

Study of y-SNPs genetic markers with forensic interest and ancestry informative power in PALOP's immigrant populations in Lisboa



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ABSTRACT

The increasing number of immigrants in Portugal is an unavoidable reality. According to Portugal Contemporary Base - PORDATA -, by the end of 2013, the total number of immigrants from PALOP (Portuguese-speaking African countries) in Portugal was about 100,000, and from those, about 75,000 are part of Lisboa population. The migratory phenomenon in Portugal can become one of the main factors for the genetic variability.

Markers located on the Y chromosome have special interest and application in origin and evolution population studies, because great part of the chromosome does not undergo recombination. Y-SNPs are single nucleotide polymorphisms, with not only ancestry and population applications, but also with forensic application.

Since there is no data for Y-SNPs markers of PALOP immigrants living in Lisboa, our aim is the characterization of those groups of individuals by typing them with a panel of Y-SNPs proposed by Rosser and collaborators in 2000, with 9 Y-SNPs markers, and compare different groups of individuals/populations. Thus, 200 bloodstain samples belonging to immigrant individuals from Angola, Guinea-Bissau and Mozambique were studied. DNA extraction was performed with Chelex[®] 100 and amplified in a multiplex PCR with primers for 9 Y-SNPs.

Few differences between the studied African populations are shown, which reveals that this Y-SNPs panel is not useful for differentiation purposes within those populations.

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1. Introduction

Since the early 70s, the flow of immigrants from African countries to Portugal has increased. According to Portugal Contemporary Base - PORDATA -, by the end of 2013, the total number of immigrants from PALOP (Portuguese-speaking African countries) in Portugal was about 100,000, and from those, 75,000 are part of Lisboa population.

The number of immigrants in Portugal is an unavoidable reality and the migratory phenomenon in this country, and particularly in Lisboa, can become one of the main factors for the genetic variability [1–7].

The single nucleotide polymorphisms (SNP) typically involve substitution of a nucleotide in DNA sequence, resulting an exchange on the sequence. SNP in forensic genetics are mainly used in samples where DNA is degraded, since only a small target

DNA region is necessary because the size of the amplified product is under 100 bp. Recent advances in the SNP markers, show us the growing interest of Forensic Genetics in their use [8,9].

Markers located on the Y chromosome have special interest and application in origin and evolution population studies. Each chromosome has the story of millions of years of evolution and Y chromosome is no exception, telling the story of a male lineage. Having diverged from the same ancestral that the X chromosome, the Y chromosome passed from generation to generation without change, except for the occurrence of mutations. Y-SNPs are single nucleotide polymorphisms, with ancestry and population applications, and also with forensic application.

Since there is no data for Y-SNPs markers of PALOP immigrants living in Lisboa, our aim is the characterization of those groups of individuals by typing them with a panel of Y-SNPs proposed by Rosser and collaborators in 2000, with 9 Y-SNPs markers, and compare different groups of individuals/populations.

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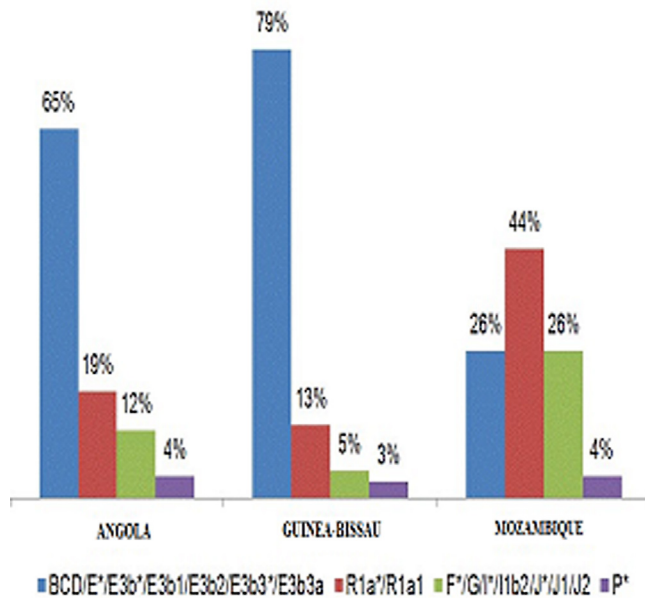


Fig. 1. Distribution of studied individuals/populations by haplogroups.

2. Materials and methods

211 bloodstain samples were studied: 123 from Angola (immigrant population inserted at Y-HRD database with number YA003921), 61 from Guinea-Bissau (immigrant population inserted at Y-HRD database with number YA003922) and 27 from Mozambique, collected from immigrant individuals inhabitants of Lisboa metropolitan area, and undergoing forensic investigations at Instituto Nacional de Medicina Legal e Ciências Forenses (INMLCF). The number of studied individuals from each one of the different origin African countries – Angola, Guinea-Bissau, and Mozambique – reflects the representation that each immigrant group has in Lisboa population Fig. 1.

An interview was conducted in order to register personal data of the studied individuals, particularly the name, the age, the birth place, the individual and the parental ethnicity.

According to Portuguese legal regulations, samples from routine forensic cases ongoing at INMLCF can be used for investigation purposes, which, naturally, include genetic studies. In our study all samples are used with special codification without any connection to personal or judicial data related to the donor.

DNA extraction was performed with Chelex[®] 100 [10] and Y-SNP typing with a multiplex PCR with primers for 9 Y-SNPs (M22, P25, SRY1532, 92R7, M173, M70, Tat, M213, M9) in a final volume of 9.5 µL. After amplification, takes place a purification step with EXO-SAP-IT[®] (USB[®]). The minisequencing reaction (single base extension, SBE) with SNaPshot[®] Multiplex (Applied Biosystems) was performed and the final step was purification with SAP (USB[®]). DNA fragments separation, detection and identification was achieved with capillary electrophoresis using an ABI PRISM[®] Genetic Analyser 3130xl sequencer (Applied Biosystems).

3. Results

Fig. 1 represents the distribution of studied individuals in different haplogroups for each population.

4. Discussion and conclusions

Through the obtained results few differences between the studied African populations are shown, which reveals that this Y-SNPs panel is not useful for differentiation purposes within African populations. To be possible to differentiate male lineages among African populations it will be necessary to use more specific Y-SNP multiplex systems. In future, it is our intention to study not only other Y-SNP panels but also all Y-STR markers available in validated multiplex systems in our Lisboa immigrant populations previously inserted in Y-HRD database.

Conflict of interest

None.

Acknowledgments

None.

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