MITOCHONDRIAL DNA CHARACTERIZATION OF GUINÉ-BISSAU IMMIGRANT POPULATION LIVING IN LISBOA

António Amorim [1,2,3,4], Marta Proença de Campos [1,2], Joana Ribeiro [1,2], Cláudia Vieira da Silva [1,4], Teresa Ribeiro [1,4], Maria João Porto [1,4], Heloísa Afonso Costa [1,4]

[1]Instituto Nacional de Medicina Legal e Ciências Forenses, Portugal; [2]Faculdade de Ciências da Universidade de Lisboa, Lisboa, Portugal; [3]Instituto Superior de Ciências da Saúde Egas Moniz, Monte de Caparica, Portugal
[4]CENCIFOR – Centro de Ciências Forenses, Coimbra, Portugal

INTRODUCTION

Guiné-Bissau, officially known as the Republic of Guiné-Bissau, is a country on the west coast of the African Continent. Colonized in the nineteenth century, Guiné-Bissau was the first Portuguese colony in Africa with independence recognized by Portugal. Before the arrival of Europeans and until the seventeenth century, almost all of the territory of Guiné-Bissau was part of the Kingdom of Gabu, part of the Mali Empire.

In the late seventies, migration flows, related to the post-colonial phase, led to Guinean community became the sixth largest immigrant community in Portugal. Most of those immigrants are based in Lisboa metropolitan region. Those individuals have contributed to increased heterogeneity in social, cultural, religious, linguistic and anthropological frame in Portugal.

In recent decades the mtDNA became an important genetic marker in human population studies as well as in forensic studies. The interest in the analysis of mtDNA is explained by the characteristics of the molecule, as high copy number per cell in the same individual, maternal inheritance, absence of recombination and high mutation rate.

Therefore, unlike nuclear DNA, mtDNA does not get shuffled every generation, so it is presumed to change at a slower rate, which is useful for the study of human evolution.

The main goal of our study was to determine the haplotypes and respective haplogroups of Guineans individuals living in Lisboa, to be possible to obtain the mtDNA genetic structure of the population under study and evaluate the impact of immigrant individuals in the Lisboa region.

MATERIALS AND METHODS

In the present study, the entire control region mtDNA was sequenced and analyzed, from blood samples of 50 individuals from Guiné-Bissau living in Lisboa.

DNA extraction
Chelex®100 method [3]

mtDNA total control region amplification
Primers L15971/H016 and Primers L16555/H639

Purification of amplified product
ExoSap-IT® (Affymetrix)

Direct and reverse sequencing
Big Dye® Terminator v.3.1 Cycle Sequence (AB)

Purification of sequenced product
BigDye® XTerminator Purification Kit (AB)

Detection of sequenced product
Genetic Analyser 3130® (*ABI Prism®)*

Sequence analysis and comparison with rCRS [4,5,6]
Sequencing Analysis v.5.2 (AB) e *SeqScape* v.3 (AB)

Haplogroup determination
Phylotree bluid 16 [7]

RESULTS

From the 50 individuals analysed, 45 different haplotypes were identified. Out of 45 haplotypes, 28 diferentnt haplogroups were determined, 41 haplotypes belong to macrohaplogroup L (characteristic of African regions); 3 to haplogroup U6 and 1 to haplogroup M1 (figure 1).

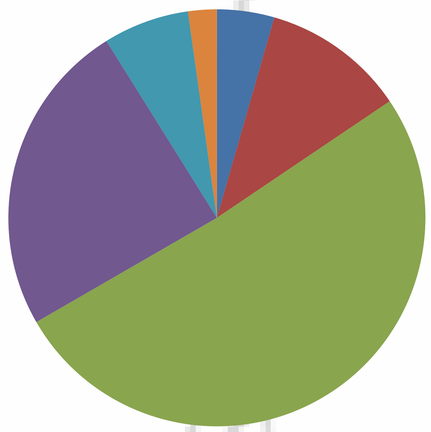

Figure 1
Schematic representation of the percentages of haplogroups for the 50 studied individuals

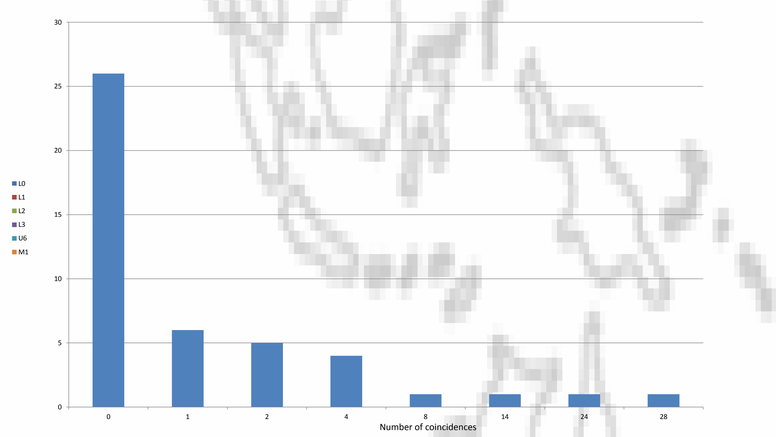

Figure 2 - Number of coincidences of haplotypes from studied population on EMPOP[8] forensic data

According to EMPOPall_R11 filter[8] there are no mutations in this samples unobserved in the set of EMPOP data.

It was verified that 26 haplotypes from our sampling had no coincidence on EMPOP Forensic data (figure 2). Still, the most common haplotype showed the highest number of coincidences and belong to haplogroup L1.

CONCLUSION

From our results, it is possible to confirm that this population of Guiné-Bissau living in Lisboa presents a high number of unique haplotypes, most of them belonging to haplogroups from Sub-Saharan regions of Africa. The results obtained in our study suggest that the genetic composition of Guiné-Bissau immigrant population presents a high genetic diversity. Those haplogroups, typical of Sub-Saharan region, will probably, in future, introduce genetic diversity in Lisboa population.

REFERENCES

[1] –Pordata, Outubro de 2013, Disponível em: www.pordata.pt
[2] –Dias P, Machado R, Estrela J, Bento AR. Relatórios de imigração, fronteiras e asilo, Oeiras, Serviços de Estrangeiros e Fronteiras, 2012
[3] – Walsh PS, Metzger DA, Higuchi R. Chelex® 100 as a medium for simple extraction os DNA for PCR-based typing from forensic material, Biotechniques 1991, 10(4);506-13
[4] – Anderson S, Bankier AT, Barrel BG, De Bruijn MH, Couson AR, Drouin J, et al. Sequence and organization of the human mitochondrial genome, Nature 1981,290:457-65
[5]– Andrews RM, Kubacka I, Chinnery PF, ligtowlers RN, Turnbull DM, Howell N. Reanalysis and revision of the Cmbridge reference sequence for human mitochondrial DNA. Nat Genet 1999; 23(2):147
[6]– Carracedo A, Bär W, Lincoln P, Mayr W, Morling N, Olasein B, et al. DNA Comission of the international society for forensic genetics: guidelines for mitochondrial DNA typing. Forensic Sci Int 200;110(2):79-85.
[7] – van Oven M, Kayser M. 2009. Updated comprehensive phylogenetic tree of global human mitochondrial DNA variation. Hum Mutat 30(2):E386-E394
[8] – Parson W, Dür A . EMPOP- a forensic mtDNA Database. Forensic Sci Int : Genetics. 2007; 1(2): 88-92