



Y-chromosome lineages from Portugal, Madeira and Açores record elements of Sephardim and Berber ancestry

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Abstract. A total of 553 Y-chromosomes were analyzed from mainland Portugal and the Archipelagos of Açores and Madeira, in order to characterize the genetic composition of their male gene pool. A large majority of the male lineages could be classified as belonging to three basic Y chromosomal haplogroups R1b, J, and E3b. While R1b, accounting for more than half of the lineages in any of the subpopulations, is a characteristic marker of many different West European populations, haplogroups J and E3b consist of lineages that are typical of the circum-Mediterranean region or Eastern Africa. The present composition of the Y chromosomes in Portugal in E3b haplogroup likely reflects a pre-Arab component shared with North African populations or testifies, at least in part, the influence of Sephardic Jews. A marginally low sub-Saharan African Y chromosome component indicates the presence of sex-related gene flow, most likely mediated by the Atlantic slave trade. © 2006 Published by Elsevier B.V.

Keywords: Portuguese; Y-chromosome; SNP; STR

1. Introduction

For centuries the Iberian Peninsula (IP) was a melting pot for various populations of different origins. However, three groups have been the focus for vigorous debate over their contribution to the present-day Portuguese genetic gene pool: the Sephardim Jews, the

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Arab–Berbers and the sub-Saharan. Jews are known to have inhabited the IP since at least the 3rd century AD. Berbers from Mauritania started to expand into IP in 711 AD. Most of the sub-Saharan component in Portugal dates back to the 15th century when the importation of sub-Saharan slaves accelerated, following the establishment of a commercial Atlantic network.

The Archipelagos of Açores and Madeira settled by the Portuguese in the first half of the 15th century played a major role in the complex Atlantic trade network. In Madeira, the non-Iberians were mainly from Italy and because of the heavy involvement with the slave trade, the island saw a considerable input of sub-Saharan males. Inter-island movements have also been recorded, and it is known that many settlers of the Açores came from Madeira. The Açores received a significant portion of male settlers from Flandria who married Portuguese women. Using Y-chromosome SNPs and STRs, we aimed to characterize the extant paternal lineages to investigate if genetic markers possibly transmitted by Sephardim Jews are detectable in today's gene pool, and to what extent North African or Berber lineages are associated with the Muslim period.

2. Material and methods

We studied 553 unrelated males from mainland Portugal (subdivided into North, Centre and South) and the archipelagos of Madeira and Açores. Blood samples were collected and genomic DNA was isolated by standard protocols. Y-SNPs were amplified with primers previously described [1,2]. The selected SNPs are shown in Fig. 1 in their phylogenetic order. Six STR loci (DYS19, DYS388, DYS390, DYS391, DYS392 and DYS393) were assayed in chromosomes belonging to haplogroups E3b and J1 using standard methodology [3]. DYS439 and A7.1 were tested to further define E3b1 subclusters [4] and DYS439 for J1 lineages. We followed the haplogroup nomenclature proposed by the YCC [5] for Y chromosome typing. Frequencies of Y haplogroups for each region and

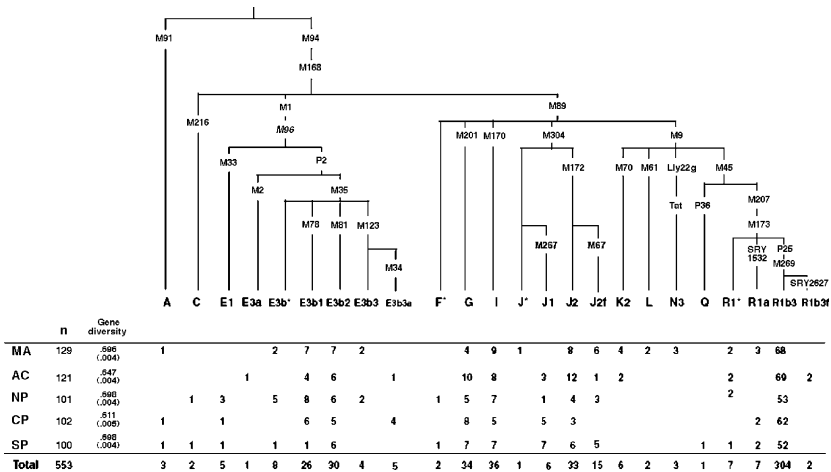


Fig. 1. Phylogenetic tree of the Y-chromosome haplotypes. Haplogroup defining mutations assayed in this study are shown along branches. Populations: Madeira—MA, Açores—AC, Portugal (North—NP, Centre—CP, and South—SP).

gene diversity measures were obtained using Arlequin v2.000 [6]. The age of STR variation within each haplogroup was estimated using calculations developed by Zhivotovsky et al. [7].

3. Results and conclusions

Haplogroup R1b3 is the most dominant in Portugal, Açores and Madeira covering more than half of the lineages in each population. This frequency is typical in all West European populations, reflecting a cline [8] and likely continuity of the Paleolithic gene pool in Europe [9]. Together with R1b, haplogroups J and E3b comprise 78–83% of the Y-chromosomal gene pool of each sampled population.

More than 10% of Portuguese Y chromosomes are classified in haplogroup J, thought to have originated in the Middle East or North East Africa [2,10], both main J1 and J2 clusters appearing in the Levant at Neolithic times [11]. The Portuguese J1 microsatellite combined haplotypes is consistent with the Cohen Modal Haplotype, this supports the view that Portuguese J1 lineages are not due to Arab gene flow, and could have entered the country independently, possibly with Sephardim and/or other Near East peoples.

Haplogroup E3b is widespread in Northwest Africa, East Africa, the Middle-East [1,4] and is also common in Europe, albeit at variable frequencies [9]. Regional sub-clustering of the STR haplotypes on the Portuguese E3b1 suggests that these lineages could have spread from the Balkans all over Europe during the Neolithic [4]. This is furthermore supported by the age of STR variation (8.8 ± 4.1 ky). E3b2 has been observed at variable frequencies in Iberian populations and is almost absent, or occurs at very low frequencies elsewhere in Western Europe [1,4,10,12]. Half of the Portuguese E3b2 is shared with Berber and/or Arab speaking populations from Morocco [13], and the age variation here is actually identical to that of North Africa (8.1 ± 3.2 ky vs. 8.6 ± 2.3 ky). These findings suggest a flow of Berber-related lineages into the male Portuguese population.

Typically sub-Saharan lineages represented by haplogroups A and E1 are virtually absent in European populations [10] and Northwest Africa, although E1 is found among Berbers [12]. The presence of both A and E1 haplogroups may be independent from the slave trade, suggesting a pre-Neolithic migration from North Africa.

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