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Gender bias in the multiethnic genetic composition of central Argentina

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Abstract A sample of central Argentina (Córdoba) was genotyped for the first hypervariable region (HVS-I) plus a set of coding region mitochondrial DNA (mtDNA) single nucleotide polymorphisms (SNPs) ($N = 102$) and compared with a data set of Y-chromosome short tandem repeats (Y-STRs; $N = 100$) previously genotyped in the same individuals. We additionally compiled a database containing more than 4,000, 6,800, and 12,000 HVS-I sequences of Native American, sub-Saharan African, and European origin, respectively. The Y-Chromosome Haplotype Reference Database (YHRD) was used as a reference for the Y-STR profiles from Córdoba. The Native American component is highly prevalent on the maternal side (~41%) in contrast to the Y-chromosome paternal contribution (~2%), indicating a strong gender bias in the colonization and admixture processes that occurred in the recent history of Argentina, in agreement with historical records. The demographic input of African slaves in Córdoba was very high in the eighteenth century (~40% of the total population) but decreased dramatically after a few decades; therefore, the minor traces of sub-Saharan Y-chromosome and mtDNA lineages observed in our sample fit well with these historical records. The European Y-chromosome component of Córdoba (~97%; in contrast to the 57% observed in the mtDNA side) also mirrors the substantial immigration experienced by Argentina during the beginning of the last century, predominantly from Italy and Spain.

Keywords Coding region · SNP · Haplotype · Native American · Phylogeny

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Introduction

The present population of central Argentina (Córdoba) is the result of a complex amalgamation of different cultures and populations with different genetic ancestries. Today, various Native American groups exist that inhabit different regions of Córdoba. A small population of Comechingones (actual population size of ~5,000) still live in a relatively isolated region of the mountain ranges of Córdoba, whereas the Olongastas, considered a subgroup of the Diaguitas (~6,000 individuals), live at lower altitudes in the northwest of the province. Other groups, such as the Sanavirones in the northeast and the Pampeanos (formerly occupying the low flat areas of the Argentinean humid *pampa*), have since become extinct. Although some of these populations have preserved their folklore and other cultural traditions relatively well, none have retained their original languages and now speak Spanish exclusively.

During colonial times, the use of slave labor for agricultural development became an economic necessity for Spanish colonialists. The relatively low density of Native Americans coupled with the resistance of these population

groups to Spanish acculturation and slavery led to the introduction of very large numbers of sub-Saharan Africans to the region. Although the demographic impact of Africans on the autochthonous population in other Latin American countries such as Colombia and Brazil was extremely high and still persists in modern populations (Alves-Silva et al. 2000; Beleza et al. 2005; Ely et al. 2006; Parra et al. 2001; Salas et al. 2005b, c; 2004), their real demographic impact in Argentina is a debatable topic. Some scholars maintain that a very significant number of African slaves were forcedly moved to Argentina, in particular to regions of intense agricultural activity: according to Victoria-Gomes (2002), in 1778 the population of Africans in Córdoba was ~44%, whereas in 1887 the official percentage of Africans was reduced to ~1.8%. The reasons for such a dramatic decrease remain uncertain; Victoria-Gomes (2002) pointed to epidemic causes (e.g., yellow fever) and colonial fights against neighboring populations (the number of Africans recruited in the army was disproportionately large compared with other ethnic groups).

During the nineteenth century, Argentina experienced a large-scale immigration of Europeans, notably from Italy and Spain (but including UK and Germany), which dramatically changed the demography of the country, mainly in urban areas. There are other well-known minorities in this region, including Jews (escaping persecution in the Second World War), Arabs, Armenians, and Japanese. Since Córdoba is one of the most important industrial centers of the country, over the past 50 years, it has attracted numerous immigrants from all over the country and from neighboring countries (i.e., Bolivia, Paraguay, etc.).

Finally, none of the existing Argentinean Native American groups live a completely isolated existence, and the degree of admixture, in particular with individuals of primary European ancestry, seems to be high in some populations. However, the magnitude of this admixture and the final demographic impact of African slaves in the region has not been properly evaluated.

We collected a sample of central Argentina (Córdoba) to estimate the different main ancestries that contributed to the present population. We analyzed mtDNA variation by sequencing the first hypervariable region (HVS-I) and a set of mtDNA coding region single nucleotide polymorphisms (SNPs). The degree of admixture on the maternal side was also contrasted with analysis of a set of Y-chromosome short tandem repeats (Y-STR) markers genotyped in the same individuals (Fondevila et al. 2003), firstly by inferring the haplogroup status of these Y-STR profiles, and secondly by searching these profiles in the worldwide Y Chromosome Haplotype Reference Database (YHRD).

Material and methods

Samples

We collected 102 healthy unrelated individuals from the province of Córdoba in central Argentina. Informed consent was given by all participants. The protocol and procedures employed were reviewed and approved by the review committee of the University of Santiago de Compostela, Spain, where genotyping was carried out. All persons gave their informed consent prior to inclusion.

PCR amplification and sequencing

All samples were amplified and sequenced (forward and reverse) for the HVS-I, analyzing the sequence range 16024–16400. Polymerase chain reaction (PCR) amplification was performed with GeneAmp 9700 thermocyclers, and sequencing analysis was performed as previously described (Álvarez-Iglesias et al. 2007). All samples were additionally genotyped for a set of ten SNPs following (Quintáns et al. 2004). Mutations are referred to the revised Cambridge Reference Sequence (rCRS) (Andrews et al. 1999). We followed a standardized forensic framework for nomenclature, as indicated in Carracedo et al. (2000) but with slight modifications considered in Salas et al. (2005a) concerning insertions. Data were checked following the phylogenetic principles described previously (Bandelt 1994; Bandelt et al. 2004a, b; Salas et al. 2007) to avoid sequence artifacts as much as possible.

Databases and statistical analysis

For phylogeographic purposes, we collected different available population data sets from the literature. Thus, for African haplotypes, we used roughly the same database reported in Černý et al. (2007), which consists of 6,856 profiles from different regions of the African continent. For Native American lineages, an HVS-I database that consists of 4,086 available sequences in the literature was employed, and for the western European database, we considered >12,000 publicly available profiles.

Only the HVS-I segment was used for population comparison, in particular, the 16090–16365 sequence range, as this is the common segment for the different populations used. Nomenclature of African haplotypes follows Salas et al. (2002, 2004), with updates in Černý et al. (2007), Kivisild et al. (2004), and Torroni et al. (2006). For Native American haplogroups, we use the most updated nomenclature from Bandelt et al. (2003) and Kong et al. (2006), and for the European profiles, Achilli et al. (2005, 2004), Loogväli et al. (2004), and Sun et al. (2006), among others. DnaSP 4.10.3 software (Rozas et al. 2003) was used for computation of different diversity indices.

Principal component analysis was performed using Stata 9.1 (http://www.stata.com/).

Y-chromosome haplogroup inference

The number of occurrences of particular profiles in a worldwide database roughly indicates their most natural geographical origin. Minimal Y-chromosome haplotypes for 100 individuals were reported in Fondevila et al. (2003) and submitted to the YHRD (http://www.yhrd.org/index.html). We searched the YHRD for each of these profiles (YHRD; release 22) with particular focus on the main European source populations of Spain and Italy together with the major continental regions (e.g., Europe, Latin America, etc.), keeping the population grouping scheme provided by the YHRD. Haplogroup Predictor (https://home.comcast.net/~hapest5/index.html) was used for inferring haplogroup status of Y-STR profiles using flat a priori probabilities.

Results and discussion

Phylogeography of mtDNA HVS-I profiles

The Central Argentinean population has two well-differentiated mtDNA ancestral components: ~57% of the lineages are of European origin (predominantly from western Europe), and ~41% are typically Native American (Fig. 1). Only two individuals carried mtDNA of sub-Saharan provenance. About 31% of European lineages belong to haplogroup H, but there are representatives of other common European haplogroups (HV0, I, J, etc.), broadly reflecting the haplogroup spectra of a typical European population (Table 1). At the haplotypic level, the degree of molecular resolution does not allow the phylogeographic allocation of these European lineages to particular regions of Europe; in fact, most of these HVS-I profiles in Córdoba occur across the whole of Europe. There are only few outlined exceptions; for instance, we did not find matches for the haplogroup H profile C16111T T16209C C16270T. Some lineages have some more restricted geographical occurrence. For example, there are seven matches for the haplogroup W sequence C16173T C16223T C16292T T16325C T16352C; curiously, all of them are in Romania (Brandstätter et al. 2007) and Georgia (Quintana-Murci et al. 2004).

With regard to the Native American component, we observed representatives of the four main haplogroups, A2 (~8%), B2 (~5%), C1 (~14%), and D1 (~14%). Five out of nine of the A2 haplotypes match with the basal A2

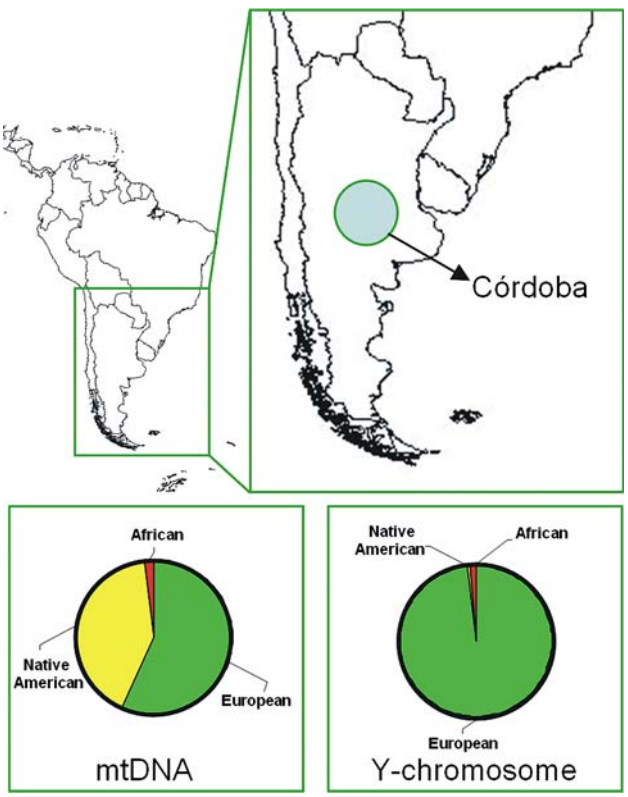

Fig. 1 Location of the sample analyzed and main ancestry of Y–chromosome and mitochondrial DNA lineages

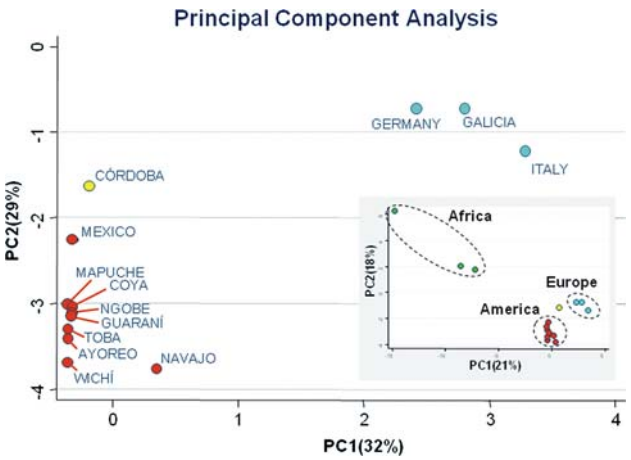

Fig. 2 Principle component analysis (PCA) based on mtDNA haplogroup frequencies. The main PCA plot includes several European and Native American samples plus the one from Córdoba analyzed in this study, whereas the nested small PCA plot includes three additional sub-Saharan African samples. References for population samples are as follows: **a** Africa (*green dots*): Mozambique (Salas et al. 2002), Angola (Plaza et al. 2004), Cabinda (Beleza et al. 2005); **b** Europe (*blue dots*): Italy (Bini et al. 2003), Galicia from Spain (Salas et al. 1998), and Germany (Lutz et al. 1998); and **c** America (*red dots*): Mexico (Green et al. 2000), Mapuche from Chile (Moraga et al. 2000), Guarani Kaiowá from Brazil (Marrero et al. 2007), Coya (Álvarez-Iglesias et al. 2007), Toba and Wichí from Argentina (Cabana et al. 2006), Navajo (Monson et al. 2002), Ayoreo from Bolivia and Paraguay (Dornelles et al. 2004), Ngobe from Panamá (Kolman et al. 1995), Córdoba (this study)

Table 1 First hypervariable region (HVS-I) and mitochondrial DNA (mtDNA) single nucleotide polymorphism (SNP) profiles in 102 Argentineans from Córdoba

Sample ID	HVS-I (minus 16000)	HG	Start	End	mtDNA SNPs									
					4216T	4529A	4580G	7028C	10398A	10400C	10873T	12308A	12705C	14766C
1	051 086 092 111 223 290 319	A2	16024	16400	–	–	–	T	–	–	–	–	T	T
2	092 111 172 218 223 290 319 362	A2	16024	16390	–	–	–	T	–	–	–	–	T	T
3	111 172 223 290 319 362	A2	16024	16390	–	–	–	T	–	–	–	–	T	T
4	111 172 223 290 319 362	A2	16024	16390	–	–	–	T	–	–	–	–	T	T
5	111 223 290 319 362	A2	16024	16400	–	–	–	T	–	–	–	–	T	T
6	111 223 290 319 362	A2	16024	16400	–	–	–	T	–	–	–	–	T	T
7	111 223 290 319 362	A2	16024	16400	–	–	–	T	–	–	–	–	T	T
8	111 223 290 319 362	A2	16024	16390	–	–	–	T	–	–	–	–	T	T
9	111 223 290 319 362	A2	16024	16383	–	–	–	T	–	–	–	–	T	T
10	093 183C 189 217 359	B2	16024	16390	–	–	–	T	–	–	–	–	–	T
11	142 183C 189 217	B2	16024	16400	–	–	–	T	–	–	–	–	–	T
12	153 183C 189 217	B2	16024	16390	–	–	–	T	–	–	–	–	–	T
13	173 182C 183C 189 217	B2	16024	16390	–	–	–	T	–	–	–	–	–	T
14	183C 189 217	B2	16024	16390	–	–	–	T	–	–	–	–	–	T
15	051 093 223 259 293C 298 325 327 357	C1	16024	16380	–	–	–	T	G	T	C	–	T	T
16	051 129 223 298 325 327	C1	16024	16370	–	–	–	T	G	T	C	–	T	T
17	071 223 298 311 325 327 368	C1	16024	16390	–	–	–	T	G	T	C	–	T	T
18	092 223 298 325 327	C1	16024	16383	–	–	–	T	G	T	C	–	T	T
19	092 223 298 325 327	C1	16024	16400	–	–	–	T	G	T	C	–	T	T
20	154 223 298 325 327	C1	16024	16390	–	–	–	T	G	T	C	–	T	T
21	183C 189 223 298 325 327	C1	16024	16390	–	–	–	T	G	T	C	–	T	T
22	223 234 298 325 327	C1	16024	16390	–	–	–	T	G	T	C	–	T	T
23	223 298 325 327	C1	16024	16390	–	–	–	T	G	T	C	–	T	T
24	223 298 325 327	C1	16024	16400	–	–	–	T	G	T	C	–	T	T
25	223 298 325 327	C1	16024	16390	–	–	–	T	G	T	C	–	T	T
26	223 298 325 327	C1	16024	16400	–	–	–	T	G	T	C	–	T	T
27	223 298 325 327 335	C1	16024	16400	–	–	–	T	G	T	C	–	T	T
28	223 298 325 327 368	C1	16024	16390	–	–	–	T	G	T	C	–	T	T
29	093 187 189 209 223 325 362	D1	16024	16383	–	–	–	T	G	T	C	–	T	T
30	126 223 325 362	D1	16024	16380	–	–	–	T	G	T	C	–	T	T
31	126 223 325 362	D1	16024	16394	–	–	–	T	G	T	C	–	T	T
32	187 189 209 223 325 362	D1	16024	16383	–	–	–	T	G	T	C	–	T	T
33	187 189 209 223 325 362	D1	16024	16390	–	–	–	T	G	T	C	–	T	T
34	189 223 325 362	D1	16024	16390	–	–	–	T	G	T	C	–	T	T
35	190 223 325 362	D1	16024	16390	–	–	–	T	G	T	C	–	T	T
36	223 242 311 325 362	D1	16024	16390	–	–	–	T	G	T	C	–	T	T
37	223 242 311 325 362	D1	16024	16400	–	–	–	T	G	T	C	–	T	T
38	223 242 311 325 362	D1	16024	16400	–	–	–	T	G	T	C	–	T	T
39	223 242 311 325 362	D1	16024	16390	–	–	–	T	G	T	C	–	T	T
40	223 242 311 325 362	D1	16024	16390	–	–	–	T	G	T	C	–	T	T
41	223 242 311 325 362	D1	16024	16383	–	–	–	T	G	T	C	–	T	T
42	223 242 311 325 362	D1	16024	16383	–	–	–	T	G	T	C	–	T	T
43	093	H	16024	16383	–	–	–	–	–	–	–	–	–	–

Table 1 continued

Sample ID	HVS-I (minus 16000)	HG	Start	End	mtDNA SNPs									
					4216T	4529A	4580G	7028C	10398A	10400C	10873T	12308A	12705C	14766C
44	189	H	16024	16384	–	–	–	–	–	–	–	–	–	–
45	189	H	16024	16390	–	–	–	–	–	–	–	–	–	–
46	189	H	16024	16400	–	–	–	–	–	–	–	–	–	–
47	189	H	16024	16390	–	–	–	–	–	–	–	–	–	–
48	304	H	16024	16380	–	–	–	–	–	–	–	–	–	–
49	304	H	16024	16390	–	–	–	–	–	–	–	–	–	–
50	362	H	16024	16390	–	–	–	–	–	–	–	–	–	–
51	111 209 270	H	16024	16390	–	–	–	–	–	–	–	–	–	–
52	172 192	H	16024	16390	–	–	–	–	–	–	–	–	–	–
53	189 301	H	16024	16390	–	–	–	–	–	–	–	–	–	–
54	193 219 362	H	16024	16383	–	–	–	–	–	–	–	–	–	–
55	293 311	H	16024	16390	–	–	–	–	–	–	–	–	–	–
56	rCRS	H	16024	16400	–	–	–	–	–	–	–	–	–	–
57	rCRS	H	16024	16395	–	–	–	–	–	–	–	–	–	–
58	rCRS	H	16024	16390	–	–	–	–	–	–	–	–	–	–
59	rCRS	H	16024	16383	–	–	–	–	–	–	–	–	–	–
60	rCRS	H	16024	16383	–	–	–	–	–	–	–	–	–	–
61	068 288	H8	16024	16380	–	–	–	–	–	–	–	–	–	–
62	172 311	HV	16024	16384	–	–	–	T	–	–	–	–	–	–
63	298	HV0	16024	16400	–	–	–	T	–	–	–	–	–	–
64	298	HV0	16024	16390	–	–	–	T	–	–	–	–	–	–
65	298	HV0	16024	16380	–	–	–	T	–	–	–	–	–	–
66	298	HV0	16024	16384	–	–	–	T	–	–	–	–	–	–
67	093 221 298	HV0	16024	16383	–	–	–	T	–	–	–	–	–	–
68	189 235 298	HV0	16024	16390	–	–	–	T	–	–	–	–	–	–
69	218 239A 298	HV0	16024	16390	–	–	–	T	–	–	–	–	–	–
70	093 129 223 391	I	16024	16380	–	–	–	T	G	–	–	–	T	T
71	129 148 192 223 294	I	16024	16383	–	T	–	T	G	–	–	–	T	T
72	129 192 223	I	16024	16383	–	–	–	T	G	–	–	–	T	T
73	069 093 126	J	16024	16390	C	–	–	T	G	–	–	–	–	T
74	069 126	J	16024	16380	C	–	–	T	G	–	–	–	–	T
75	069 126	J	16024	16380	C	–	–	T	G	–	–	–	–	T
76	069 126 183C 189	J	16024	16380	C	–	–	T	G	–	–	–	–	T
77	069 126 189	J	16024	16390	C	–	–	T	G	–	–	–	–	T
78	069 126 193 278	J	16024	16380	C	–	–	T	G	–	–	–	–	T
79	069 126 274	J	16024	16400	C	–	–	T	G	–	–	–	–	T
80	069 126 145 231 261	J1a	16024	16400	C	–	–	T	G	–	–	–	–	T
81	069 126 145 231 261	J1a	16024	16390	C	–	–	T	G	–	–	–	–	T
82	069 126 145 222 256 261 278	J1b	16024	16400	C	–	–	T	G	–	–	–	–	T
83	093 224 234 311	K	16024	16400	–	–	–	T	–	–	–	G	–	T
84	224 234 311	K	16024	16390	–	–	–	T	–	–	–	G	–	T
85	224 293 311	K1	16024	16390	–	–	–	T	G	–	–	G	–	T
86	224 311	K1	16024	16400	–	–	–	T	G	–	–	G	–	T

Table 1 continued

Sample ID	HVS-I (minus 16000)	HG	Start	End	mtDNA SNPs									
					42 16 T	45 29 A	45 80 G	70 28 C	103 98 A	104 00 C	108 73 T	123 08 A	127 05 C	147 66 C
87	224 311	K1	16024	16383	–	–	–	T	G	–	–	G	–	T
88	224 311	K1	16024	16390	–	–	–	T	G	–	–	G	–	T
89	126 187 189 193 223 264 270 278 293 311	L1b1	16024	16390	–	–	–	T	G	–	C	–	T	T
90	129 163 187 189 209 223 278 293 294 311 360	L1c1	16024	16386	–	–	–	T	G	–	C	–	T	T
91	126 172 294 304	T	16024	16383	C	–	–	T	–	–	–	–	–	T
92	086 126 163 189 294	T1	16024	16380	C	–	–	T	–	–	–	–	–	T
93	126 163 186 189 294	T1	16024	16390	C	–	–	T	–	–	–	–	–	T
94	126 163 186 189 294	T1	16024	16391	C	–	–	T	–	–	–	–	–	T
95	126 163 186 189 294	T1	16024	16384	C	–	–	T	–	–	–	–	–	T
96	126 294 296 304	T2	16024	16390	C	–	–	T	–	–	–	–	–	T
97	192 256 270 362	U5a	16024	16390	–	–	–	T	–	–	–	G	–	T
98	239 256 270	U5a	16024	16390	–	–	–	T	–	–	–	G	–	T
99	146 162 342	U8a	16024	16380	–	–	–	T	–	–	–	G	–	T
100	173 223 292 325 352	W	16024	16400	–	–	–	T	–	–	–	–	T	T
101	223 292	W	16024	16383	–	–	–	T	–	–	–	–	T	T
102	111 189 223 278	X	16024	16400	–	–	–	T	–	–	–	–	T	T

A dash in the SNP columns indicates a match with the revised Cambridge Reference Sequence (rCRS) (Andrews et al. 1999)

haplotype, which is common across the American continent. There are four instances of the basal C1 haplotype in Córdoba, also common in populations living at different continental latitudes. In contrast, the basal HVS-I haplotype of D1 (T16362C T16325C) was not detected in our sample. However, we found 11 identical matches of the most common D1 haplotype C16223T C16242T T16311C T16325C T16362C, ten of them in other Argentinean populations: four matches in the Mapuches (Ginther et al. 1993) and the rest in the Pilagá and Wichí (Cabana et al. 2006) and in the Coyas (Álvarez-Iglesias et al. 2007). Two other matches were also observed in the Genographic database (https://www3.nationalgeographic.com/genographic/resources.html). There are other interesting matches for the D1 haplotypes of Córdoba. Haplotype C16187T T16189C T16209C C16223T T16325C T16362C appears four times in the database, three of them in the aboriginal and in the general Chilean population (Horai et al. 1993; Moraga et al. 2000) and one in the ancient Kaweskar DNA samples (Patagonia-Tierra del Fuego; South of Argentina, and Chile) studied by García-Bour et al. (2004).

As shown in Table 2, for the sequence range 16090–16385, there are 24 (out of 42) different Native American haplotypes in Córdoba. Most of them ($N = 17$) are only observed once, four sequences appear twice, two sequences occur five times, and one haplotype appears seven times (C16223T C16242T T16311C T16325C T16362C). Sequence (S) and nucleotide (π) diversities and average number of nucleotide diversity (M) are quite high for the Native American component ($S = 0.948$; $\pi = 0.01767$; $M = 6.04$) in contrast to the average values for the continental Native American component ($S = 0.945$; $\pi = 0.01519$; $M = 3.43$), indicating that the admixture process with Europeans was relatively gradual, allowing the preservation of a significant part of the Native American original gene pool in today's general population of Córdoba. There are 13 identical matches between the Native American lineages and South America: nine in central America and ten in North America. However, six haplotypes are found across America (North, Central, and South) (Table 2). Moreover, there are nine haplotypes with only one representative in Córdoba that were still not observed in our large database.

We only detected two sequences of sub-Saharan origin belonging to haplogroups L1b1 and L1c1. For the L1b1 sequence (see Table 1), we did not find exact matches, but there are some one-step-mutation neighbors—such as

Table 2 Córdoba first hypervariable region (HVS-I) Native American sequences (haplogroups A2, B2, C1, and D1) matches in the main American continental regions (North, Central, South)

HVS-I sequences	Haplogroup	North ($N = 2,005$)	Central ($N = 485$)	South ($N = 1,596$)	Córdoba ($N = 42$)	Total ($N = 4,128$)
092 111 172 218 223 290 319 362	A2	–	–	–	1	1
092 111 223 290 319	A2	–	–	–	1	1
111 172 223 290 319 362	A2	1	1	–	2	4
111 223 290 319 362	A2	161	45	50	5	261
093 189 217 359	B2	–	–	–	1	1
142 189 217	B2	–	–	–	1	1
153 189 217	B2	–	–	–	1	1
173 189 217	B2	–	–	–	1	1
189 217	B2	24	38	90	1	153
092 223 298 325 327	C1	–	1	1	2	4
093 223 259 293A 298 325 327 357	C1	–	–	–	1	1
129 223 298 325 327	C1	5	3	10	1	1
154 223 298 325 327	C1	–	–	1	1	2
189 223 298 325 327	C1	4	–	1	1	6
223 234 298 325 327	C1	3	–	–	1	4
223 298 311 325 327	C1	5	4	4	1	14
223 298 325 327	C1	111	19	192	5	327
223 298 325 327 335	C1	1	–	1	1	3
093 187 189 209 223 325 362	D1	–	–	–	1	1
126 223 325 362	D1	–	–	–	2	2
187 189 209 223 325 362	D1	–	–	4	2	6
189 223 325 362	D1	3	7	8	1	19
190 223 325 362	D1	–	–	1	1	2
223 242 311 325 362	D1	–	1	5	7	13
Totals		318	119	368	42	829

Only the sequence range 16090–16365 was considered here. Length variation around 16189 and variants 16182C and 16183C were omitted from the comparison

A16166G on top of T16126C C16187T T16189C C16193T C16223T C16264T C16270T C16278T A16293G T16311C, which is found in Cabinda (Beleza et al. 2005)—in the Bakaka of south Cameroon (Coia et al. 2005) but also in the Tacuarembó from Uruguay. The typical central African L1c1 lineage G16129A A16163G C16187T T16189C T16209C C16223T C16278T A16293G C16294T T16311C C16360T also appears at moderate frequencies in Cabinda (Beleza et al. 2005) and other American countries.

Principal component analysis

We carried out a principal component analysis (PCA) based on mtDNA haplogroup frequencies and using several European, sub-Saharan African and two Native American data sets as population references (see Fig. 2 legend). PC1 (which accounts for 21% of the variability) primarily separates African from non-African populations, whereas PC2 (18%) basically splits European from Native American samples (see nested plot in Fig. 1). The plot also shows a clear-cut heterogeneity pattern in the African samples. In contrast, the Native American groups are tightly grouped in a single cluster, reflecting the fact that most of them essentially carry Native American lineages. A second round of PC analysis (the two first PCs accounting for 61% of the variability) was carried out excluding the African samples (main plot in Fig. 2). PC1 (32%) shows in one pole the European samples and in the other extreme the Native Americans ones. The European component of the Córdoba sample (57%) is clearly reflected in the PC2 (29%), as it is also the case for the Mexican data set (Green et al. 2000), which also has an important European mtDNA background ($\sim$15%).

The most likely ancestry of Y-STR profiles

Inference of the haplogroup status based on the Bayes approach implemented in Haplogroup Predictor showed that $\sim$70% of the profiles yielded a posteriori

Table 3 Inference of haplogroup status of Y-chromosome short tandem repeat (Y-STR) profiles in central Argentineans and number of matches in the Y-Chromosome Haplotype Reference Database (YHRD)

Sample	Y-STRs								No.	HG	Prob.	Worldwide	Europe	Spain	Italy	Latin-Am.	Asia	Africa
ID[a]	319	385	389I	389II	390	391	392	393				*N* = 5,0867	(*N* = 26,305)	(*N* = 1,708)	(*N* = 2,285)	(*N* = 7,035)	(*N* = 11,439)	(*N* = 2,750)
01	14	11–14	14	30	24	11	13	13	1	R1b	99.9	313	154	47	9	114	–	4
02	14	12–14	14	30	24	11	13	14	1	R1b	86.7	2	1	1	–	1	–	–
03	14	11–14	13	30	24	11	13	13	3	R1b	100	196	105	21	11	55	1	6
04	15	11–14	13	28	24	10	11	13	1	R1a	89.1	3	–	–	–	3	–	–
05	15	14–16	13	29	23	11	13	13	1	I1b2a	33.9	1	–	–	–	1	–	–
06	14	11–14	13	29	23	10	13	13	1	R1b	100	189	139	11	8	29	–	5
07	14	11–14	13	29	24	11	13	13	4	R1b	100	1045	637	126	70	276	3	33
08	14	11–11	13	29	24	11	13	13	1	R1b	100	64	45	4	5	8	1	–
09	15	14–17	14	30	23	10	11	12	1	J2	58.4	4	2	–	–	1	1	–
10	14	13–20	13	30	23	11	11	12	1	J1	97.9	4	–	–	–	2	–	2
11	13	16–18	13	30	25	10	11	13	1	E3b	100	35	32	1	5	3	–	–
12	14	11–14	14	30	24	11	14	13	1	R1b	53.1	14	7	1	1	7	–	–
13	13	16–16	13	30	24	10	11	13	1	E3b	99.9	21	11	3	2	6	–	–
14	14	11–14	13	30	24	10	13	13	1	R1b	99.9	94	47	4	4	19	2	4
15	15	11–14	13	29	24	10	13	12	1	R1b	99.8	8	5	1	–	2	1	
16	14	11–12	13	29	24	11	13	13	1	R1b	99.9	14	7	1	1	5	–	–
17	14	11–14	12	28	23	10	11	13	1	I1a	96.4	2	1	–	1	1	–	–
18	15	14–14	13	29	23	11	13	13	1	R1b	96.3	1	–	–	–	1	–	–
19	17	11–14	13	31	25	11	11	13	1	R1a	100	35	28	–	–	2	–	4
20	14	11–14	13	29	23	11	13	13	3	R1b	100	443	325	20	16	49	1	15
21	14	13–14	12	28	23	10	10	13	1	I1a	99.5	1	–	–	–	1	–	–
22	16	12–14	12	29	22	10	10	12	1	G2	97.6	1	–	–	–	1	–	–
23	15	12–13	13	29	23	10	13	13	1	R1b	99.6	1	–	–	–	1	–	–
24	14	11–15	14	30	24	11	13	13	1	R1b	100	93	49	19	6	18	15	2
25	14	12–14	13	29	23	10	13	13	1	R1b	99.5	22	17	2	–	4	–	–
26	14	11–14	13	28	24	10	14	13	1	R1b	98.4	5	3	–	1	1	–	–
27	13	13–15	14	30	23	10	13	12	1	J1	84.1	1	–	–	–	1	–	–
28	16	13–17	12	28	25	10	11	12	1	J2	98.6	4	1	–	–	2	1	–
29	14	12–14	13	29	24	11	13	13	2	R1b	100	126	79	17	9	27	–	6
30	17	12–12	14	29	25	9	10	13	1	I1b1	100	1	–	–	–	1	–	–
31	14	14–16	12	30	22	10	10	15	1	G2	99.4	1	–	–	–	1	–	–
32	14	12–13	13	29	24	11	13	13	1	R1b	99.8	33	20	2	4	11	–	2
33	14	11–14	13	29	24	11	13	14	1	R1b	99.3	57	32	5	4	18	–	1
34	15	14–15	12	29	23	10	10	15	1	G2	98	1	–	–	–	1	–	–

Table 3 continued

Sample	Y-STRs								No.	HG	Prob.	Worldwide	Europe	Spain	Italy	Latin-Am.	Asia	Africa
ID[a]	319	385	389I	389II	390	391	392	393				*N* = 5,0867	(*N* = 26,305)	(*N* = 1,708)	(*N* = 2,285)	(*N* = 7,035)	(*N* = 11,439)	(*N* = 2,750)
35	14	10–13	12	28	24	10	14	13	1	R1b	85.5	1	–	–	–	1	–	–
36	17	12–13	13	28	23	10	11	13	1	I1b1	99.8	3	1	1	–	1	–	–
37	15	12–16	12	29	22	10	11	13	1	G2	90.4	3	–	–	–	2	–	–
38	14	13–13	13	30	23	11	11	12	1	J1	89.2	3	–	–	–	2	1	–
39	15	11–15	13	29	24	10	13	12	1	R1b	99.4	2	–	–	–	1	–	–
40	14	11–16	13	29	24	11	13	13	1	R1b	100	45	23	3	4	16	–	–
41	14	12–13	14	30	23	10	11	12	1	J1	52.3	1	–	–	–	1	–	–
42	14	10–10	13	30	24	12	12	13	1	R1b	100	1	–	–	–	1	–	–
43	14	11–14	14	30	23	11	13	13	1	R1b	99.6	101	55	3	4	25	–	13
44	17	13–16	12	28	25	10	11	13	1	J2	56.4	2	1	–	–	1	–	–
45	16	10–14	13	30	25	10	11	13	1	R1a	100	83	77	–	–	1	4	–
46	17	11–13	13	30	25	10	11	13	1	R1a	99.8	10	8	–	1	1	1	–
47	15	13–16	13	29	23	9	11	13	1	J2	87.5	7	5	1	–	2	–	–
48	14	11–11	14	31	24	11	11	13	1	R1a	82.1	1	–	–	–	1	–	–
49	15	14–14	12	29	22	10	13	14	1	G2	99.4	1	–	–	–	1	–	–
50	15	13–14	14	31	23	11	11	12	1	J2	85.3	1	–	–	–	1	–	–
51	14	11–14	13	31	24	10	13	14	1	R1b	97.9	5	1	–	–	4	–	–
52	17	11–12	13	28	23	10	11	13	2	I1b1	99.8	6	4	3	–	2	–	–
53	15	11–14	13	29	24	11	13	13	1	R1b	99.6	148	92	12	17	46	–	3
54	17	12–12	13	28	24	10	13	13	1	I1b1	100	1	–	–	–	1	–	–
55	17	9–18	13	30	21	10	11	15	1	E3a	95.8	1	–	–	–	1	–	–
56	14	13–15	13	29	24	10	13	13	1	R1b	66	4	2	–	–	1	–	–
57	15	11–12	13	29	24	11	13	13	1	R1b	99.5	5	–	–	–	4	–	–
58	14	13–15	13	30	23	10	11	12	1	J1	95	39	22	1	2	13	3	1
59	14	11–14	13	30	24	12	13	13	1	R1b	100	11	4	–	–	7	–	–
60	13	16–18	12	29	24	10	11	12	1	E3b	98.5	1	–	–	–	1	–	–
61	14	11–15	13	29	27	11	13	13	1	R1b	89.2	1	–	–	–	1	–	–
62	13	15–18	13	30	24	10	11	13	1	E3b	99.9	39	28	2	3	7	–	–
63	15	11–14	13	28	24	11	13	13	1	R1b	99.8	11	4	3	–	5	–	–
64	15	13–16	13	29	23	9	11	12	1	J2	95.8	59	41	2	7	8	4	–
65	13	16–18	13	30	23	10	11	13	1	E3b	99.9	20	12	–	6	4	1	1
66	13	11–14	14	30	25	12	13	13	1	R1b	99.6	1	–	–	–	1	–	–
67	14	12–12	13	29	24	12	13	13	1	R1b	99.7	1	–	–	–	1	–	–
68	14	11–14	15	31	24	10	13	13	1	R1b	99.9	4	–	–	–	3	–	–
69	14	11–15	13	30	24	10	13	13	1	R1b	99.9	33	22	3	1	6	–	1

Table 3 continued

Sample ID[a]	Y-STRs								No.	HG	Prob.	Worldwide	Europe	Spain	Italy	Latin-Am.	Asia	Africa
	319	385	389I	389II	390	391	392	393				*N* = 5,0867	(*N* = 26,305)	(*N* = 1,708)	(*N* = 2,285)	(*N* = 7,035)	(*N* = 11,439)	(*N* = 2,750)
70	14	12–15	13	30	24	11	13	13	1	R1b	99.6	10	4	–	–	4	–	–
71	17	11–14	13	30	25	11	11	13	1	R1a	100	43	35	–	–	1	6	1
72	14	11–13	14	30	24	11	13	13	1	R1b	97.5	21	12	4	–	8	–	–
73	13	16–18	13	29	24	10	11	13	1	E3b	99.9	16	10	–	3	4	1	–
74	15	12–14	12	28	21	10	11	14	1	G2	99.4	1	–	–	–	1	–	–
75	14	11–14	13	28	23	11	13	15	1	R1b	99	1	–	–	–	1	–	–
76	15	13–17	12	29	22	10	11	14	1	G2	98.3	3	2	–	2	1	–	–
77	17	11–13	13	30	25	11	11	13	1	R1a	99.7	22	17	–	–	1	2	1
78	14	16–16	13	30	24	10	15	13	1	Q	93.7	1	–	–	–	1	–	–
79	14	13–17	13	29	23	10	11	12	1	J1	77.9	24	17	2	4	3	3	–
80	14	14–14	12	28	22	10	11	13	1	I1a	99.5	151	138	1	1	7	–	1
81	14	11–14	12	27	24	10	13	13	1	R1b	100	5	2	–	–	2	–	1
82	15	11–14	13	29	25	11	11	13	1	R1a	99.9	44	35	–	–	1	6	–
83	15	14–15	12	29	23	10	11	15	1	G2	96.5	4	–	–	–	4	–	–
84	14	12–18	13	30	23	10	11	12	1	J1	95.9	4	1	–	1	1	2	–
85	14	11–14	12	28	25	10	13	14	1	R1b	99.7	2	1	–	–	1	–	–
86	14	11–15	13	29	24	11	13	13	1	R1b	100	279	179	22	18	44	8	7
87	14	11–14	12	28	24	10	13	13	1	R1b	100	29	16	3	3	10	–	–
88	14	11–16	13	29	24	11	13	14	1	R1b	99.7	3	1	–	–	2	–	–
89	16	14–14	12	28	22	10	11	15	1	G2	96.8	1	–	–	–	1	–	–
90	15	14–15	12	29	22	10	11	14	1	G2	99.7	40	24	2	3	4	2	6
91	14	13–18	13	30	23	10	11	12	1	J1	96.7	44	9	–	1	11	12	8

HG haplogroup, *Prob.* probability

[a] Note that the haplotypes considered here have already been reported in Y-Chromosome Haplotype Reference Database (YHRD) (Fondevila et al. 2003). Sample ID corresponds with those used in Fondevila et al. (2003)

probabilities >0.98 in a unique and well-defined haplogroup (Table 3). In contrast to the mtDNA variation, most Y-STR lineages (~97%) can be allocated to typical European haplogroups, whereas only a small fraction correspond to lineages of likely Native American (~2%) and African ancestry (~1%) (Fig. 1). The most common European haplogroup is R1b (42%). This lineage is supposed to have spread into the rest of Europe from Iberian and other southern European refugia after the Last Glacial Maximum and today is the most frequent Y-chromosome lineage in Europe. Some other typical Mediterranean clades are also present in our central Argentinean sample. For instance, J2 is of near-Eastern origin but today it is prevalent across Mediterranean coastal regions, including the Iberian Peninsula. Its sister clade, J1, is probably of a more southern European origin and it is now common in the Mediterranean coastal regions. G2 is the most common G lineage in western Europe and makes up 8–10% of several Mediterranean populations (Spain, Italy, Greece, and Turkey).

Y-STRs profiles were also searched in the YHRD to investigate the number of times a particular profile was previously observed in other worldwide populations. This procedure provides additional indication regarding their most natural geographical origin. Strikingly, 25% of the Y-STR profiles do not find a single match in the YHRD. In agreement with their inferred haplogroup status, most haplotypes are more frequently observed in Europe or in populations with important European ancestry (e.g., USA). In agreement with historical documentation, a substantial number of profiles, 29 and 28%, have a higher frequency in Spain and Italy, respectively. For instance, profile 14/11–14/13/30/24/11/13/13 (inferred haplogroup status = R1b) appears three times in Córdoba with 25 matches in Spain (1.5%). The number of profiles that match certain central European samples is relatively high; however, this may just reflect the fact that the YHRD is substantially enriched with profiles obtained from specific areas (the YHRD European data set comprises one third of profiles obtained from Germany and Poland).

Haplotype 27 is of Native American ancestry (a posteriori probability for its haplogroup Q status = 0.94). Profile 5 has a more ambiguous haplogroup allocation (e.g., a posteriori probability of belonging to haplogroup Q = 0.17). When searching the YHRD, we did not find a single match in the whole database; however, a total of seven one-step mutation derivatives are only observed in Americans. Finally, haplotype 55 belongs to the sub-Saharan haplotype E3a (a posteriori probability for its haplogroup status = 0.96), and there are neither matches nor one-step mutation profiles in the YHRD (which could again reflect sampling bias due to the limited presence of sub-Saharan samples in the database).

Final remarks

There is a clear gender bias in the mtDNA and Y-chromosome composition of central Argentina. PCA, based on mtDNA haplogroup frequencies, with the two first principal components accounting for 61% of the variability), together with phylogeographic inferences, clearly indicates the halfway position of the Córdoba population between Europeans and Native Americans. In contrast, most Y-chromosomes in the general population of Córdoba are of European origin, with evidence for an important input from Italy and Spain. This is in agreement with historical records indicating that between 1869 and 1991, the average contribution of Italians and Spanish was 34% and 22% of total newcomers, respectively (source: INDEC, Instituto Nacional de Estadísticas y Censo from Argentina; http://www.indec.gov.ar). The presence of mtDNA and Y-chromosome lineages of sub-Saharan origin in Córdoba is low (<2%) but fits well with the demographic inferences of another study (Victoria-Gomes 2002).

Our results contrast with those obtained for other Argentinean populations. For instance, Dipierri et al. (1998) show that the Native American component in two northwestern Argentinean populations is ~65%, the introgression being more evident on the Y-chromosome side with frequencies of ~28% in Quebrada de Humahuaca and ~64% in San Salvador de Jujuy. On the other hand, the population of La Plata (Argentina) shows a Native American component of ~46% and a paternal contribution of ~11% (Martinez-Marignac et al. 2004). In the very isolated Argentinean village of Acuña (Bailliet et al. 2001), mtDNA is mainly Native American, whereas the Y-chromosome part is essentially European. As in Córdoba, all the above-mentioned studies clearly indicate directional mating in Argentina. Moreover, all these studies indicate Argentina shows a clear pattern of population substructure on the specific maternal and paternal genomes, which also corroborates the findings of previous studies based on autosomal STR markers (Toscanini et al. 2006). The forensic field and medical genetic studies will benefit from population studies across the Argentinean territory that would allow detailed knowledge of population structure and its consequences when estimating the weight of forensic haploid evidence (Egeland and Salas 2008) or evaluating the possibility of spurious positive results in medical genetic studies (Salas and Carracedo 2007).

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