

The Y-chromosome in the Azores Islands: Phylogeny and diversity

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Abstract. The Azores, a Portuguese archipelago located in the North Atlantic Ocean, had no native population when the Portuguese first arrived in the 15th century. The islands were populated mainly by the Portuguese, but Jews, Moorish prisoners, African slaves, Flemish, French and Spaniards also contributed to the initial settlement. To understand the paternal origins and diversity of the extant Azorean population, we typed genomic DNA samples from 172 individuals using a combination of 10 Y-biallelic markers (YAP, SRY-1532, SRY-2627, 92R7, M9, sY81, Tat, SRY-8299, 12f2 and LLY22g) and the following Y-chromosomal STR systems: DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393 and DYS385 a/b. We identified nine different haplogroups, most of which are frequent in Europe. Haplogroup J* is the second most frequent in the Azores (13.4%), but it is modestly represented in mainland Portugal (6.8%). The other non-European haplogroups, N3 and E3a, which are prevalent in Asia and sub-Saharan Africa, respectively, have been found in the Azores (0.6% and 1.2%, respectively) but not in mainland Portugal. A Y-chromosomal haplotype was constructed for each individual using the seven loci. In total, 118 different haplotypes were observed in the 172-sample set (68.6% discriminatory capacity). Haplotype diversity value was high (0.9994), due to high variability of the Y-STRs. Moreover it is important to notice the great genetic diversity observed within the Azorean group. Microsatellite data indicate that the mean gene diversity (D) value for all the loci analysed in our sample set is 0.590 (values range from 0.4592 for DYS393 to 0.8212 for DYS385). Taken together, our analysis suggests that the current paternal pool of the Azorean population is, to a great extent (59.3%), of Portuguese descent with significant contributions from people with other genetic backgrounds. © 2005 Elsevier B.V. All rights reserved.

Keywords: Y-chromosome; Y-STR; Y-SNP; Azores Islands

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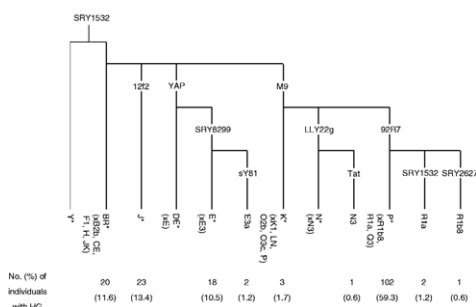


Fig. 1. Phylogenetic tree of the Y-chromosome haplogroups and their percent frequencies in the Azorean sample.

venipuncture into EDTA tubes. DNA was extracted using the PUREGENE® kit (Gentra Systems Inc.).

A total of 10 Y-biallelic markers were selected: YAP, SRY-1532, SRY-2627, 92R7, M9, sY81, Tat, SRY-8299, 12f2 and LLY22g and the following seven Y-chromosomal STR systems: DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393 and DYS385 a/b. Primer sequences were obtained in the Y-STR haplotype database (<http://www.ystr.org>).

Population differentiation between the Azores and other populations was assessed using haplogroup frequencies included in Arlequin software.

3. Results and discussion

A Y-chromosomal haplotype was constructed for each individual, using seven loci. Overall, 118 different haplotypes were observed in the 172-sample set (68.6% discriminatory capacity). The mean gene diversity value across loci in the Azorean sample (Table 1), $D=0.590$, is higher than the value reported for northern Portugal and than that observed for the Europeans [2]. Likewise, haplotype diversity value in Azores (0.9994) is higher than in northern Portugal and Europe [2]. The diversity found in Azorean Y-chromosome is probably due to the admixture of Portuguese with other populations.

The presence of haplogroups of widespread distribution in Europe (Fig. 1), in combination with others of clear sub-Saharan, Asian and Middle East origin, reflects the diverse patterns defining the present Azorean Y-chromosome pool. We conclude that the current paternal Y-chromosome pool in the Azores is of Portuguese descent, with a considerable contribution of individuals from multiple origins [3].

References

- [1] L. Mota-Vieira, et al., Human DNA bank in São Miguel Island, Azores: assembly and analysis, *Eur. J. Hum. Genet.* 12 (Suppl. 1) (2004) P0995.
- [2] A. Gonzalez-Neira, et al., Distribution of Y-chromosome STR defined haplotypes in Iberia, *Forensic Sci. Int.* 110 (2000) 117–126.
- [3] P.R. Pacheco, et al., The Y-chromosomal heritage of the Azores Islands population, *Ann. Hum. Genet.* 69 (2005) 145–156.