LISBON POPULATION MTDNA ANALYSIS: STUDY OF A GENETIC MARKER WITH POPULATION, EVOLUTION AND FORENSIC INTEREST

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INTRODUCTION

Mitochondrial DNA (mtDNA) is remarkable in population genetic studies to clarify the history and demographic past of human populations. For forensic purposes mtDNA analysis is particularly useful when there are degraded or low level DNA samples. Nonetheless, only being in possession of the genetic reference data of the populations it is possible to attribute statistical weight to an evidence in forensic casework.

The National Institute of Legal Medicine and Forensic Sciences in Portugal carried out a study with the aim of portraying the genetic diversity of immigrants living in Lisbon. However, the nonimmigrant population of Lisbon, the reference population, had not yet been studied. The present study intends to determine the mtDNA variability of the Lisbon metropolitan native population.

MATERIALS AND METHODS

A total of 90 unrelated individuals were sampled. The entire control region of the mtDNA was amplified with primers L15971 and H639 and sequenced with BigDye® Terminator v.3.1 Cycle Sequence and primers L15971, L16555, H016 and H639. Sequences were detected in a 3130 Genetic Analyzer. Results were analyzed and compared with revised Cambridge Reference Sequence in SeqScape v.3. The frequency of haplotypes was calculated by direct counting using EMPOP database.

DNA extraction
Chelex®100 method [1]

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 [2, 3, 4]
Sequencing Analysis v.5.2 (AB) e *SeqScape* v.3 (AB)

Haplogroup determination
Phylotree bluid 16 [5]

RESULTS AND CONCLUSION

Among the 90 samples, 87 unique mtDNA haplotypes were identified. It was verified that 32 of the haplotypes had no coincidence and 14 of the haplotype had only one coincidence on EMPOP database. This low frequency of the majority of the haplotypes emphasizes the importance of studying this population. The most frequent haplotypes, encloses haplogroups H1, HV, K1, R0 and U5, that belong predominantly to west Eurasian mtDNA. Haplotypes that belong to Haplogroups L0, L1, L2, L3 were also identified in Lisbon population (figure 1).

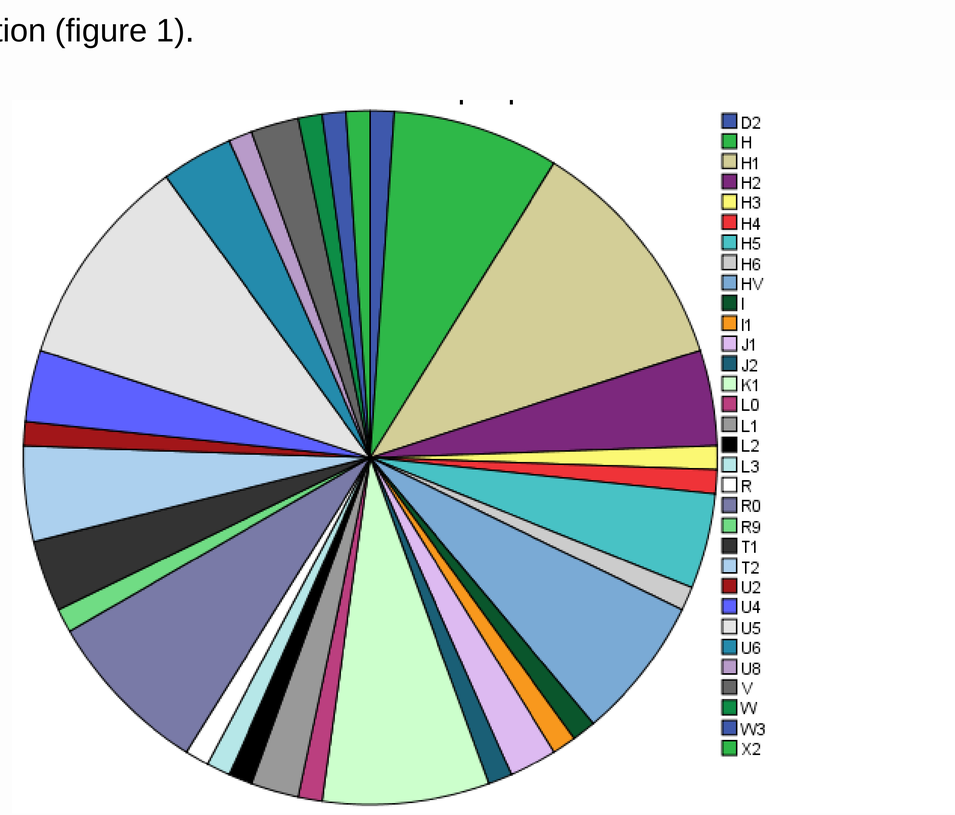

Figure 1
Schematic representation of the percentages of haplogroups for the 90 studied individuals

According to EMPOPall_R11 filter [6] there are no mutations in this samples unobserved in the set of EMPOP data.

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