# Mitochondrial DNA studies of Lisbon immigrants from Portuguese Speaking African Countries: bioinformatics contributions

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## Introduction

Since the end of the 1970s Portugal had an important role in migratory movements, becoming a destiny for immigrants of a wide range of nationalities, mainly from African countries. According to PORDATA, until the end of 2014 there were approximately 40,000 immigrants from Cape Verde, 20,000 immigrants from Angola, 18,000 immigrants from Guinea-Bissau and 3,000 immigrants from Mozambique living in Portugal, and from those, more than 80% living in Lisbon region [1]. This reality can be one of the main contributors for genetic variation of Lisbon population at the present and in the future.

Mitochondrial DNA (mtDNA) has certain features that make it desirable for forensics, namely, high copy number, lack of recombination, and matrilineal inheritance. These mtDNA features are also important in evolutionary and population studies [2,3].

We aim to characterize mtDNA of immigrants from Portuguese Speaking African Countries (PALOP) living in Lisbon and their potential contribution to genetic variation of Lisbon population.

## Material and Methods

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| **DNA extraction of blood samples: Chelex®100 method [4]** | **Sequence analysis: Sequencing Analysis v.5.2 (AB) [5,6]** |
| **mtDNA total control Region amplification: Primers L15971/H016 and Primers L16555/H639** | **Sequence comparison with rCRS: SeqScape v.3 (AB) [5,6]** |
| **Purification of amplified product: ExoSap-IT®** | **Haplogroup determination: Phylotree build 17 [7]** |
| **Direct and Reverse sequencing: BigDye® Terminator v.3.1 Cycle Sequence (AB)** | **Genetic distances calculation: Arlequin software ver.3.5** |
| **Purification of sequenced product: BigDye® XTerminator Purification Kit (AB)** | **Graphic/Tree representation: PHYLIP - Phylogeny Inference Package ver.3.2.** |
| **Detection of sequenced product: Genetic Analyser 3130® (ABI Prism®)** | **Genetic distances and Tree representation: VEME Workshop 2017 methods** |

## Results

For each sample, the complete sequence of the total control region was obtained. The comparison of the obtained sequences with the Revised Cambridge Reference Sequence (rCRS), among the 508 analysed individuals, allowed the identification of 261 different haplotypes, 253 of which are unique. The other haplotypes were present in more than one individual. Between the 261 haplotypes, we determined 114 different haplogroups. The most common haplogroup is the haplogroup L with 160 haplotypes. We also found that the haplogroup H has 20 haplotypes and the haplogroup U and K have 10 and 8 haplotypes, respectively. The haplogroup J, M and T appear only once. In Fig.1 we present haplogroups and correspondent haplotypes determined in some of studied African immigrants.

At Fig. 2, the graphical representation of calculated genetic distances between all populations shows the establishment of two distinct groups: one composed by the Portuguese population and another, more distant, by the African populations, as expected. Comparing the different immigrant populations living in Lisbon, the genetically closest community of Portuguese population is Mozambique and the furthest is Cape Verde, followed by Guinea-Bissau and Angola.

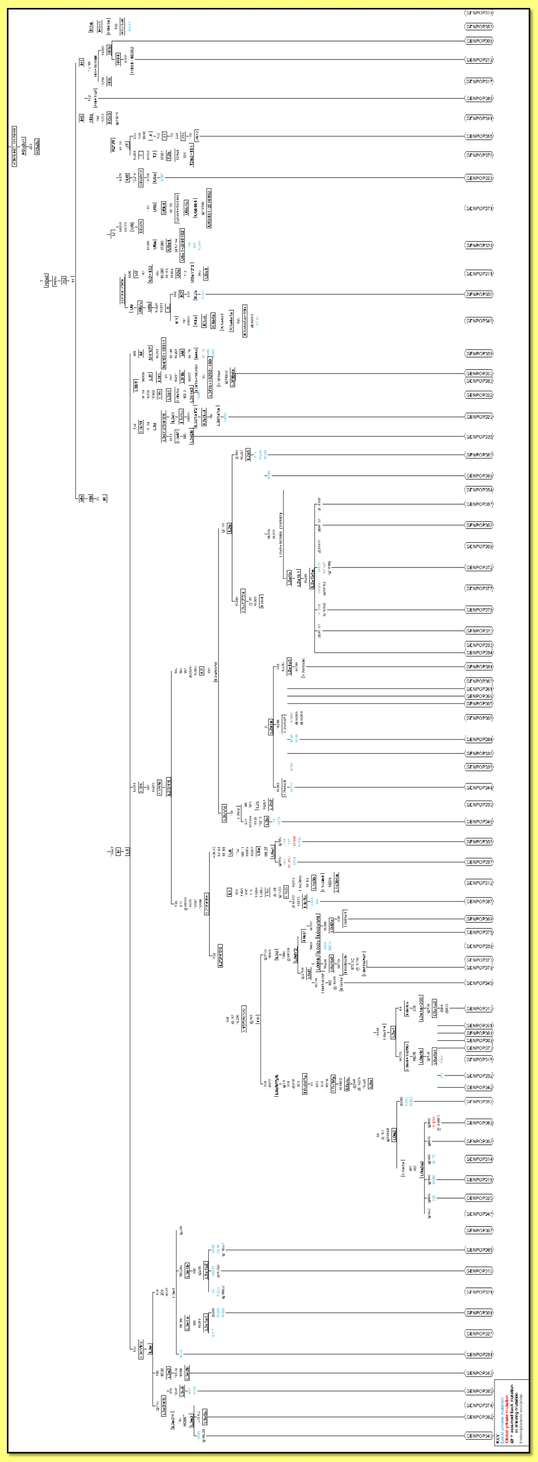

Figure 1 - Graphical presentation of all studied haplotypes and correspondent haplogroups.

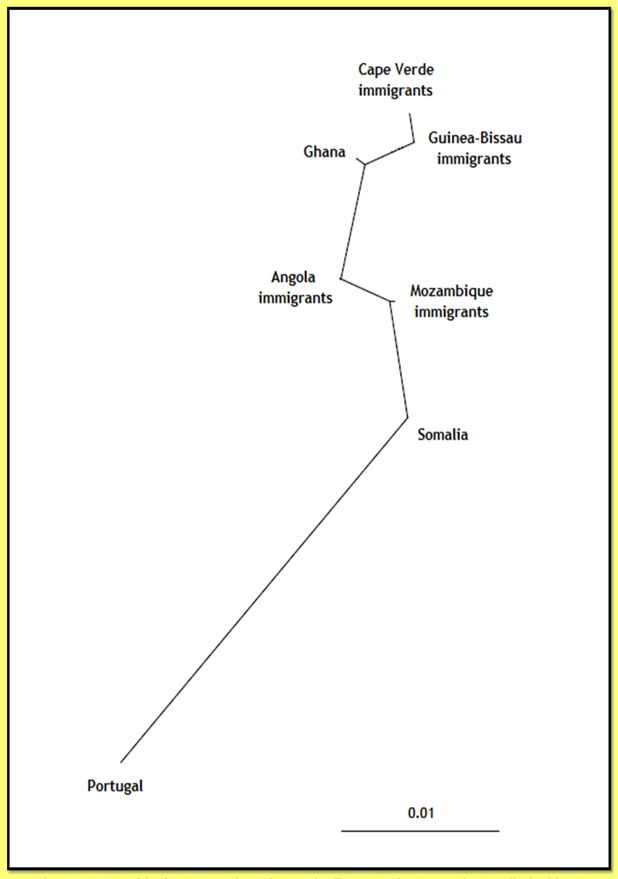


Figure 2 - Graphical presentation of genetic distances between the studied African immigrant populations and other selected populations including Lisboa population (Portugal).

## Discussion and Conclusions

PALOP immigrants present high number of unique haplotypes, most of them belonging to haplogroups L, R, J, H, U and M from Sub-Saharan regions of Africa. Haplogroup L was the most prevalent. This haplogroup is uncommon in European and Portuguese populations.

Our results indicate that the PALOP immigrants introduce high genetic diversity in Lisbon population not only due to high number of unique haplotypes of the studied populations but regarding to high genetic distances between all studied populations as show in the presented phylogenetic tree representative of genetic distances.

Our results suggests that the integration of African immigrants among Lisbon population is going to lead to an increase in the genetic variability in this region. Therefore, it is very important to study the immigrant communities currently living in Lisbon.

## References

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