

## Human DNA bank in Sao Miguel Island (Azores): A resource for genetic diversity studies

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**Abstract.** The peopling of São Miguel Island in the 15th century was made by Portuguese and settlers of foreign origin (Flemish, Jews, Moorish prisoners and black slaves), generating an admixture signature. Thus, to unravel São Miguel's population genetic background and to characterize its population's polymorphisms, we decided to establish a human DNA bank. Here, we describe the construction of the DNA bank and analyse the information of 997 samples obtained from healthy blood donors. The bank follows the international ethical guidelines, which include informed consent, confidentiality, anonymity of personal data, and abandonment in case of expressed will. DNA was isolated from blood samples, coded and immediately stored in a locked refrigerator. The identifiable DNA bank has self-reported data concerning sex, age, birth, current place of living, and parental birthplaces. The samples are representative of all the island's municipalities ( $r=0.995$ ,  $p<0.01$ ). The majority (87%) of the participants is male, with mean age of 36.3 years (18–64 years). Birthplace analysis reveals that 902 (90%) have both parents born in São Miguel. Moreover, 477 (54%) have their parents born in the same locality, confirming a relatively high rate of consanguinity in rural area. To date, this DNA bank was used to assess the Y-chromosome phylogeny and diversity in Azorean population. Now, we are analysing autosomal STRs for the better understanding of the gene pool and genetic structure of the archipelago's people. © 2005 Elsevier B.V. All rights reserved.

*Keywords:* Human DNA sampling; DNA bank; Informed consent; Confidentiality; Molecular epidemiology

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## 1. Introduction

The establishment of a human DNA bank is of particular importance in the characterization of a population's polymorphisms, particularly in geographically isolated places, as well as for molecular epidemiological studies. Here, we describe briefly the building of a DNA from the population living in São Miguel Island (Azores Archipelago, Portugal, Fig. 1), in order to characterize their genetic background and to carry out biomedical research. Discovered in the 15th century, the peopling of São Miguel was made by Portuguese and settlers of foreign origin (Flemish, Jews, Moorish prisoners and black slaves), creating a signature of admixture throughout 25 generations.

## 2. Materials and methods

The bank consists of DNA from blood donors and follows the international ethical guidelines for sample collection, processing and storage, which include informed consent, confidentiality and anonymity of personal data, and abandonment of the study in case of expressed will [1,2]. Two departments at the Hospital were involved: the Hematology Department (HD) and the Molecular Genetics and Pathology Unit (MGPU). At the HD, all blood donors received information concerning the construction of the DNA bank, through the distribution of a leaflet and of complementary explanation by a technician if requested. Sampling (7.5 ml) and labelling (code 1) was performed at the time of blood donation, only after signing the informed consent. Each DNA has self-reported data. The blood samples were delivered to the MGPU with copies of records (without name identification). At this unit, the samples received a second code, and the records were computerized in an Excel database. DNA was extracted using the PUREGENE® kit (Gentra Systems Inc.) and quantified. All DNA samples were stored in a locked refrigerator.

## 3. Results and discussion

The identifiable DNA bank from São Miguel Island consists of 997 samples, doubly coded, each sample having a self-reported data concerning sex, age, birth, current place of living, and parental birthplaces. The 997 samples are representative of all the island's municipalities ( $r=0.995$ ,  $p<0.01$ , Table 1). The majority (87%) of the participants are male, with mean age of 36.3 years (18–64 years). Birthplace analysis reveals that 902 (90%) have both parents born in São Miguel. Moreover, 477 (54%) have their parents born in the same locality, confirming a relatively high rate of consanguinity in rural area [3].

The use of whole-blood specimens to obtain genomic DNA yields large quantities of high-quality DNA that provides sufficient material for the current and future molecular applications at a

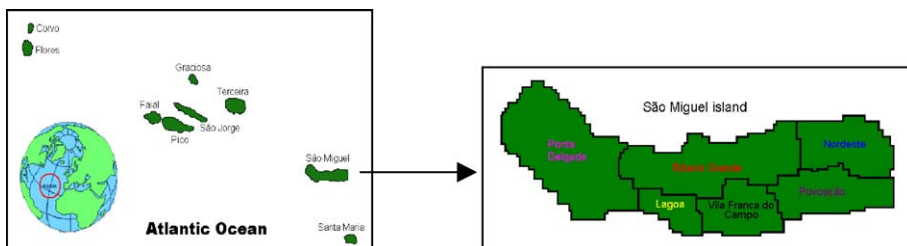


Fig. 1. Map of Azores archipelago and São Miguel island.

Table 1  
Demographic data from São Miguel Island and DNA sample distribution

Municipalities of São Miguel Island	Inhabitants (2001 Census)		DNA sample distribution	
	Number	%	Number	%
Lagoa	14,126	11	88	9
Nordeste	5291	4	28	3
Ponta Delgada	65,854	50	549	55
Povoação	6726	5	42	4
Ribeira Grande	28,462	22	188	19
Vila Franca do Campo	11,150	8	102	10
Total	131,609	100	997	100

sustainable cost of storage. The identifiable DNA bank described here contains samples from healthy individuals that can be used in the characterization of polymorphisms in São Miguel's population. To date, this DNA bank was used to assess the Y-chromosome phylogeny and diversity in Azorean population [4]. Now, we are analysing autosomal STRs for the better understanding of the gene pool and genetic structure of the archipelago's population [5–7].

This DNA bank provides us opportunities to collaborate in international projects. No commercial benefit is involved in the establishment of this DNA bank.

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