



Relevant genetic contribution of Amerindian to the extant population of Argentina

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Abstract. The extant population of Argentina claims to be the most European country of Latin America. The statement has been held for over one century, and started when the Campaign Against the Desert took place in 1870. This military mission was directed against the aboriginal communities in order to obtain their possessions. No scientific approach was available, till recent, for checking the supposed to be pure European ancestries. Uniparentally inherited genetic markers, such as mitochondrial DNA (mtDNA) and Single Nucleotide Polymorphisms (SNP) located in the non-recombinant regions of the Y-chromosome may clarify the strict European ancestry of the nowadays population of Argentina. A set of 325 unrelated males were chosen to evaluate if Amerindian markers such as the mitochondrial haplogroups A, B, C and D, as well as the C to T transition in locus DYS199 of the Y-chromosome were present. Its detection in the tested individuals might suggest an ancestral contribution of Amerindian lineages. Hypervariable Regions (HVR) I and II were sequenced by Big Dye termination approach and detected in automated platforms (ABI, 310 and 3100Avant). DYS199 C to T transition detection was carried out by primer specific PCR. Over 50% of the individuals tested carried either mtDNA or Y Amerindian markers, 10% both, 20% were of Amerindian patrilineage and less than 40% denoted non-Amerindian contribution in the uniparentally inherited markers. By this simple approach a different contribution can be suggested within the most European country of Latin America. © 2005 Published by Elsevier B.V.

Keywords: Amerindian population; Uniparentally inherited genetic marker

1. Introduction

The analysis of uniparentally inherited genetic markers included in the non-recombining linkage groups of the Y-chromosome and the sequences of the mitochondrial

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Table 1

	mtDNA-Amer/Y-Amer		mtDNA-Amer		Y-Amer		mtDNA-no Amer/Y-no Amer	
	N	%	N	%	N	%	N	%
Northeastern	14	13.7	52	51.0	7	6.9	29	28.4
Center	13	10.6	48	39.0	7	5.7	55	44.7
Southwestern	13	13.0	47	47.0	4	4.0	36	36.0
Total	40	12.3	147	45.2	18	5.6	120	36.9

DNA have shown to be efficient molecular tools for tracing back the origin of the extant populations.

Amerindian lineages are well characterized by specific Y-chromosome polymorphisms, with reduced mutation rate, such as the locus DYS199 able to discriminate between people of Amerindian ancestry and others with different ancestries (Caucasian or African). A C→T transition defines the Amerindian haplogroup called M3-Q3*. The analysis of this marker may identify Amerindian patrilineages.

On the other hand, in the mitochondrial DNA in the Hypervariable Regions I and II (HVRI and HVR II) in the non-coding D-Loop it is also possible to identify ethnic specific sequences motif defined as Haplogroups A, B, C and D, all four characteristics of the Amerindian lineages.

Accordingly, in order to evaluate the aboriginal contribution to the extant population of Argentina either Y-chromosome specific or mtDNA Amerindian haplogroups were investigated. A sample of 325 unrelated male volunteer donors was investigated by DYS199 typing and HVRI and II mtDNA sequencing.

2. Material and methods

Populations: Samples were obtained from three different regions of Argentina.

Southwestern region: including Mendoza, Rio Negro and Chubut Provinces ($N=100$).

Central region: including Buenos Aires and Santa Fe Provinces ($N=123$).

Northeastern region: including Formosa, Chaco, Corrientes, and Misiones Provinces ($N=102$). All samples were obtained by finger puncture with written consent of the donors or required by judicial order. Samples were spotted onto FTA paper and extracted with FTA extraction reagents.

DYS199 typing: was carried out by primer specific PCR [1]. Amplicon detection was performed by means of 2% agarose electrophoresis in $0.5 \times$ TBE and visualized by staining with EthBr through UV light.

Table 2
DYS199 distribution

	DYS199-C		DYS199-T		Total
	N	%	N	%	
Northeastern	81	79.4	21	20.6	102
Center	103	83.7	20	16.3	123
Southwestern	83	83.0	17	17.0	100
Total	267	82.1	58	17.9	325

Table 3
mtDNA Haplogroup distributions

	A		B		C		D		Total
	N	%	N	%	N	%	N	%	
Northeastern	10	15.2	26	39.4	19	28.8	11	16.6	66
Center	12	19.7	20	32.8	20	32.8	9	14.7	61
Southwestern	3	5.0	20	33.3	24	40.0	13	21.7	60
Total	25	13.4	66	35.3	63	33.7	33	17.6	187

mtDNA HVRI and II sequencing: was carried out by Big Dye sequencing system, using as templates HVRI and HVRII amplicons obtained with primers L15997 and H16410 and L15 and H480, respectively. Electrophoretic separation was carried out with an ABI 310 or ABI 3100Avant automated sequencer.

3. Results and discussion

Although it was for a long time claimed that Argentina was the “European” country of Latin America, the genetic analysis of uniparentally inherited markers strongly suggests a different landscape. Individuals with neither Y-chromosome nor mtDNA Amerindian haplogroups were detected in 28.43% in the Northeastern, 44.71% in Central region and 36% in Southwestern. A differential contribution was detected when the matrilineage and the patrilineage aboriginal components were compared to the extant population of Argentina. Only 17.9% of the samples depicted Amerindian M3-Q3* haplogroup and over 57.5% showed A, B, C or D mtDNA haplogroups. Both Amerindian traits were detected in solely 13% (in the Northeastern and Southwestern) and 10% of the samples, in the Center (Table 1).

The distribution of the M3-Q3* haplogroup in the three geographical regions is depicted in Table 2. Table 3 shows the frequency distribution of the different mtDNA haplogroups in Northeastern, Central and Southwestern regions.

The results presented herein strongly suggest an intense admixture between the Amerindian and the foreign settlers that invades since the early XVI century the territory known at present as Argentina. These observations are supported by previous reports in which the urban populations, restricted to the cities of La Plata and Buenos Aires, were investigated [2–4].

A more extensive research involving both autosomal and uniparentally inherited genetic markers, in a bigger sample size, considering all the Argentinian provinces, will help to precisely define the genetic contribution of the aboriginal Amerindians in the extant population of Argentina.

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