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Y-chromosomal STR haplotype diversity in a sample from the Metropolitan Area of Buenos Aires (Argentina)

*M. Laura Parolin *, Sergio A. Avena*, Cristina B. Dejean **, Stella M. Jaureguiberry ***, Lorena A. Sambuco *** and Francisco R. Carnese ***

*Sección de Antropología Biológica, Instituto de Ciencias Antropológicas, Facultad de Filosofía y Letras, Universidad de Buenos Aires, Argentina. Departamento de Ciencias Naturales y Antropología, Centro de Estudios Biomédicos Biotecnológicos Ambientales y Diagnóstico (CEBBAD), Universidad Maimónides, Buenos Aires, Argentina. CONICET ** Sección de Antropología Biológica, Instituto de Ciencias Antropológicas, Facultad de Filosofía y Letras, Universidad de Buenos Aires, Argentina y Centro de Estudios Biomédicos Biotecnológicos Ambientales y Diagnóstico (CEBBAD). *** Laboratorio de Genética Forense de San Martín, Buenos Aires, Argentina.

Abstract

The aim of this work was to analyze the origin of Y-chromosome haplotypes in a sample from Buenos Aires Metropolitan Area (BAMA), and compare these results with those obtained at a mitochondrial level. In order to reach this objective, 17 Y-STRs were determined from 85 unrelated blood donors. A total of 85 unique haplotypes were observed. The haplotype diversity was 1.000 +/- 0.0018, and the average genetic diversity 0.680+/-0.095. Paternal lineages showed a genetic homogeneity of European roots (93%), mainly from Italy and Spain. Amerindian paternal contribution associated to sub-haplogroup Q1a3a was relatively low (6%). The low proportion of Amerindian haplotypes and the high number of maternal lineages (44%) from the same sample reveal a distinguishing input by gender in the history of this population. A single profile, E1b1a, was found, predominant in sub-Saharan Africa. These data, together with the historical and demographic information, allow us to state that the small Amerindian and sub-Saharan contribution observed in the sample from BAMA would result from recent migrations started in the mid twentieth century, mainly from the north of Argentina and bordering countries of high native composition, and, to a lesser extent, from African origin.

Keywords: Y-chromosome, haplotypes, population genetics, Buenos Aires.

Diversidad de Haplotipos del cromosoma Y en una muestra del área metropolitana de Buenos Aires (Argentina)

Resumen

El objetivo de este trabajo fue analizar el origen de los haplotipos del cromosoma Y en una muestra poblacional del Área Metropolitana de Buenos Aires (AMBA), y comparar estos resultados con los obtenidos previamente a nivel mitocondrial. Se determinaron 17 marcadores Y-STRs en 85 donantes no emparentados. Un total de 85 haplotipos únicos fueron observados. La diversidad haplotípica fue de 1,000 +/- 0.0018, y la diversidad genética media de 0,680+/-0,095. Los linajes paternos evidenciaron una homogeneidad genética de raíces Europeas (93%), procedentes principalmente de Italia y España. La contribución amerindia paterna asociada al sub-haplogrupo Q1a3a fue relativamente baja (6%). La menor proporción de haplotipos amerindios y el elevado número de linajes maternos (44%) de ese origen, revela que ha habido un aporte diferencial por género en la historia de mestizaje de esa población. Se observó un único perfil E1b1a, el cual es predominante en África subsahariana. Estos datos, conjuntamente con la información histórica y demográfica, nos permite afirmar que el bajo aporte amerindio y subsahariano observado en la muestra del AMBA, sería el resultado de las migraciones recientes, iniciadas a mediados del siglo XX, principalmente desde el norte de Argentina y de países limítrofes de elevada composición nativa y, en menor medida, africana.

Palabras clave: cromosoma Y, haplotipos, genética de poblaciones, Buenos Aires.

Human populations can be characterized by their genetic structure, which is the result of microevolution factors that have acted upon them and determined their biological diversity. In contrast to isolated or semi isolated populations, where the genetic drift would play an important role in the decrease of intrapopulational variability and in the tendency to homozygosity, in cosmopolitan populations the most important

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microevolution factor is gene flow that, as a consequence of migrations, produces an increase of biological diversity and, as a result, modifies the original genetic composition of the receiving group.

In Argentina the massive migratory currents of European origin, which mainly arrived from 1880 to 1930, produced substantial changes at demographic and cultural levels, particularly in Buenos Aires city. Of a total of 3,500,000 immigrants of European origin, approximately 800,000 settled in the city of Buenos Aires (Sánchez Alonso 1992). Therefore, as expected, in the middle of the twentieth century, the frequencies of ABO and Rh systems were similar to those of Spain and Italy (Avena 2003).

Later, as a consequence of industrial development initiated in the mid-1940s, and concomitantly with the decrease in the arrival of Europeans, there was an increase of internal migrants, mainly from the northern provinces and also from neighboring countries (Avena 2003), with a high Amerindian composition, and a low African one. The latest arrived in America due to the slave trade (Lorandi 1992, Avena et al. 2001, Avena 2003). These population's movements have produced new changes in Buenos Aires Metropolitan Area (BAMA), which are reflected in census information. In Buenos Aires city in 1947, 94% and 6% of the foreigners came from European and South American countries respectively (Dirección Nacional del Servicio Estadístico 1947). On the other hand, in 2010 these values were 34% and 66% respectively (INDEC 2010).

Previous studies carried out by our research team showed that, at the level of protein markers, there was an European, Indigenous and African participation of 81%, 15%, and 4% respectively in the gene pool of the BAMA population. Likewise, in these works it can be observed that native contribution was differentially distributed among the different districts analyzed: 5% in Buenos Aires city, 11% in the first outer-ring suburb (O-R1) and 33% in the second one (O-R2) (Avena *et al.* 2001, 2006, Avena 2003). At the same time, by means of mitochondrial haplotypes analyses, a 44% aboriginal maternal contribution was estimated, showing a similar distribution in Buenos Aires city (37%) and O-R1 (38%), and a marked growth of the native component in O-R2 (67%), while the sub-Saharan component was 1% (Dejean *et al.* 2003, Carnese 2006).

At a different level of analysis, the same sample was studied by Fejerman et al. (2005), who determined twelve molecular autosomal markers and obtained 2.2% of gene admixture with Africans, although the proportion of individuals with some sub-Saharan influence would be of 10%. This is compatible with a crossbreeding process which occurred several generations back. In this study, admixture with Amerindian people was not determined.

These estimations, in correspondence to historic and demographic information for every district, demonstrated that Indigenous and sub-Saharan participation has a considerably more important incidence than is normally accepted at academic and popular levels.

The researches in cosmopolitan populations mentioned so far have employed biparental markers that allow us to estimate the population gene admixture degree, while the mtDNA haplogroups allowed us to analyze the direction of maternal gene flow. It was possible to observe that the major presence of Amerindian uniparental lineages by maternal via, in relation to the paternal ones, is concordant with a scheme where the crossbreed mainly occurred between native women and men of different origins, which has been widely observed in our country, as shown in the works of Dipierri et al. (1998) in two villages at different altitudes in the Quebrada de Humahuaca; Martínez Marignac et al. (1999, 2004) in La Plata; Corach et al. (2006), and Marino et al. (2007) in samples of ten Argentine provinces; García and Demarchi (2006) in two admixed populations from Córdoba province; Ramallo et al. (2007) in an isolated rural area of Catamarca province, and the works developed by our research team in three cosmopolitan populations from Patagonian region (Avena et al. 2007, 2009, 2010). This process was also detected in other South American countries, such as Uruguay (Sans et al. 2002), Brazil (Bortolini et al. 1999, Marrero et al. 2007), Chile (Rocco et al. 2002), and Colombia (Bedoya et al. 2006). Recently, Wang et al. (2008) have analyzed the geographical patterns of miscegenation in mestizo individuals of Argentina, Brazil, Chile, Colombia, Costa Rica, Guatemala, and México, corroborating that the differential mixing process was the rule in Latin America.

However, although the degree of participation of the Amerindian maternal lineages in the BAMA is known, there have not carried out studies on the geographical origin of the paternal lineages in that region. In this respect, the analysis of the polymorphisms of the Y-chromosome, and especially the haplotypes derived of the analysis of STRs (short tandem repeats), constitutes an excellent tool for researching the historical and evolutionary processes of human populations. Therefore, the aim of this work was to typify in the same population sample of the BAMA, 17 Y-STRs in order to analyze the geographic origin of the Y-chromosome haplotypes, the paternal ancestral migratory movements, and their distribution in the different areas of this region: Buenos Aires city, O-R1, and O-R2 of the suburbs. Also, these results were contrasted with the genealogical information of the donors, and compared with the data obtained from the same sample concerning the mitochondrial haplogroup, in order to evaluate the asymmetry by sexes that was observed between native women and men of other origin.

The information obtained will allow us to know with greater precision the genetic structure of the BAMA population, and to analyze and rebuild the history, the migration movements, as well as the mixture degree of

that region. Meanwhile, the data obtained will contribute to develop the worldwide databases of Y-Chromosome STRs, which are highly useful in anthropological, archaeological and forensic research.

Subjects and methods

Biological samples

The present study was performed in a sample of 85 non-related blood donors that concurred to the Hemotherapy Service of Hospital Italiano and Hospital de Clínicas from Buenos Aires city from 1998 to 2002, and who reside in three zones of the BAMA: Buenos Aires city (CABA; n=28), O-R1 (n=23), and O-R2 (n=34) (Figure 1). All of them were adequately informed about the aims of the study, and gave their consent to perform this research. The samples were collected in sterile tubes with anticoagulant (ACD), and were codified, registered as "anonymous", and preserved in the DNA bank of the Sección de Antropología Biológica, Instituto de Ciencias Antropológicas, Facultad de Filosofía y Letras of the University of Buenos Aires. Also, a survey was conducted on each one of the participants, in order to collect data on place of birth, present residence, and genealogical information of the two preceding generations.

DNA extraction and genotyping

DNA extraction was performed by means of the phenol-chloroform method, following the protocol of Sambrook *et al.* (1989). The standardization of the 17 located Y-STRs markers in the Y-chromosome non-recombinant region was carried out using the AmpFlSTR® Yfiler amplification kit, following the manufacturer instructions (AmpFlSTR® Yfiler PCR Amplification kit, User's Manual Applied Biosystems). The PCR products were separated and analyzed in an ABI Prism 3130 capillary sequencer. For the study of the mutation M3, a single nucleotide polimorphism (SNP), that defines the Amerindian sub-haplogroup Q1a3a, was followed the protocol of Underhill *et al.* (1996). In South Amerindians, Q1a3a has a 77- 90 % frequency, and is absent in other worldwide populations (Bianchi et al. 1997; Bortoloni et al. 2003; Zegura *et al.* 2004).

Statistical analysis

Frequencies calculation and allelic and haplotypes diversity, analysis of molecular variance (AMOVA), and the genetic differences between population pairs (pairwise F_{ST}) were determined by executing the Arlequin 3.1 program (Excoffier et al. 2005). The corresponding dendrogram of biological affinities was constructed by the UPGMA (Unweighted Pair Group Method with Arithmetic Averaging) algorithm (Sneath and Sokal 1973), using the statistical package Phylip v.3.65 (Felsenstein 2004) and the Treview32 program (Page 1996).

The data obtained in this study were compared with those published by other authors in relation to seven worldwide populations, two European: Italy (N= 163; Ferri et al. 2008) and Spain (N= 164; Martin et al. 2004), two African: Mozambique (N= 102; Alves et al. 2003) and Equatorial Guinea (N= 112; Arroyo-Pardo et al. 2005), a cosmopolitan Argentine population: Córdoba (N= 100; Salas *et al.* 2008) and two South Amerindian populations: Toba of the Argentine Chaco (N= 49; Toscanini *et al.* 2008) and Guaraní-Kaingang of Brazil (N= 46; Leite *et al.* 2008).

The comparation of the minimum haplotypes for 9 loci Y-STR was analyzed: DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, and DYS385a/b.

The contribution of paternal lineages arising from different geographical origins was estimated, first of all, by inferring the Y-chromosome haplogroup with the Haplogroup Predictor program (Athey, 2006). For this, the extended haplotypes were used (17 Y-STRs markers), and only those profiles that were defined for a unique haplogroup with a ≥ 99% a posteriori probability were considered in the analysis. They were named according to the alpha numerical system updated by Karafet et al. (2008). Secondly, the minimum haplotypes were entered into the world-wide YHRD reference database (http://www.yhrd.org), focusing our analysis mainly on finding these profiles in the metapopulations of European (Spain and Italy), African and Native American origins. Also, the proportion of men carrying the M3 mutation

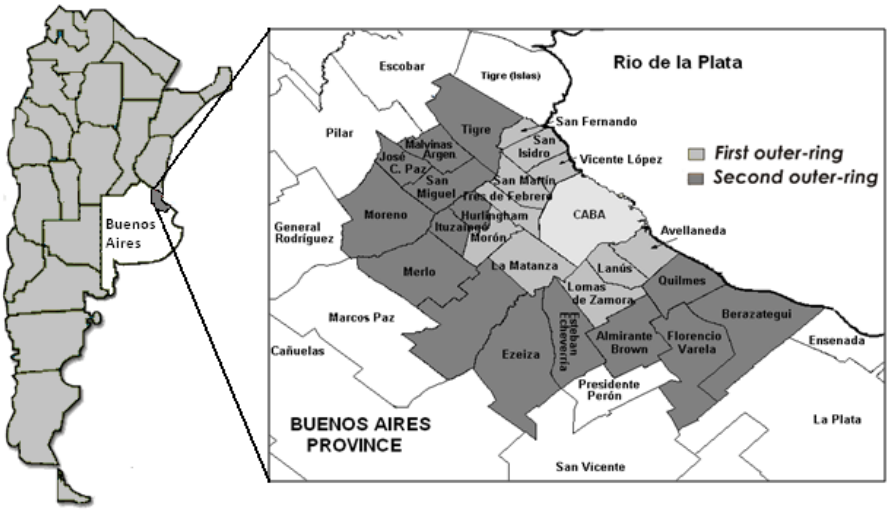

Figure 1. Map of Argentina indicating the city of Buenos Aires (CABA) and first (O-R1) and second (O-R2) outer suburbs of Greater Buenos Aires (34° 36'S; 58° 22'W).

Figura 1. Mapa de la Argentina indicando la ciudad de Buenos Aires (CABA) y el Primer (O-R1) y Segundo anillo (O-R2) del conurbano (34° 36'S; 58° 22'W).

that defines the Amerindian sub-haplogroup Q1a3awas considered by direct count and it was contrasted with the geographical distribution of the haplotypes found in the YHRD base and with the haplogroup obtained inferred by the Predictor program.

These results were also compared with the donor's paternal genealogical information and with the mitochondrial haplogroup previously determined in each individual in the same sample by means of RFLP (Restriction Fragments Length Polymorphisms) (Dejean et al. 2003, Carnese 2006).

Results and discussion

Y-STRs haplotypes

From the analysis of 17 Y-STR markers, a total of 85 unique haplotypes were observed in the BAMA (Table 1). The haplotype diversity was 1.000 +/- 0.0018, and the genetic diversity average 0.680 +/- 0.095. By means of the analysis of 9 Y-STR, 82 different profiles were observed, with an haplotype diversity and genetic average of 0.998 +/- 0.0018 and 0.674 +/- 0.088, respectively. In addition, the observed allelic diversity varied between 0.514+/- 0.056 for the less polymorphic locus DYS393 and 0.928 +/- 0.019 for the marker of greater variability DYS385 (combination of the genotype a/b) (Table 2). These diversity indicators show values similar to those registered in different European populations (Roewer et al. 2001, Schmidt et al. 2003, Martin et al. 2004, Ferri et al. 2008), and in cosmopolitan groups from Argentina (Marino et al. 2007, 2010, Salas et al. 2008).

Haplogroup inference and phylogeography of the Y-STR profiles

The Y-chromosome haplogroups were inferred from the extended haplotypes by means of a bayesian method executed by Haplogroup Predictor program. 95% (81/85) of the haplotypes could be assigned to a unique haplogroup (HG) with a > 99% a posteriori probability (Table 1). The most frequent haplogroup was R1b (38%: 31/81), that is also the lineage of major prevalence in Europe, mainly in the Western and Mediterranean regions, and is associated to modal haplotype: DYS19*14, DYS390*13, DYS389I*29, DYS389II*24, DYS391*10 or DYS391*11, DYS392*13, DYS393*13, DYS385*11-14 (Presciuttini et al. 2001, Roewer et al. 2001, Gusmaõ et al. 2003, Schmidt et al. 2003, Brion et al. 2004, Sims et al. 2007, Ferri et al. 2008, Marino et al. 2010). Secondly, the HG E1b1b was registered (15%: 12/81), which predominates in the northwest, south and east of Africa, and in the Middle East, while in Europe it has a higher presence in the Balkan Mountains, followed by Italy and Spain (Underhill et al. 2001, Semino et al. 2002, Cruciani et al. 2004, 2007, Luis et al. 2004). In smaller proportions, G2 (11%), I1 and I2 (10%), J2 (8.6%) and R1a (7.4%) lineages were observed. They showed a wide distribution in populations of European origin and in the Near East (Underhill et al. 2001; Jobling and Tyler-Smith, 2003; Brion et al. 2004; Cruciani et al. 2007). The HG T (formerly haplogroup K2) showed low frequency (2%), and it was observed in Western Africa and the Horn of Africa, Arabian Peninsula, and India (Luis et al. 2004). The HG N, which is distributed mainly from the northeast of Eurasia to the south of Siberia (Rootsi et al. 2007), was carried by a single donor (1%).

A single profile (ID= 72; Table 1) was assigned to the HG E1b1a, which is almost exclusive and predominant in sub-Saharan Africa, and it corresponds with the modal haplotype Bantú, described by Thomas et al. (2000): DYS19*15; DYS390*21; DYS391*10; DYS392*11 and DYS393*13. This HG is observed with high frequency in Senegal and Cameroon (81-95%) (Semino et al. 2002; Luis et al. 2004) and in a 60% in Afro-American populations (Sims et al. 2007).

The Amerindian paternal contribution associated to sub-haplogroup Q1a3a was relatively low (6%: 5/85), having been observed in five samples (ID: 6, 9, 46, 50, 70, Table 1) which registered probabilities of inference between 99.7 - 100%, and presence of the precise mutation M3 in locus DYS199. In addition, these profiles exhibited the markers of the modal haplotype described for Amerindian populations: DYS19*13, DYS390*24, DYS391*10, DYS392*14 and DYS393*13 (Bianchi et al. 1998, Bortolini et al. 2003, Zegura et al. 2004) presenting one and two step differences. As it was already mentioned, HG Q1a3a is restricted to the American continent, and among South Amerindians it reaches frequencies of 77-90% (Bianchi et al. 1997; Bortolini et al. 2003; Zegura et al. 2004).

When entering the minimum Y-STR haplotypes into the YHRD reference database (with 72,946 haplotypes at the time of the search), 25% (21/85) of them were not detected. As for the rest, 86% (55/64) were observed in metapopulations of Eurasia, 64% (41/64) of which were found in populations of Italy and Spain. The profiles that match with a greater prevalence in the worldwide base were assigned to the HG R1b, and present one step variations in respect to European modal haplotype (Roewer et al. 2001; Gusmaõ et al. 2003) (see Tables 1 and 3). As well, 14% (9/64) were observed in Asian groups (Sino Tibetano), African (Tunisia, Egypt, Somalia and Ethiopia), and Mestizo of Latin America (Argentina, Colombia and Peru). The profile ID: 72, assigned to the HG E1b1a was only found in two individuals from sub-Saharan origin and one from Colombia. Of the five haplotypes that displayed the Amerindian variant M3, three of them were in the YHRD base, one (ID: 9) in Colombia and the remaining two (ID: 6 and 70) in mixed populations of the Argentine provinces of Salta, Mendoza, and Rio Negro. When the bi-allelic locus DYS385 was not considered, it was observed that 4 out of 5 carrying samples of C-T transition match with Amerindians, native populations of the Australian Continent and Mestizo populations of Latin America. Also, the haplotype ID: 72 was indeed observed in greater proportion in African metapopulations (Table

ID	DYS19	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393	DYS385a/b	DYS438	DYS439	DYS437	DYS448	DYS456	DYS458	DYS635	GATA H4	HG	Pb (%)	M3 mutation
1	11	13	31	24	10	12	13	16-18	12	11	14	20	15	16	21	12	E1b1b	100	C
2	13	12	29	25	10	11	13	17-17	10	12	14	20	15	14	22	11	E1b1b	100	C
3	13	13	30	24	10	11	13	16-19	10	12	14	20	17	15	22	12	E1b1b	100	C
4	13	13	30	24	10	11	13	15-18	10	12	14	20	16	15	22	12	E1b1b	100	C
5	13	13	30	24	10	11	13	15-18	10	12	14	19	17	15	21	11	E1b1b	100	C
6	13	13	30	24	10	14	12	15-15	12	12	14	20	15	16	22	12	Q1a3a	100	T
7	13	13	31	24	10	11	13	16-18	10	12	14	20	17	15	23	11	E1b1b	100	C
8	13	13	31	25	10	11	13	16-18	10	12	14	20	15	16	22	11	E1b1b	100	C
9	13	13	32	24	10	14	13	15-15	11	11	13	19	16	16	22	11	Q1a3a	100	T
10	13	14	30	24	9	11	13	13-14	10	10	14	20	16	18	23	12	E1b1b	100	C
11	13	14	30	24	10	11	13	13-14	10	10	14	20	16	18	23	12	E1b1b	100	C
12	13	14	31	24	10	11	13	16-18	10	12	14	20	15	15	23	11	E1b1b	100	C
13	14	12	26	23	11	13	13	11-14	12	13	14	19	16	16	22	12	R1b	100	C
14	14	12	27	23	10	11	14	13-15	10	11	16	20	13	15	21	11	I1	99.9	C
15	14	12	27	23	10	13	13	14-15	9	11	14	19	15	16	21	11	T	100	C
16	14	12	28	22	10	11	14	15-16	10	13	16	21	17	15	21	12	G2	99.8	C
17	14	12	28	23	11	12	13	11-14	12	13	13	18	15	18	23	12	R1b	99.9	C
18	14	12	28	23	11	13	13	11-14	12	13	13	18	15	18	23	12	R1b	100	C
19	14	12	28	24	11	13	13	11-14	12	12	15	20	15	17	23	11	R1b	100	C
20	14	12	29	23	9	11	13	13-15	10	11	16	20	14	15	21	11	I1	100	C
21	14	12	29	24	11	13	14	11-14	12	12	15	19	15	17	23	12	R1b	100	C
22	14	12	29	25	9	11	14	14-15	10	11	16	21	15	17	20	12	G2	100	C
23	14	13	28	24	11	15	12	11-16	12	12	15	18	16	20	23	12	R1b	100	C
24	14	13	29	23	10	11	12	13-16	9	11	15	21	14	15	22	12	J2a	100	C
25	14	13	29	23	10	11	12	13-16	9	10	14	20	15	15	21	11	J2a	100	C
26	14	13	29	23	10	11	12	13-18	9	11	15	22	15	15	21	11	J2a	100	C
27	14	13	29	23	10	11	13	11-13	12	12	15	19	15	16	23	11	R1b	100	C
28	14	13	29	23	10	13	13	13-13	12	12	15	19	15	17	24	12	R1b	100	C
29	14	13	29	23	10	13	13	12-13	12	12	15	19	17	17	23	12	R1b	100	C
30	14	13	29	23	11	13	13	11-15	12	12	14	19	15	17	25	12	R1b	100	C
31	14	13	29	24	10	11	12	13-17	9	10	15	20	16	15	21	11	J2a	100	C
32	14	13	29	24	10	13	13	11-14	12	12	15	19	15	17	23	12	R1b	100	C
33	14	13	29	24	10	13	13	11-15	12	12	15	19	15	17	23	12	R1b	100	C
34	14	13	29	24	11	12	12	11-16	12	12	15	19	15	19	24	12	R1b	100	C
35	14	13	29	24	11	13	13	11-14	11	12	15	19	16	17	24	12	R1b	100	C
36	14	13	29	24	11	13	13	11-15	12	12	15	19	16	18	23	12	R1b	100	C
37	14	13	29	24	11	13	13	12-15	12	13	14	18	16	17	23	10	R1b	100	C
38	14	13	29	24	11	14	13	11-12	12	13	15	19	15	16	23	12	R1b	100	C
39	14	13	29	24	12	12	13	11-14	12	12	14	17	15	19	24	11	R1b	100	C
40	14	13	29	25	12	13	13	11-14	12	12	14	18	16	17	23	12	R1b	100	C
41	14	13	29	26	11	13	13	11-14	12	12	15	19	15	17	24	12	R1b	100	C
42	14	13	30	23	10	11	13	17-19	10	13	14	20	16	15	21	11	E1b1b	100	C
43	14	13	30	23	10	13	13	14-17	9	12	14	19	15	17	20	11	T	100	C
44	14	13	30	23	11	13	13	11-14	12	13	15	19	16	16	24	11	R1b	100	C
45	14	13	30	24	11	13	14	11-14	12	12	14	17	15	16	23	11	R1b	100	C
46	14	13	31	25	10	14	13	13-18	11	12	14	20	15	16	22	12	Q1a3a	99.9	T
47	14	14	29	24	11	13	13	11-14	12	11	15	18	15	17	23	11	R1b	100	C
48	14	14	30	22	11	14	14	11-15	10	11	15	19	15	17	20	12	N	99.2	C
49	14	14	30	23	10	10	12	13-21	10	12	14	21	15	16	23	11	J1	97.3	C
50	14	14	30	24	9	14	13	14-15	13	14	14	20	15	17	22	12	Q1a3a	99.9	T
51	14	14	30	24	11	13	13	11-14	12	11	14	19	15	17	23	12	R1b	100	C
52	14	14	30	24	11	13	13	12-14	12	12	14	18	16	17	23	11	R1b	100	C
53	14	14	30	25	11	13	13	11-14	12	12	15	18	16	18	24	12	R1b	100	C
54	14	14	30	25	11	13	13	12-14	12	11	15	19	16	17	23	12	R1b	100	C
55	14	14	30	26	11	13	13	11-14	12	12	14	18	16	16	23	12	R1b	100	C
56	14	14	31	22	9	12	12	13-14	9	12	15	20	15	17	24	11	J2a	99.9	C
57	14	14	31	23	10	11	13	14-15	9	11	14	21	17	17	23	11	J2a	99.9	C
58	14	14	31	24	10	14	13	12-15	12	12	14	18	15	17	23	11	R1b	100	C
59	15	12	28	22	10	11	13	13-15	10	12	16	21	17	17	21	11	G2	100	C
60	15	12	29	20	10	11	15	15-16	11	11	16	21	15	18	21	12	G2	97.2	C
61	15	12	29	22	10	11	13	12-18	10	11	16	21	16	16	21	12	G2	100	C
62	15	12	29	22	10	11	13	13-14	10	12	16	20	14	15	21	11	I1	99.4	C
63	15	12	29	22	10	11	15	14-14	10	11	16	21	15	16	23	12	G2	100	C
64	15	12	29	23	10	11	12	13-19	10	11	14	19	16	15	20	11	J1	76.7	C
65	15	13	28	24	10	12	15	16-16	10	11	15	20	14	16	19	11	I2b1	100	C
66	15	13	29	23	10	12	15	16-16	10	11	14	20	14	16	22	11	I2b1	100	C
67	15	13	29	23	11	12	14	15-15	10	11	15	20	18	15	21	11	I2b1	100	C
68	15	13	29	24	11	13	13	11-14	12	12	15	19	16	18	23	11	R1b	100	C
69	15	13	30	24	10	11	13	11-14	11	10	14	20	16	16	23	12	R1a	100	C
70	15	13	31	25	10	14	13	12-19	11	13	14	20	15	16	22	12	Q1a3a	99.7	T
71	15	14	30	23	11	13	13	12-15	12	11	15	19	16	18	23	12	R1b	100	C
72	15	14	31	21	10	11	13	16-19	9	12	15	19	15	19	21	11	E1b1a	99.9	C
73	16	12	28	22	10	11	14	14-14	10	11	16	21	15	16	20	12	G2	100	C
74	16	12	28	22	10	11	14	14-14	11	11	16	21	15	16	20	13	G2	100	C
75	16	12	28	24	10	11	12	10-17	9	12	16	19	15	15	21	11	J2b	100	C
76	16	12	29	22	10	11	14	14-15	10	13	15	21	15	16	20	13	G2	100	C
77	16	12	30	24	10	11	13	16-19	9	12	14	20	14	15	21	11	E1b1b	98.4	C
78	16	13	29	25	10	11	13	12-14	11	11	14	20	18	16	23	13	R1a	100	C
79	16	13	30	22	11	11	12	11-15	11	10	14	21	16	16	24	12	R1a	100	C
80	16	13	31	24	10	11	13	12-15	10	12	14	20	16	16	23	11	I2a	99.7	C
81	16	13	31	24	10	11	14	11-14	11	10	14	20	14	15	23	12	R1a	100	C
82	16	13	31	24	11	11	13	14-15	10	13	15	20	15	17	24	11	I2a	100	C
83	16	14	30	22	11	11	14	13-14	10	12	15	20	15	17	21	12	G2	99.6	C
84	16	14	31	26	10	11	13	11-14	11	10	14	19	17	16	23	12	R1a	100	C
85	18	13	30	25	11	11	13	11-13	11	11	14	20	16	15	23	13	R1a	100	C

Table 1. Haplotypes observed in BAMA (N= 85) for 17 Y-STR markers, the haplogroup (HG) inferred with its corresponding a posteriori probability (Pb), and the presence of the M3 mutation defining the Amerindian sub-haplogroup Q1a3a. ID: identification number assigned to each sample.

Tabla 1. Haplotipos observados en AMBA (N=85) para 17 marcadores del Y-STR, El haplogrupo (HG) es inferido con la correspondiente probabilidad (Pb) y la presencia de la mutación M3 definiendo al subhaplogrupo Amerindio Q1a3a. ID: número de identificación asignado a cada muestra.

4). These observations show a correspondence with the geographical origin of the inferred HG.

Population structure analysis

By means of pairwise analysis , no significant differences were found in the distribution of the Y-STR haplotypes between Buenos Aires city, O-R1 and O-R2 of the suburbs (Pairwise F_{ST} CABA vs O-R1 = 0.008; F_{ST} CABA vs O-R2= 0.013; F_{ST} OR1 vs O-R2 = 0.009; F_{ST} P-values > 0.05). Also, the analysis of molecular variance (AMOVA) exhibited a low degree of genetic differentiation among these three areas: F_{ST} = 0.0003 (interpopulation variation = 0.03%, and intrapopulation variation = 99.97%). This fact would demonstrate a considerably homogenous distribution of the Y- chromosome haplotypes in the sample, possibly due to the high proportion of European paternal lineages in both subsamples. On the other hand, in this same sample, a 54 % of native maternal contribution was observed (unpublished data). This value is higher than the 44% already indicated for the total sample (Dejean

DYS19				DYS389I				DYS389II				DYS390			
Allele	N	Freq.	± SD	Allele	N	Freq.	± SD		N	Freq.	± SD	Allele	N	Freq.	± SD
11	1	0.012	0.012	12	22	0.259	0.048	26	1	0.012	0.012	20	1	0.012	0.012
13	11	0.129	0.037	**13**	**44**	**0.518**	**0.055**	27	2	0.024	0.017	21	1	0.012	0.012
14	**46**	**0.541**	**0.054**	14	19	0.224	0.045	28	10	0.118	0.035	22	12	0.141	0.038
15	14	0.165	0.040					**29**	**33**	**0.388**	**0.053**	23	22	0.259	0.048
16	12	0.141	0.038					30	24	0.282	0.049	**24**	**36**	**0.424**	**0.054**
18	1	0.012	0.012					31	14	0.165	0.040	25	10	0.118	0.035
								32	1	0.012	0.012	26	3	0.035	0.020
GD		0.651	0.044	GD		0.622	0.031	GD		0.661	0.036	GD		0.727	0.030
DYS391				DYS392				DYS393							
9	5	0.059	0.026	10	1	0.012	0.012	12	12	0.141	0.038				
10	**49**	**0.576**	**0.054**	11	41	0.482	0.055	**13**	**57**	**0.671**	**0.051**				
11	29	0.341	0.052	12	8	0.094	0.032	14	12	0.141	0.038				
12	2	0.024	0.017	**13**	**26**	**0.306**	**0.050**	15	4	0.047	0.023				
				14	8	0.094	0.032								
				15	1	0.012	0.012								
GD		0.554	0.036	GD		0.664	0.034	GD		0.514	0.056				
DYS385a/b															
10-17	1	0.012	0.012	12-14	3	0.035	0.020	13-17	1	0.012	0.012	15-16	2	0.024	0.017
11-12	1	0.012	0.012	12-15	4	0.047	0.023	13-18	2	0.024	0.017	15-18	2	0.024	0.017
11-13	2	0.024	0.017	12-18	1	0.012	0.012	13-19	1	0.012	0.012	16-16	2	0.024	0.017
11-14	**20**	**0.235**	**0.046**	12-19	1	0.012	0.012	13-21	1	0.012	0.012	16-18	4	0.047	0.023
11-15	5	0.059	0.026	13-13	1	0.012	0.012	14-14	3	0.035	0.020	16-19	3	0.035	0.020
11-16	2	0.024	0.017	13-14	5	0.059	0.026	14-15	6	0.071	0.028	17-17	1	0.012	0.012
12-13	1	0.012	0.012	13-15	3	0.035	0.020	14-17	1	0.012	0.012	17-19	1	0.012	0.012
				13-16	2	0.024	0.017	15-15	3	0.035	0.020				
												GD		0.928	0.019
DYS438				DYS439				DYS437				DYS448			
9	11	0.129	0.037	10	8	0.094	0.032	13	3	0.035	0.020	17	2	0.024	0.017
10	29	0.341	0.052	11	25	0.294	0.050	**14**	**40**	**0.471**	**0.054**	18	10	0.118	0.035
11	12	0.141	0.038	**12**	**40**	**0.471**	**0.054**	15	30	0.353	0.052	19	27	0.318	0.051
12	**32**	**0.376**	**0.053**	13	11	0.129	0.037	16	12	0.141	0.038	**20**	**32**	**0.376**	**0.053**
13	1	0.012	0.012	14	1	0.012	0.012					21	13	0.153	0.039
												22	1	0.012	0.012
GD		0.713	0.024	GD		0.674	0.032	GD		0.640	0.029	GD		0.728	0.025
DYS456				DYS458				DYS635				GATA H4			
13	1	0.012	0.012	14	1	0.012	0.012	19	1	0.012	0.012	10	1	0.012	0.012
14	7	0.082	0.030	15	20	0.235	0.046	20	7	0.082	0.030	11	37	0.435	0.054
15	**42**	**0.494**	**0.055**	**16**	**26**	**0.306**	**0.050**	21	19	0.224	0.045	**12**	**43**	**0.506**	**0.055**
16	25	0.294	0.050	17	25	0.294	0.050	22	12	0.141	0.038	13	4	0.047	0.023
17	8	0.094	0.032	18	9	0.106	0.034	**23**	**35**	**0.412**	**0.054**				
18	2	0.024	0.017	19	3	0.035	0.020	24	10	0.118	0.035				
				20	1	0.012	0.012	25	1	0.012	0.012				
GD		0.661	0.036	GD		0.761	0.019	GD		0.749	0.030	GD		0.559	0.025

Table 2. Allelic frequencies (Freq.) and genetic diversity (GD) of the17 Y-STR markers analyzed with their corresponding standard deviations (SD). N: number of alleles. In bold: the most frequent allele/genotype observed.

Tabla 2. Frecuencias alélicas (Freq.) y diversidad genética (GD) de los 17 marcadores del Y-STR analizados con su correspondiente desviaciones estándar (SD). N: número de alelos. En negrita: los alelos/genotipo observados con más frecuencia.

et al. 2003), because in this study a greater proportion of resident people in O-R2 had been included, where there is a higher Amerindian contribution.

The smaller value of native paternal contribution (6%) would be indicating us that, in the sample from BAMA, there were a clear asymmetry by sex in the mixing process. As previously indicated, this noticeable incidence of biological unions between native women and men of another origin has been previously observed in different urban centers of Argentina and other Latin American countries. Among European immigrants, the amount of men was always proportionally greater, both in colonial times (Assadourian et al. 1986) and after the national independence in 1816 (Seefeld 1986). According to the Argentine national censuses of the years 1895 and 1914, the proportion of foreigners was 172 men per 100 women. In conclusion, it is appraised that the high index of masculinity of European immigrants has remained constant from 1580, when Buenos Aires city was founded for the second time, until the beginning of the twentieth century, which had brought about interethnic unions (Lorandi 1992, Avena et al. 2006).

On the other hand, it is worth noting that 4 out of 5 men carrying the Q1a3a sub-haplogroup also exhibited an Amerindian mitochondrial haplogroup. This has also been observed in previous studies conducted by our research team, and, at present, out of 27 men with native paternal lineage, 26 also had a native maternal lineage in a sample from Comodoro Rivadavia (Chubut province) (Avena et al. 2009). This fact would be indicating us that, although this has been clearly an unequal contribution of the native uniparental lineages, no evidence of marked panmixia has been found, which could be explained by the persistence of several factors considered in the election of a partner, such as origin, migratory history, residence place and socioeconomic stratification (Carnese 2006).

When comparing the minimum Y-STR haplotypes with those obtained by other authors in seven worldwide populations, significant differences were observed (F_{ST} P-values < 0.05) among all the pairs analyzed, with the exception of BAMA with Italy (F_{ST} P-values = 0.0991) and Spain (F_{ST} P-values = 0.0810), that did not show significant differences among each other. Table 5 shows the matrix of Pairwise, with the respective values for Fst distances. In the same table, it can be observed that the BAMA presents smaller distances (0.0007< F_{ST} <0.0011) with respect to the groups of Europe and Córdoba (Argentina), and greater values (0.0032< F_{ST} <0.0247) than those of the Amerindian and African populations (Table 5).

In relation to these results, the corresponding dendrogram of genetic affinities exhibited, on the one hand, a subgroup of biological similarity integrated by the populations of Europe, BAMA and Córdoba (Argentina) and, on the other hand, the populations of Africa, whereas the Toba from the Argentinean Gran Chaco and the Guaraní-Kaingang from Brazil, were located in different ends of the tree (Figure 2). These observations show a close relation with the historical-demographic phenomena previously described, in connection with the massive inmigratory movements that took place in the province of Buenos Aires since the end of the XIXth Century until the middle of the twentieth century, presenting high European

ID	Minimal haplotype	Worldwide n= 72946	Eurasian n= 42447	European n= 36109	Italy n= 3175	Spain n= 1959	Argentina n= 1937
35	14-13-29-24-11-13-13-11,14	1369	988	981	91	139	65
32	14-13-29-24-10-13-13-11,14	635	491	486	39	64	45
51	14-14-30-24-11-13-13-11,14	403	269	269	12	52	35
36	14-13-29-24-11-13-13-11,15	358	292	287	25	25	21

Table 3. Minimal haplotype frequently observed in the YHRD worldwide database. ID: identification number assigned to each sample.

Tabla 3. Mínimo haplotipo observado con frecuencia en la base de datos YHRD en todo el mundo. ID: número de identificación asignado a cada muestra.

ID	HG	YHRD Worldwide N= 74724; 7 loci Y-STR,
6	Q1a3a	Admixed: 2 Mendoza, 1 Salta, 1 Buenos Aires, 1 Rio Negro (Argentina) and 5 Peru,
9	Q1a3a	Amerindian: 3 toba and 2 pilaga (Formosa Argentina); Admixed: 2 Mexico, 1 Colombia, 1 Peru and 1 Costa Rica
46	Q1a3a	Amerindian: 2 Quichua (Ecuador); Admixed: 2 Quito (Ecuador)
50	Q1a3a	0
70	Q1a3a	Natives from Oceanía: 2 Trobriand Islands (Papua New Guinea); Admixed: 1 Mendoza (Argentina), 1 Peru
72	E1b1a	Subsaharians and Afroamericans: 28; Admixed: 4 Colombia, 1 Mexico, 1 Brasil

Table 4. Haplotypes assigned to an Amerindian and sub-Saharan origin, and their distribution in the YHRD database for 7 loci Y-STR (DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392 and DYS393). ID: identification number assigned to each sample; HG: haplogroup inferred.

Tabla 4. Haplotipos asigandos a origenes Amerindios y Subsaharianos y su distribucion en la base de datos YHRD para 7 loci de Y-STR (DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392 and DYS393). ID: número de identificación asignado a cada muestra; HG: haplogrupo inferido.

	BAMA	CDB	ITAL	SPN	MOZB	EC-GUIN	TOB-CH	GUAR-KG
BAMA	0.0000							
CDB	0.0018	0.0000						
ITAL	0.0007*	0.0031	0.0000					
SPN	0.0011*	0.0045	0.0018	0.0000				
MOZB	0.0063	0.0073	0.0075	0.0089	0.0000			
EC-GUIN	0.0032	0.0046	0.0043	0.0053	0.0073	0.0000		
TOB-CH	0.0247	0.0259	0.0255	0.0264	0.0305	0.0275	0.0000	
GUAR-KG	0.0193	0.0208	0.0206	0.0208	0.0254	0.0226	0.0456	0.0000

Table 5. Distances matriz Fst obtained from comparisons between population pairs. * Pairs of populations exhibiting non-significant Fst P-values (> 0.05). BAMA: Buenos Aires Metropolitan Area, Argentina (N= 85); CDB: Córdoba, Argentina (N= 100); ITAL: Italy (N= 163); SPN: Spain (N= 164); MOZB: Mozambique (N= 102); EC-GUIN: Ecuatorial Guinea (N= 112); TOB-CH: Toba Chaco, Argentina (N= 49); GUAR-KG: Guarani-Kaingang, Brazil (N= 46).

Tabla 5.Distancias matriz Fst obtenidas mediante comparación entre pares de poblaciones. * Pares de poblaciones que muestran valores de Fst-P no significativos (> 0.05). BAMA: Área Metropolitana de Buenos Aires, Argentina (N= 85); CDB: Córdoba, Argentina (N= 100); ITAL: Italia (N= 163); SPN: España (N= 164); MOZB: Mozambique (N= 102); EC-GUIN: Guinea Ecuatorial (N= 112); TOB-CH: Toba Chaco, Argentina (N= 49); GUAR-KG: Guarani-Kaingang, Brasil (N= 46).

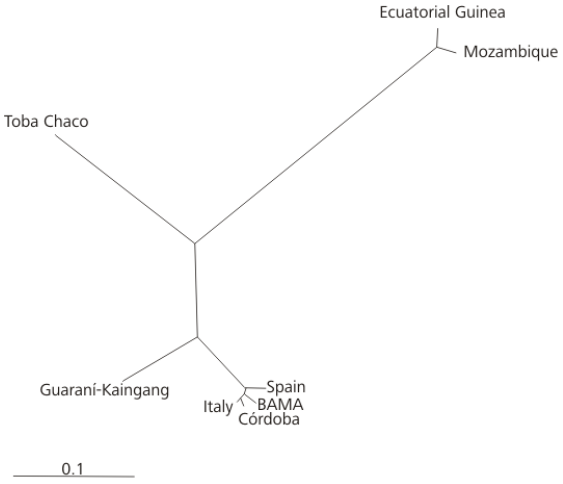

Figure 2. UPGMA tree based on Fst values between pairs of populations.

Figura 2. Fenograma UPGMA basado en los valores Fst entre pares de poblaciones.

Place of birth	Paternal Grandfather (N)	%
BAMA/BsAs Prov.	18	28.6
Centre	6	9.5
NWA	3	4.8
NEA	5	7.9
Cuyo	5	7.9
Bordering countries	5	7.9
Spain	6	9.5
Italy	12	19
Others Europe	3	4.8
Total *	63	100

Table 6. Birth place of donors' paternal grandfather.

Tabla 6. Lugar de nacimiento del abuelo paterno del donante.

composition, mainly of Italian and Spanish origin.

Genealogical data

The data obtained from the Y-STR haplotypes was compared with the genealogical data of the donors, focusing our analysis on the geographic origin of the paternal grandparents that have contributed to those lineages, and the proportion in which they have done it. In Table 6 can be observed that 28.6% of the grandparents were born in the BAMA and in Buenos Aires Province (by historical reasons they would be mainly European descendants), while 33.3% are natives of Europe, with a high prevalence of Italians (19%). Meanwhile, 30.1% of the men ancestors correspond to migrants from the argentine provinces, mainly from the center of the country (9.5%). Five foreigners came from bordering countries, 3 were born in Paraguay, 1 in Chile and 1 in Uruguay. Also, Figure 3 shows that there is generally a noticeable relation between the grandparent's birthplace and the assigned geographical origin of the Y-chromosome lineages. In this sense we observed that the originating ancestors of Cuyo an Argentine Northwest make up the aggregate of Amerindian paternal haplotypes, while those who were born in Buenos Aires, in the Center, Northeast, and in different regions of Europe mainly present profiles associated to this last continent and to the Middle East. On the other hand, the unique sub-Saharan lineage comes from Paraguay. These data, together with the historical and demographic information available, allow us to state that the Amerindian and sub-Saharan contribution observed in men from the BAMA, would be the result of recent migrations, initiated in the middle of twentieth century, principally from the north of the country and bordering countries, of high indigenous and, to a lesser extent, African composition.

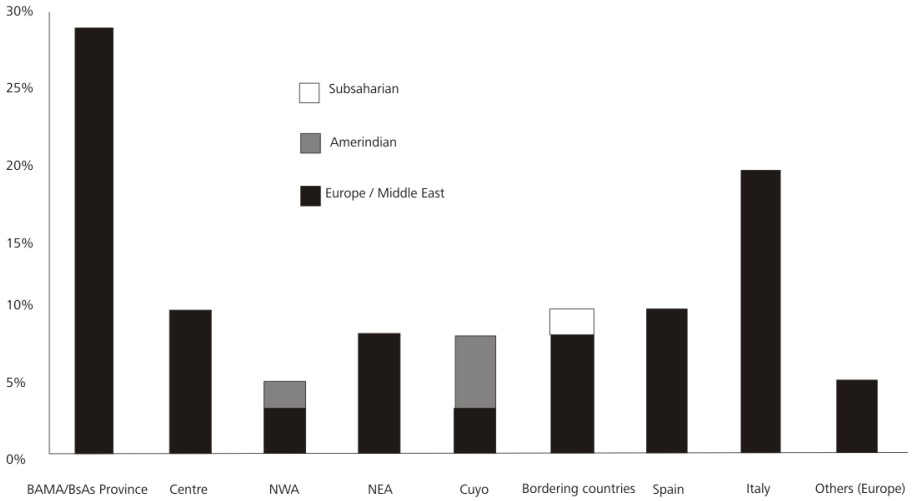

Figure 3. Birthplace of paternal grandparents and geographic origin assigned to the corresponding Y-chromosome haplotypes and donors.

Figura 3. Lugar de nacimiento del abuelo paterno y origen geográfico asignado correspondiente a los haplotipos del cromosoma Y de los donantes.

Conclusion

Previous studies carried out in the BAMA have shown that the population genetic composition in this area is product of the contributions of different geographical groups. Nevertheless, in this work we have demonstrated that the input of paternal lineages in the BAMA presents a genetic diversity level similar to that observed in other cosmopolitan populations of Europe and Argentine, showing a genetic homogeneity with European roots, principally arising from Italy and Spain. Also, the low proportion of Amerindian paternal haplotypes, and the high number of maternal lineages found before in the same sample, reveals that there has been a differential input by gender in the history of this population, in the sense of an important contribution of native women to the mixing process, which seems to have been the rule in the cosmopolitan population of Latin America.

The historical, demographic and genealogical information allowed us to state that the native and sub-Saharan paternal participation would be explained to a great extent by the migratory movements occurred since the middle of twentieth century, mainly arising from the Argentine north and west, and also from the bordering countries, of high native American incidence, and, to a less extent, African composition. This illustrates the importance of considering the genealogical data for genetical ancestry studies. Finally, it should be stated that the data obtained from this work is also of interest in the field of forensic anthropology.

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