STUDY OF Y-SNPs GENETIC MARKERS WITH FORENSIC INTEREST AND ANCESTRY INFORMATIVE POWER IN PALOP'S IMMIGRANT POPULATIONS IN LISBOA

Ana Inácio[1,2], Heloísa Afonso Costa[1,6], Cláudia Vieira da Silva[1,6], Teresa Ribeiro[1,6], Maria João Porto[1,6], Jorge Costa Santos[1,3,4,6], Gilberto Igrejas[2], António Amorim[1,4,5,6]

1 - Serviço de Genética e Biologia Forenses, Delegação do Sul, Instituto Nacional de Medicina Legal e Ciências Forenses, Lisboa, Portugal
2 - Departamento de Genética e Biotecnologia, Universidade de Trás-os-Montes e Alto Douro, Vila Real, Portugal
3 - Faculdade de Medicina, Universidade de Lisboa, Lisboa, Portugal
4 - Instituto Superior de Ciências da Saúde Egas Moniz, Monte da Caparica, Portugal
5 - Faculdade de Ciências, Universidade de Lisboa, Lisboa, Portugal
6 - CENCIFOR - Centro de Ciências Forenses, Coimbra, Portugal

Introduction

Since the early 70's, 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 [1,2,3,4,5,6,7].

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 [8,9,10].

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.

Material and Methods

Sample collection and DNA extraction

We study 211 bloodstain samples, from those, 123 from Angola (immigrant population inserted at Y-HRD database with number YA003921), 61 from Guiné-Bissau (immigrant population inserted at Y-HRD database with number YA003922) and 27 from Moçambique, collected from immigrant individuals inhabitants of Lisboa metropolitan area, and undergoing forensic investigations in INMLCF. The number of studied individuals from each one of the different African countries - Angola, Guiné-Bissau, Moçambique - traduce the representation that each immigrant group has in Lisboa population.

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. DNA extraction was performed with Chelex® 100 [11].

DNA typing

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®), then 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 3130 xl sequencer (Applied Biosystems).

Results

The haplotypes of each individual was obtained in a electropherogram in which we verify the panel of the 9 Y-SNP markers. In figure 1 it's possible to observe the distribution of studied individuals in each haplogroup, for each population.

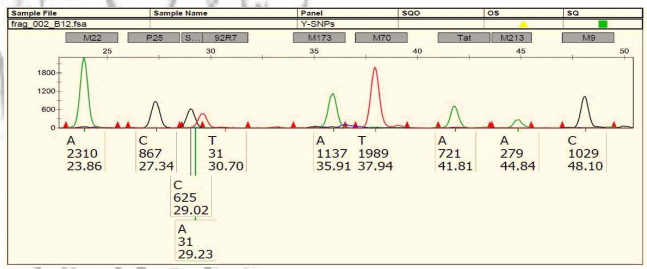

Figure 1 – Example of an electropherogram from an individual's haplotype with the various markers used in this study.

Through the haplotypes obtained for the three populations in study, it was possible to verify the existence of a few genetic differences between populations. In figure 2 we have the percentages of each haplogroup presented in each African population.

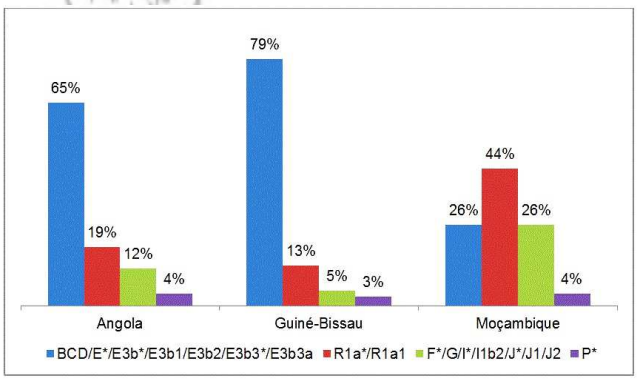

Figure 2 – Percentages of each haplogroup presented in each African population. In blue is presented the haplogroups BCD/E*/E3b*/E3b1/E3b2/E3b3*/E3b3a; in red the haplogroups R1a/R1a1; in green the haplogroups F*/G/I*I1b2/J*/J1/J2 and in purple the haplogroup P*.

Discussion/Conclusion

Through the results obtained we can confirm that those populations exhibit some differences between them. In African populations only a few differences are shown with the used markers in the present study, which reveals that this Y-SNPs panel isn't useful for differentiation purposes within African populations.

To be possible the differentiation among African populations it will be necessary to use more specific Y-SNP multiplex systems than the one used in our study.

Furthermore, we intend to study, not only with other Y-SNP panels but also with all Y-STR available; all immigrant populations previously studied and insert at Y-HRD database by our group - Angola (YA003921), Guiné-Bissau (YA003922), Cabo Verde (YA003906) and Moçambique (to be inserted) -.

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