

Research article

Y-chromosome lineages in São Tomé e Príncipe and Cabo Verde islands: Different input of European influence

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Abstract

The Y-chromosome haplogroup composition of the population of São Tomé e Príncipe and Cabo Verde Archipelagos was profiled by using 24 biallelic markers, and compared with populations from Europe, Africa and the Middle East. According to the traditional view, these archipelagos colonized by the Portuguese in the 15th century were settled mainly by West African slaves, with the addition of a minor fraction of male colonizers from Europe. Although the major proportion of the founding population of São Tomé e Príncipe cluster in haplogroup E3a (84.2%), very common among sub-Saharan, this lineage was observed at a frequency of only 15.9% in Cabo Verde. Haplogroups I, J and R1, characterized of populations of Europe and the Middle East account for more than half of the paternal lineages of Cabo Verdeans (53.5%). These West Eurasian haplogroups are found at a frequency of only 12.5% in the population of São Tomé e Príncipe. Our findings suggest that despite the sub-Saharan genetic background of these archipelagos, a relevant contribution of European paternal lineages is present in nowadays populations indicating that gene flow from multiple sources have been important in the formation of the diversity of the islanders, nevertheless with a different degree of admixture.

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1. Introduction

The São Tomé e Príncipe (STP) and Cabo Verde (CV) Archipelagos were inhabited at the time of their discovery in the 15th century. In CV, the first settlers were mostly European males recruited from Portuguese nobles, Genovese adventurers, exiles, and convicts of the Crown. Among the first slaves brought into the Archipelago from the West African coast of the region now known as Senegambia, were also Guanches from the Canary Islands and North Africans. The population grew quickly, mainly due to the slave trade from the present-day Guiné-Bissau region [1,2]. Following the discovery of STP Islands in the Gulf of Guinea by Portuguese sailors in 1470, CV started to decline as a slave-trading center, which led to a minimal input of new European settlers. Nevertheless, the European population in CV has never been numerous.

By its turn, the settlement process of STP Archipelago was composed mainly by sub-Saharan African slaves from the Gulf of Guinea, Congo and Angola, recruited to work in local

plantations, and in a minor extent Portuguese. In the first centuries after their discovery STP acted as an outpost for slave trade between Africa and America. Besides Portuguese, other Europeans were involved in this trade along the coast of Africa, namely French, Spanish, Dutch and English, which could have contributed in a minor scale to the present-day genetic pool of the islanders [3]. In the 19th century, due to a new economic cycle supported by coffee and cacao plantations, the islands received a new wave of sub-Saharan African people recruited from CV Archipelago, Angola and Mozambique.

The main aim of the present work has been to analyze the Y-chromosome gene pool of the present-day STP and CV populations and to quantify the relative paternal input of European and African origin on both Archipelagos.

2. Material and methods

The population included in this study consisted of a total of 351 unrelated males, 201 from CV Archipelago, and 150 from STP Archipelago. Blood samples were collected and genomic DNA was isolated by standard protocols. Y-SNPs were amplified with primers previously described [4,5]. The selected SNPs are shown in Fig. 1 in their phylogenetic order. We

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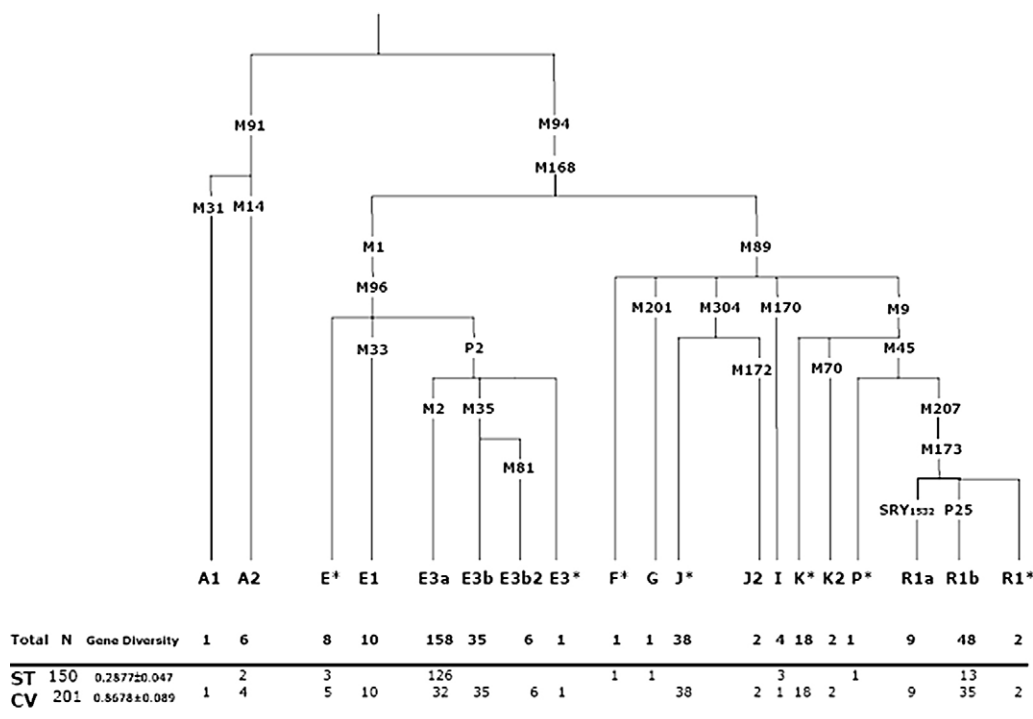


Fig. 1. Phylogenetic tree of the Y-chromosome haplogroups. Haplogroup defining mutations assayed in this study is shown along branches. Populations: São Tomé e Príncipe (STP), Cabo Verde (CV).

followed the haplogroup nomenclature proposed by the YCC [6] for Y-chromosome typing. Frequencies of Y haplogroups for each region and gene diversity measures were obtained using *Arlequin v2.000* [7].

3. Results and conclusions

The most frequent haplogroup found in STP is E3a (84.2%) also the most widespread clade in sub-Saharan Africa and the most frequent in West African populations [8–10], emphasizing the origin of the main settlers of these islands. This clade reaches 80% among Senegalese [9], 71% of the Guinean paternal lineages, 15.9% in CV [11] but is absent in Europe [8] and Iberia [12,13]. In northwest Africa, E3b is the most common cluster reaching ~75% of Y-lineages [12]. This haplogroup was not found on our STP population but constitutes 20.4% of CV [11].

The second most common haplogroup in STP is R1b (8.7%) a typical haplogroup from Western Europe, most likely carried by Portuguese settlers suggesting their genetic contribution to this population. West Europeans almost completely lack R1a, but show the highest frequency of R1b. Iberian populations in particular, show 77% of R1b lineages and 1% or less of R1a lineages [12]. More than 17% of CV Y-chromosomes are R1b, a West European influence in the archipelago settlement process [11]. Haplogroup I, also a characteristic clade for many different European populations, constitute 2% of STP population. Other haplogroups, like P* and G, with a Eurasian origin, and F* with a Middle East origin, show only marginal frequencies.

Haplogroups I, J, K, common in circum-Mediterranean populations of Europe, the Middle East, and North Africa but absent in sub-Saharan Africa, account for 30.3% of the CV sample. Haplogroup J appears with a frequency of 19.4% contrasting with 2–7% in Iberian and North African [8,12].

Haplogroups A1 and A2, basal clades in the Y-human phylogenetic tree, typical in sub-Saharan Africans at modest frequencies [4,6,9] constitute 2% of our samples.

Conflict of interest

None.

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