



Phylogeny of the mtDNA haplogroup U6. Analysis of the sequences observed in North Africa and Iberia

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Abstract

Comparison was performed between 41 U6 sequences observed in North Africans, 14 in Iberia and 42 in the Canary Islands. Only 2 of the 14 Iberian sequences belong to the Canarian-specific U6b1 sub-haplogroup, and thus represent introduction from these islands. The remaining 12 sequences are very diverse (only 4 share the same haplotype), which do not support a single founder for the U6 lineages in Iberia.

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1. Introduction

The haplogroup U6, defined by transitions at np 16172 and 16219 in the hypervariable region I (HVRI) of the mitochondrial DNA (mtDNA), is characteristic of North African populations, reaching the highest frequencies (~ 20%) in Berbers [1]. The cluster U6 has been subdivided into two subgroups: U6a defined additionally by the transition at np 16278 and U6b having a transition at np 16311 [2].

Outside North Africa, the highest frequencies (10–16%) are observed in the Canary Islands [2], which are located in the Atlantic Ocean close to the northwest African coast, and which were a centre of the Iberian overseas expansion from the 15th century. There is indeed a typical Canarian U6 sub-haplogroup, U6b1, defined by an additional substitution at np 16163, which encompasses 93% of the U6 sequences observed in the Canary Islands, pointing to a unique introduction of the North African lineages into these islands

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around 2800 ± 900 years ago, with the less frequent sequences likely to have been introduced subsequently [2].

In Iberia, 14 U6 sequences have also been reported, a much higher frequency than in the rest of Europe (only 1 sequence in Sweden and another in Sicily, in a European-wide database). Out of 14 U6 sequences, 11 were from the north of Iberia: 7 in North Portugal (7.0%) [3], 2 in Galicia (2.2%) [4], and 2 in northeastern Spain (1.7%) [5]; the remaining 3 being described in a general sample from Portugal (5.5%) [6].

To clarify whether the Iberian U6 sequences could have been introduced in one major prehistoric event, as it seems to have been the case in the Canary Islands, or if they are the outcome of more recent introductions, for example, during the Islamic period, we took a close look at the different HVRI sequences.

2. Materials and methods

U6 sequences were collected from several publications summing a total of: 41 in North Africa [1,6,8,9], 42 in the Canary Islands [2], 14 in Iberia [3–6], and 9 in sub-Saharan Africa [7,8].

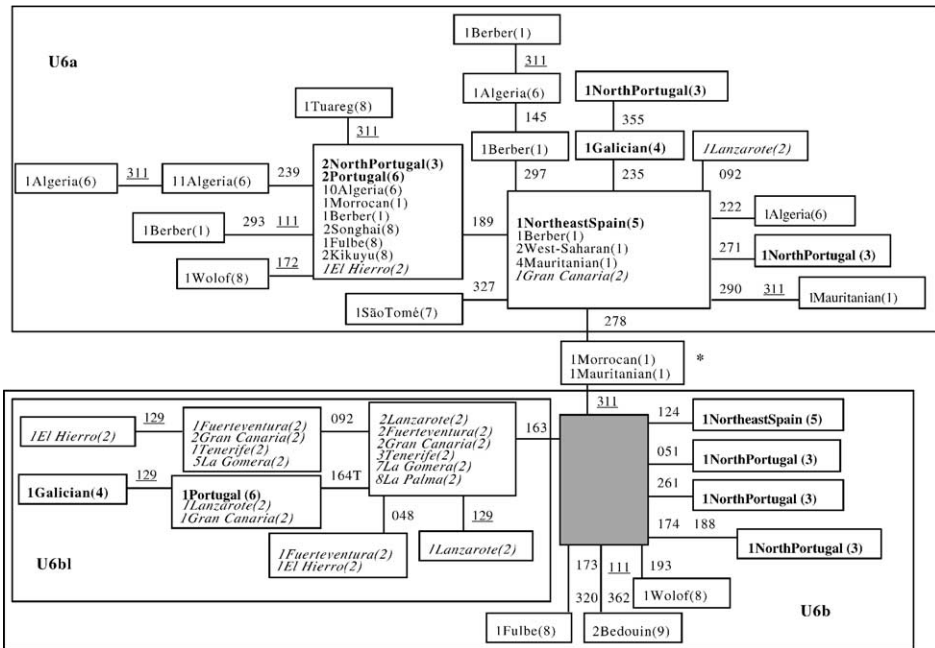


Fig. 1. A most parsimonious tree of sequences belonging to cluster U6. Root motif 172–219 indicated with an asterisk. Branches are labelled by the nucleotide positions in HVRI (minus 16000) to designate transitions; transversions are further specified and positions underlined represent parallel mutations. The grey rectangle corresponds to an empty node. Iberian samples are in italic and Canarian in bold. Numbers preceding the population refer to the number of sequences observed with that motif and numbers inside brackets represent the bibliographic reference.

3. Results

Of the Iberian sequences, 2 belong to the Canarian-specific sub-haplogroup U6b1, and these were likely to have been introduced from these islands (a short-lived Guanche slave trade is recorded). The remaining 12 are very diverse, corresponding to 9 different haplotypes, 5 of which belong to U6a and 4 to U6b. Only 4 sequences share the same widespread U6a substitution pattern 16172-16189-16219-16278 (Fig. 1).

4. Discussion

Taken together, these results do not support the hypothesis of a single founder for this haplogroup in Iberia. Whether it was introduced in a single event from a source population with a diverse set of U6 lineages or whether it was introduced over a long period from several sources is not clear. Since it is currently not possible to infer the U6 founder types in Iberia, it is impossible to date its introduction there. An enlarged North African database would aid the solution of this puzzle.

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