Mitochondrial DNA data of Cabo Verde Immigrant Population Living in Lisboa

Afonso Costa H[1,2,*], Morais P[1,3], Amorim A[1,2,4], Vieira Silva C[1,2], Matos S[1,3], Marques Santos R[1,2], Espinheira R[1,2], Costa Santos J[5,2,6]

[1]Servico de Genética e Biologia Forense, Delegação do Sul do Instituto Nacional de Medicina Legal e Ciências Forenses, I.P., Lisboa, Portugal
[2]CENCIFOR - Centro de Ciências Forenses, Coimbra, Portugal
[3]Biologia Molecular em Saúde, Escola Superior de Saúde Egas Moniz, Almada, Portugal
[4]Antropologia Forense, Faculdade de Ciências e Tecnologia da Universidade de Coimbra, Coimbra, Portugal
[5]Servico de Clinica Forense, Delegação do Sul do Instituto Nacional de Medicina Legal e Ciências Forenses, I.P., Lisboa, Portugal
[6]Medicina Legal e Ciências Forenses, Faculdade de Medicina da Universidade de Lisboa, Lisboa, Portugal

Introduction

Mitochondrial DNA (mtDNA) analysis found an important role in forensic genetics, especially when nuclear DNA analysis does not give a conclusive response. It is a powerful tool to exclude samples as originating from the same matriline. Features that increase the vested interest of mtDNA are the high copy number per cell, maternal inheritance [1], absence of recombination [2], and high mutation rate. Due to higher overall mutation rate, control region is comparatively enriched in sequence variation and therefore its analysis is important to establish haplotypes and haplogroups. Haplogroup assignment became noteworthy to clarify the history and demographic past of a population.

As well as occurs all over Europe, in Portugal, and particularly in Lisboa, immigrant populations are increasing. The Instituto Nacional de Medicina Legal e Ciências Forenses is carrying out a comprehensive genetic study with the aim of portraying the genetic diversity of the immigrants who live in Lisboa [3, 4, 5]. Within that objective the present study intends to: obtain the mtDNA variability of Cabo Verde Immigrant Population Living in Lisboa and classify haplotypes into haplogroups.

Material and Methods

Blood samples were obtained from twenty four, Cabo Verde immigrants living in Lisboa. DNA was extracted using Chelex® 100 method [6]. PCR amplification was performed with 2 pairs of primers L15971/ H017 and L16450/ H599 in a primer mix using QIAGEN® Multiplex PCR Master Mix in a final volume of 10 µl. Thermocycling conditions were performed in a GenAmp® PCR system 9700 (Applied Biosystems). The cycle sequencing was performed using the ABI Prism® BigDye® Terminator v.3.1 Cycle Sequence Kit (Applied Biosystems); BetterBuffer (Microzone Limited, Sussek, UK) has been incorporated into the sequencing procedure. Before DNA analysis a simple bead purification method (XTerminator/SAM) was made, to remove the unincorporated BigDye terminators, unnecessary salts, and unused diluent buffer. The sequences were analysed in the sequencer 3130 – Genetic Analyser (ABI PRISM®) with the ABI DNA Sequencing Analysis Software v.5.2 and the SeqScape® Software v.2.5. The obtained haplotypes were compared with the Cambridge Reference Sequence (CRS) [7] and typed following the nomenclature recommendations of the IUPAC [8]. Haplogroups were determined on a Web-based tool for management and quality analysis of mitochondrial DNA [9].

Results and discussion

All twenty four samples were successfully typed, all being unique. It was possible to include 100% of the mtDNA sequences into a specific mtDNA haplogroup according to nucleotide substitutions of the non-coding region (figure 1). The majority of mtDNA sequences were included into specific African mtDNA haplogroups and a minority, 8.3% of mtDNA lineages, belongs to West Eurasian haplogroups. The haplogroups determined are in accordance with historical colonization that took place with caucasian Portuguese and African slaves.

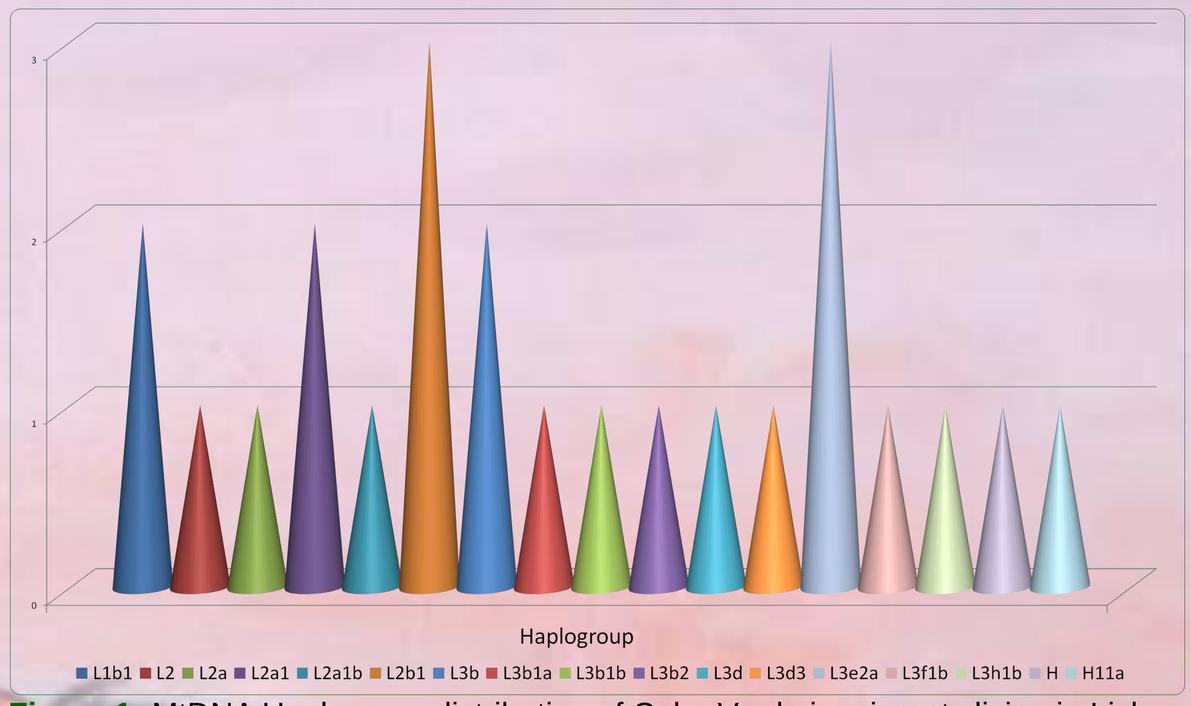

Figure 1: MtDNA Haplogrups distribution of Cabo Verde immigrants living in Lisboa.

Conclusions

The studied population shows a great interpopulation genetic variability due to the high frequency of unique haplotypes. Cabo Verde immigrants living in Lisboa exhibit haplotypes that belong to haplogroups observed in native Africans and in West Eurasian. MtDNA control region typing is extremely useful as a technique to differentiate among degraded samples frequently found in forensic genetics and to establish its global frequency when having knowledge of the genetic structure of populations.

References

[1] R.E. Giles, et al., Proc. Natl. Acad. Sci. 77 (1980) 6715–6719.
[2] J.W. Ballard, M. O. Whitlock, Mol. Ecol. 13 (4) (2004) 729–744.
[3] A. Amorim, et al., Forensic Sci Int Genetics, doi:10.1016/j.fsigen.2012.05.006.
[4] A. Amorim, et al., Forensic Sci Int Genetics 6 (2012) e121–e124.
[5] R. Marques dos Santos, et al., Forensic Sci Int Genetics, doi:10.1016/j.fsigen.2012.05.010.
[6] P.S. Walsh, D.A. Metzger, R. Higuchi, BioTechniques 10 (1991) 506-513.
[7] S. Anderson, A.T. Bankier, B.G. Barrel, et al. Nature 1981; 290: 457-465.
[8] A. Carracedo, W. Bär, P. Lincoln, et al. Forensic Sci Int 2000; 110(2): 79-85.
[9] H.Y. Lee, I. Song, et al. BMC Bioinformatics 2008; 9: 483.

"DNA in Forensics: Exploring the Phylogenies"
September 06-08, Innsbruck, Austria