# Genetic portrait of Lisboa immigrant population from Mozambique with mitochondrial DNA

M. Proença de Campos [(1)], H. Afonso Costa [(2,3)], C. Vieira da Silva [(2,3)], T. Ribeiro [(2,3)], M. J. Porto [(2,3)], A. Amorim [(2,3)]

(1) Faculdade de Ciências da Universidade de Lisboa, Lisboa, Portugal; (2) Instituto Nacional de Medicina Legal e Ciências Forenses, Portugal; (3) CENCIFOR – Centro de Ciências Forenses, Coimbra, Portugal

## Introduction

Since the end of the 1970s, Portugal had a role in the migratory movements, becoming a destiny for immigrants from a wide range of nationalities, especially from the African continent. According to statistical data, until the end of 2014, there were approximately 99 000 African immigrants living in Portugal. The Mozambican community is about 3 000 individuals, of which more than a half are currently living in Lisbon metropolitan region [1].

Mitochondrial DNA (mtDNA) is a useful tool, not only in evolutionary and population studies, but also in forensic genetics. Due to its unique features, including maternal inheritance, lack of recombination, high copy number and high mutation rate, mtDNA has been used to measure the genetic impact of human migration and also for human identity testing [2,3]. The analysis of mtDNA allows to trace the ancestral origin of each population, revealing the evolutionary history of maternal lineages across several generations.

The aim of our study is the genetic characterization of Mozambican immigrants living in Lisbon, in order to emphasize their genetic contribution to Lisbon population. We also pretend to classify haplotypes into haplogroups and to compare this population with other African populations.

## Material and Methods

| |
|---|
| **DNA extraction of blood samples:** **Chelex®100 method [4]** |
| **mtDNA total control Region amplification:** **Primers L15971/H016 and Primers L16555/H639** |
| **Purification of amplified product:** **ExoSap-IT®** |
| **Direct and Reverse sequencing:** **BigDye® 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 [5,6]** **Sequencing Analysis v.5.2 (AB) e SeqScape v.3 (AB)** |
| **Haplogroup determination:** **Phylotree build 17 [7]** |

## Results

For each sample we sequenced the total control region using the forward and reverse primers. In order to define haplotypes we compared our sequences with the Revised Cambridge Reference Sequence (rCRS). Among the 82 analysed individuals, 72 different haplotypes were identified, 65 of which are unique. The other 7 haplotypes were present in more than one individual. Inside the 72 haplotypes, we determined 49 different haplogroups. The most common haplogroup is the haplogroup L with 66 haplotypes. We also found that the haplogroup H has 7 haplotypes and the haplogroup U and K have 4 and 2 haplotypes, respectively. The haplogroup J, M and T appear only once (Fig.1).

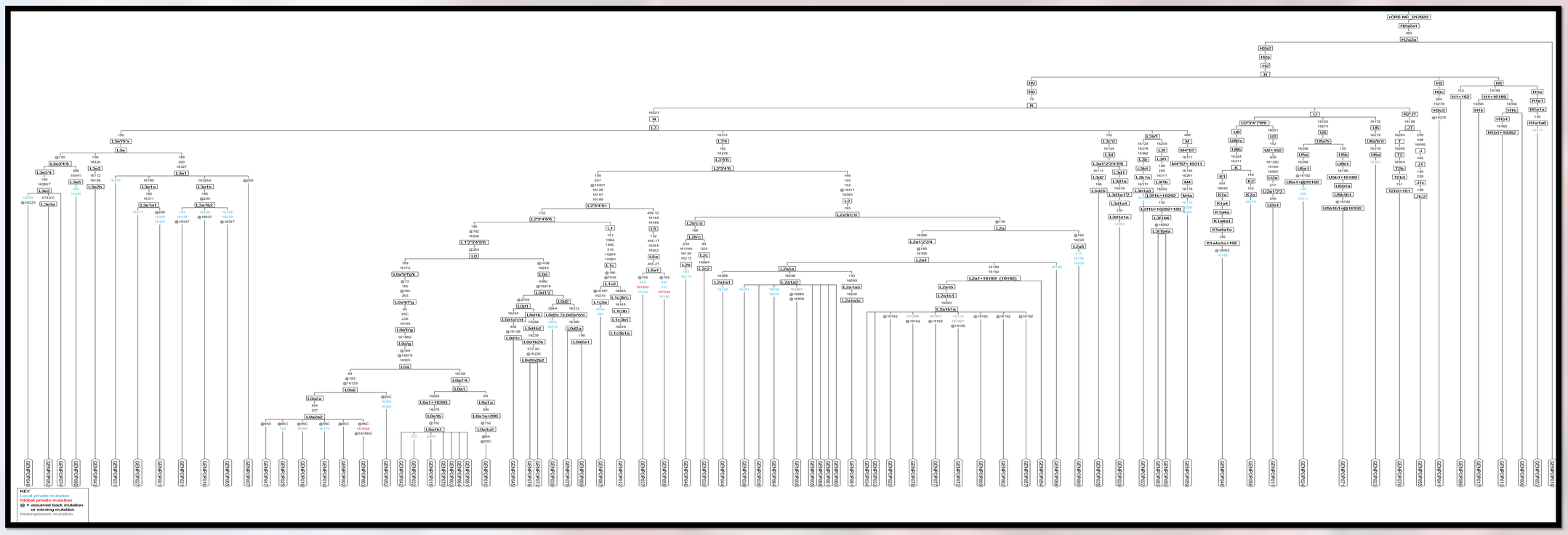


Figure 1 - Graphical presentation of all studied haplotypes and correspondent haplogroups.

## Conclusion

The studied population shows a great genetic variability due to the high frequency of unique haplotypes. The majority of haplotypes fit into the macrohaplogroup L, very characteristic of Sub-Saharan region of Africa, where Mozambique is framed. This result evidences the importance of studying this Mozambican community living in Lisbon, once it demonstrates the genetic diversity that this immigrant population will introduce into the Lisbon population.

## References

[1] PORDATA, Mach 2016. Available in: www.pordata.pt.

[2] M. Ingman, H. Kaessmann, S. Pääbo, and U. Gyllensten, "Mitochondrial genome variation and the origin of modern humans.," *Nature*, vol. 408, no. 6813, pp. 708–713, 2000.

[3] J. C. Alvarez, D. L. E. Johnson, J. a. Lorente, E. Martinez-espin, L. J. Martinez-Gonzalez, M. Allard, M. R. Wilson, and B. Budowle, "Characterization of human control region sequences for Spanish individuals in a forensic mtDNA data set," *Leg. Med.*, vol. 9, no. 6, pp. 293–304, 2007.

[4] P. S. Walsh, D. a. Metzger, and R. Higuchi, "Biotechniques 30th anniversary gem Chelex 100 as a medium for simple extraction of DNA for PCR-based typing from forensic material," *Biotechniques*, vol. 54, no. 3, pp. 506–513, 2013.

[5] R. M. Andrews, I. Kubacka, P. F. Chinnery, R. N. Lightowlers, D. M. Turnbull, and N. Howell, "Reanalysis and revision of the Cambridge reference sequence for human mitochondrial DNA.," *Nat. Genet.*, vol. 23, no. 2, p. 147, 1999.

[6] W. Bär, B. Brinkmann, B. Budowle, a Carracedo, P. Gill, M. Holland, P. J. Lincoln, W. Mayr, N. Morling, B. Olaisen, P. M. Schneider, G. Tully, and M. Wilson, "Guidelines for mitochondrial DNA typing. DNA Commission of the International Society for Forensic Genetics.," *Vox Sang.*, vol. 79, no. 2, pp. 121–125, 2000.

[7] M. van Oven and M. Kayser, "Updated comprehensive phylogenetic tree of global human mitochondrial DNA variation.," *Hum. Mutat.*, vol. 30, no. 2, pp. 386–394, 2009.

7th International EMPOP meeting, Belin, Germany - 2016