was found in four individuals, 27 were observed in unique copies and seven haplotypes were shared by two individuals. Haplotype diversity was estimated at 0.987 ± 0.008 (Table 1).

Table 1
Genetic diversity parameters in Balearic Archipelago

Population	Autosomal STRs			Y-chromosome STRs			mtDNA		
	Number of alleles	Observed heterozygosity	PD ^a	Gene diversity	Haplotype diversity	PD ^a	Haplotype diversity	Haplogroup diversity	PD ^a
Ibiza	8–9	0.797	>0.999	0.415	0.987	0.778	0.938	0.811	0.380
Majorca	9–10	0.800	>0.999	0.459	0.988	0.824	0.957	0.870	0.689
Minorca	8–9	0.822	>0.999	0.473	0.984	0.868	0.987	0.902	0.761

^a PD = power of discrimination.

3.3. *mtDNA* variation

With respect to *mtDNA* [2], Ibiza showed 19 different haplotypes, 26 polymorphic sites and 3.8 pairwise differences. Ten different haplogroups were represented, with a low (0.811) haplogroup diversity. In contrast with the other Balearic populations, haplogroup T (original from the Near East) was well represented in this population. The African haplogroup L was also detected.

It is highly significant the low diversity that shows Ibiza, especially in the case of *mtDNA* (Table 1). The correlation between the different markers elaborated by means of Mantel test was high significant between *mtDNA* and STR, but not significant between Y-chromosome and STR and Y-chromosome and *mtDNA*. The *mtDNA* and autosomal STRs evidence the differentiation, but Y-chromosome polymorphisms did not show clear differences between Ibiza and the other islands.

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