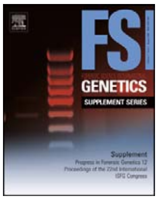
Forensic Science International: Genetics Supplement Series 2 (2009) 329–330

Contents lists available at ScienceDirect

Forensic Science International: Genetics Supplement Series

journal homepage: www.elsevier.com/locate/FSIGSS



Research article

Common mitochondrial DNA haplogroups observed in an argentine population database sample

Laura Catelli [a,*], Carola Romanini [a], Alicia Borosky [b], Mercedes Salado Puerto [a], Lourdes Prieto [c], Carlos Vullo [b]

[a] EAAF, Rivadavia 2443, 2P Of 3-4, Buenos Aires, Argentina
[b] L.I.D.M.O., Independencia 644 – 4ª, Córdoba, Argentina
[c] Instituto Universitario de Investigación en Ciencias Policiales (IUICP), Comisaría General de Policía Científica, Sección de Biología-ADN, Madrid, Spain

ARTICLE INFO

Article history:
Received 1 September 2009
Accepted 4 September 2009

Keywords:
mtDNA
HVI
HVII
Haplotypes
Haplogroups

ABSTRACT

Mitochondrial DNA hypervariable regions I and II were sequenced from 403 unrelated Argentine individuals. The aim of this study was to create a population database as well as to identify the population diversity for this genetic marker by classifying it into haplogroups.

The sequence polymorphisms of the HVI and HVII regions were determined by PCR and direct sequencing. The haplotypes found were checked by phylogenetic haplogroup analysis to decrease haplotype assignation errors and to avoid artificial recombination.

We found 78 different haplogroups in this set of samples. A high percentage of haplotypes (53%) belong to European haplogroups due to the large flow of European immigrants from colonial times. However, we also observed a high percentage of haplotypes that belong to Amerindian haplogroups (39%), which were conserved through the female Amerindian population contribution. Furthermore, we found a small group of haplotypes with Sub-Saharan African origin (3.5%) due to the slave trade at the beginning of Argentina's colonization.

The sequences found showed that this set of samples has an abundant haplogroup diversity because of the European and Amerindian ethnic group contribution.

1. Introduction and objectives

Mitochondrial DNA analysis is a powerful tool in the forensic area due to some of its characteristics, such as the high polymorphism observed in the hypervariable regions, the fact that mtDNA does not undergo recombination and the high copy numbers per cell. But its discrimination power depends on the population frequency of a particular sequence; thus, it is essential to establish a mitochondrial reference database. Moreover, the generation of this database will allow us to know some aspects of the phylogenetic composition and origin of the population studied.

The aim of this study was to analyze the hypervariable regions of mitochondrial DNA (HVI and HVII) of unrelated individuals from the Argentine population and to generate a database from the results. This database can be useful to interpret aspects related to the genetic diversity of the Argentine population by having data on which are the haplogroups most commonly found in this population.

2. Materials and methods

A total of 403 blood stain samples from unrelated individuals from Argentina were studied. DNA was isolated by using the modified salting out method [1]. Both HVI and HVII mitochondrial DNA regions were amplified using primers 15997F, 16401R, 048F and 408R [2]. Sequencing reactions were completed by using the Big Dye Terminator 3.1 kit (Applied Biosystems) and both strands of each fragment were sequenced. The analysis of the sequences was done using Sequencher 4.8 software.

Recommendations from Refs. [3–6] were followed to establish haplogroup designation.

3. Results and conclusions

The analysis of mitochondrial DNA hypervariable regions (HVI/HV2) performed on 403 samples revealed a great diversity of haplotypes in the Argentine population. Table 1 shows 287 different haplotypes found in this sampling. These haplotypes were classified as belonging to 78 haplogroups and sub-haplogroups.

Only 17 samples (4.5%) were not classified in haplogroups. The sequences found in these samples were confirmed through a second analysis in order to rule out the possibility of artificial recombination.

---

[*] Corresponding author.
E-mail address: lidmo-forense@datamarkets.com.ar (L. Catelli).

1875-1768/$ – see front matter © 2009 Elsevier Ireland Ltd. All rights reserved.
doi:10.1016/j.fsigss.2009.09.016

Table 1
Haplogroups and sub-haplogroups observed in the Argentine population sample.

Hgs	*n* samples[a]	*n* haplotypes[b]	Hgs	*n* samples	*n* haplotypes	Hgs	*n* samples	*n* haplotypes
L0a1b	1	1	U1	1	1	J2	1	1
L1b	3	3	U2e1	1	1	J2a1	2	2
L2a1a3	1	1	U3	2	2	J2b	2	2
L2a1c1	1	1	U4	1	1	T	5	5
L2c2	1	1	U4a1	1	1	T1a	8	3
L3b	2	1	U4a2	1	1	T2	6	6
L3e1a	1	1	U5a	11	11	T2b	4	4
L3e2b	1	1	U5b	5	5	X2e1a1a	1	1
L3e4	1	1	U5b1	1	1	I	1	1
L3f1b1	2	2	U5b1c	1	1	W	5	5
M1	1	1	U6a	1	1	W1b	1	1
M7a1b	1	1	U6c	1	1	A	2	2
N1b	2	2	U8b	1	1	A2	34	17
R0	1	1	K	8	8	A2a	5	3
R0a2a	1	1	K1a1a	1	1	A4	3	3
HV0	7	5	K1a1b1	5	3	C1	35	25
HV0b′c	2	2	K1b2	1	1	C1a	2	2
HV1	2	2	K2	1	1	C1d	7	4
H	83	54	K2a	6	2	B4	29	7
H1a	3	3	J	1	1	B4a′g	1	1
H1b	2	2	J1	1	1	D	1	1
H2a1	1	1	J1a′b′e	1	1	D1	36	20
H2a2b1	3	2	J1b	1	1	D4b1	1	1
H2a3	1	1	J1b1	1	1	D4h3	2	2
H5	3	2	J1c	4	4	?[c]	17	17
H6	2	1	J1c1	2	2			
H11	1	1	J1c7	1	1			
Total							403	287

[a] Number of samples belonging to each haplogroup.
[b] Number of different haplotypes.
[c] Undetermined haplogroup.

Haplotypes: The most frequent haplotype (263G, 315.1C – sometimes with 309.1C and 309.2C), which belongs to the European haplogroup H, was observed 15 times. Furthermore, a low variability was observed among haplotypes belonging to haplogroup H (only 67 different haplotypes were observed in 99 samples belonging to haplogroup H) in comparison with haplogroup U which showed a high variability (all the 28 haplotypes belonging to haplogroup U were different). Native American haplotype variability was similar among haplogroups with no significant prevalence.

Haplogroups: Among the most frequent haplogroups observed were those from European origin (RO-HV-H-I-J-K-T-U-W-X) (53%) and from Native American origin (A-B-C-D) (39%), which shows that the extant Argentine population is represented mainly by an admixture of European and Native American origin. There was only a small number of haplotypes belonging to African (L) and Asian origin (M, N) (see Fig. 1).

By analyzing the data obtained from the sequencing of the mitochondrial DNA HVI and HVII regions, it can be concluded that the Argentine population has a wide diversity of haplotypes from different origins. This coincides with the colonization that occurred in this country leading to the occurrence of a high percentage of European haplotypes as a result of two immigration waves; the first one, occurring during colonization times, was mainly from Hispanic origin but the second one, which occurred at the beginning of the 20th century, had a component mainly from Hispanic and Italian origin and also from other European areas. Moreover, the haplotypes that belong to Native American haplogroups are also frequently found due to the female contribution of the local indigenous population. Haplotypes of African origin are attributed to the slave trade which took place during earlier colonization times.

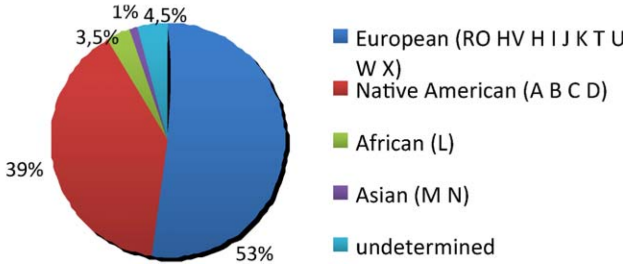


Fig. 1. Origin of haplogroups found in the Argentine population.

Apart from its contribution to the knowledge of the genetic composition and the origin of the Argentine population, this study represents an important tool for the statistical interpretation of the results obtained from the mitochondrial DNA analysis.

Conflict of interest

None.

Role of funding

Equipo Argentino de Antropología Forense (EAAF) and LIDMO provided the financial support for this article. The funding source had no involvement in study design.

Acknowledgements

The authors wish to thank Meredith Moon and Marcela Villar for providing language help and writing assistance.

References

[1] A.S. Miller, D.D. Dykes, H.F. Polesky, A simple salting out procedure for extracting DNA for human nucleated cells, Nucleic Acids Res. 16 (1988) 1215.
[2] M.R. Wilson, J.A. DiZinno, D. Polanskey, J. Replogle, B. Budowle, Validation of mitochondrial DNA sequencing for forensic casework analysis, Int. J. Legal Med. 108 (1995) 68–74.
[3] http://www.phylotree.org—Global mitochondrial DNA tree.
[4] S. Finnilä, M. Lehtonen, K. Majamaa, Phylogenetic network for European mtDNA, Am. J. Hum. Genet. 68 (2001) 1475–1484.
[5] A. Salas, H.J. Bandelt, V. Macaulay, M.B. Richards, Phylogoegraphic investigations: the role of trees in forensic genetics, Forensic Sci. Int. 168 (1) (2007) 1–13.
[6] W. Parson, A. Dür, EMPOP—a forensic mtDNA database (2007) 88–92.