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High-resolution analysis of Y-biallelic markers in three populations from São Tomé e Príncipe

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Abstract. Twenty Y-chromosome biallelic polymorphisms were analysed in samples from Angolares, Forros and Tongas, three population groups from the African archipelago of São Tomé e Príncipe. Although most male lineages belonged to Sub-Saharan haplogroups, the component of European origin reached 23.9% in the archipelago as a whole. This value contrasts with the previously reported absence of European female lineages in the São Tomean mtDNA pool and reflects the strong sex-biased admixture process between Europeans and Africans that characterized the demographic history of the archipelago. The lowest diversity levels as well as the smallest proportion of putative male European ancestry were registered among the Angolares, representing clear genetic signs of their past relative isolation from other São Tomé e Príncipe's inhabitants.

Keywords: Y-chromosome biallelic markers; São Tomé e Príncipe; Population groups; Admixture proportions

1. Introduction

Angolares, Forros and Tongas are three population groups from São Tomé e Príncipe, a small African archipelago (Gulf of Guinea) whose population history traces back to the end of the XV century when Portuguese navigators discovered the uninhabited islands.

Previous data derived from Y-chromosome STRs [1] and mtDNA [2] indicated that the Angolares presented reduced genetic diversity and a slight differentiation from the other

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São Tomean groups. In order to gain further insights into the genetic relationships between the groups and to obtain estimates of the relative contribution of Sub-Saharan and European male lineages in shaping its present day gene pool, we have extended the genetic characterization to the variation determined by 20 biallelic markers, SNPs and indels, from the non-recombining region of the Y-chromosome.

2. Material and methods

Blood samples were collected from 55 unrelated Angolares, 39 Forros and 44 Tongas. DNA was extracted using standard phenol–chloroform or Chelex methods. Markers were typed using a hierarchical approach, according to [3] and [4]. Genetic diversities and distances were calculated with the software ARLEQUIN ver 2.000 [5].

3. Results and discussion

The 20 Y biallelic markers tested yielded 22 different haplogroups, out of which 11 were present in the São Tomean groups analysed (Fig. 1).

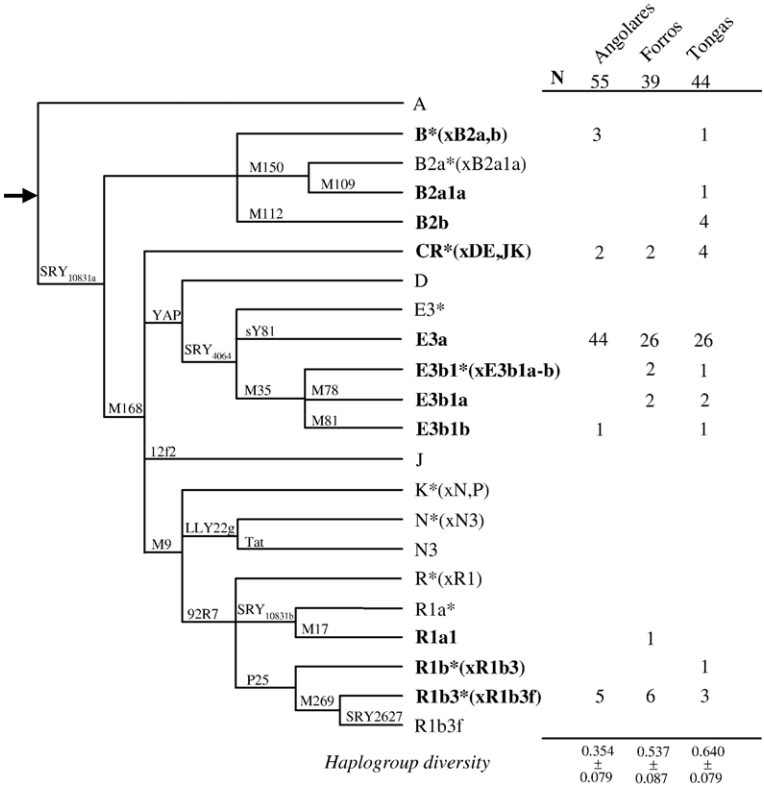

Fig. 1. Tree of 20 Y-chromosome biallelic makers typed and haplogroup proportions and diversities in Angolares, Forros and Tongas.

None of the genetic distances between Angolares, Forros and Tongas was statistically significant, but the Angolares exhibited the lowest number of different haplogroups and levels of diversity (Fig. 1) reinforcing previous data pointing to reduced diversity in the group [1,2]. This seems to represent a drift effect that can be explained by their history of isolation and confinement to the south-eastern tip of São Tomé for many years [1,2].

Among the different haplogroups detected in this study, the E3a, B2a1a, B2b and B*(xB2a,b) are highly specific of Sub-Saharan Africa. E3a represented the major fraction of male lineages being present in 69.1% of the São Tomeans, which is a value in the range of those usually found in other Sub-Saharan Western populations. Among the São Tomean groups, it reached the highest frequency, 80.0%, in the Angolares. The B lineages detected – B2a1a, B2b and B*(xB2a,b) – are representatives of one of the oldest clades in the Y-chromosome phylogeny and globally comprised 6.5% of the São Tomean Y-chromosomes. The frequency of 5.5% of B*(xB2a,b) lineages in Angolares constitutes the highest value so far registered for the haplogroup that until now only has been sporadically detected in a few African populations. Once more, this finding suggests the occurrence of drift effects during the history of the Angolares.

The remaining haplogroups were representatives of CR*(xDE,JK), E3b1 or R1a,b lineages, which are common and widespread in Europe but virtually absent in Sub-Saharan Africa. Since these lineages were likely introduced in São Tomé e Príncipe by European males, they can be used to infer the degree of admixture between Europeans and Africans in the archipelago. In São Tomé e Príncipe as a whole, the proportion of male lineages of European origin was 23.9% which is a rather high value particularly when confronted with the previously reported absence of European mtDNA sequences in the archipelago [2]. This disproportion of European component in the male and female pools testifies the strong bias in the admixture process that characterized the demographic history of the archipelago.

The male component of European ancestry was 33.3% in Forros, 27.3% in Tongas and approximately two-fold less, 14.5%, in Angolares. This last value indicates that gene flow between Europeans and Angolares was more restricted than between Europeans and other São Tomean inhabitants, and represents an additional sign of the past relative isolation of the group.

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