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Genetic attributes of the YHRD minimal haplotype in 10 provinces of Argentina

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Abstract

We investigated nine Y-STRs on 1136 unrelated males from 10 provinces and three aboriginal tribes of Argentina. The urban populations depicted 654 different haplotypes (66%) and a haplotype diversity (HD) value of 0.9967. Meanwhile, the Amerindian groups ranged from 20 to 26 different haplotypes (62.5–38.2%) and the HD values from 0.8635 to 0.9586. By AMOVA it was determined a remarkable homogeneous haplotype distribution, nevertheless a certain degree of genetic substructure was detected in the North region, in particular in Salta population. Genetic distance allowed to identify three clusters one of them included Salta population and the Amerindian tribes. The results presented herein showed the impact of the European male genetic contribution on the aboriginal gene pool that can be, at present, assessed by analyzing the nowadays extant population.

Keywords: Y-chromosome; Population structure; Y-STR haplotype; YHRD; Amerindian; Argentina

1. Introduction

The population of Argentina is known to be genetically heterogeneous, with an important Native American and European genetic contributions.

The country is composed by 24 provinces and a Federal District; and can be subdivided into four major regions: Northwestern, Northeastern, Central and Southern. Each of which has their own social and economical attributes influenced by the quality of the land, the climate, the aboriginal population that originally occupied the areas and the influence of migratory waves that settled in each region. At present, they are inhabited by admixed populations whose genetic contribution was provided by the Amerindian aboriginal populations that arrived to the nowadays territory of Argentina around 12,000 years before present [1–3], and the different migratory waves from Europe. This immigration flow started at the moment of the Spanish conquest, occurred in 1536 and increased in the eighteenth and nineteenth centuries.

The analysis of uniparental markers, like those harbored onto the non-recombining portion of the Y-chromosome, provides a unique system for the study of the human origins, migration and admixture. These markers have been proven to be valuable tools for the study of population history and to determine genetic distances between populations [4–6].

The Y-chromosome has special features; is in haploid state, is inherited from father to son and except the pseudoautosomal regions the DNA sequences do not recombine, therefore, this portion contains a record only of the mutations events that occurred in the past. As a consequence the Y-STRs loci, and specially the haplotype constructed with different loci have been successfully applied to human evolutionary history [7–10] and forensic casework [11–14] investigations. A basic informative core set of nine Y-STRs is, at present, the minimal haplotype recommended for forensic application (see www.yhrd.org).

In order to investigate genetic attributes, establish the genetic distance between the sampled populations and determine if genetic substructure exist in Argentina, a set of 1.197 unrelated males, inhabiting 10 provinces as well as three Argentinean Amerindian tribes (Pilaga, Toba and Guarani) and a set of Bolivian Amerindian (Lecos, Moxeños and Mosetenes) were analyzed. In addition, in order to investigate the European contribution, it was also included for comparison a data set

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from Spain [15]. Our results indicate that the male population of Argentina reflects a homogeneous haplotype distribution similar to that depicted by European populations and only in the Northern region a discrepant component was observed. This might be related to a preponderant Amerindian contribution.

2. Materials and methods

2.1. Individuals

A set of 991 samples from unrelated male individuals were selected based on their geographical origin. This sample set is considered as urban populations and included samples from the following provinces: Buenos Aires $n = 237$; Santa Fe $n = 73$; Rio Negro $n = 224$; Chubut $n = 108$; Mendoza $n = 129$; Misiones $n = 70$; Formosa $n = 54$; Salta $n = 35$; Chaco $n = 33$; Corrientes $n = 28$. In addition, 145 males from three Argentinean Amerindian populations were typed, including 68 members of the Guaraní tribe (Misiones province), 37 Tobas and 40 Pilaga Amerindians (Formosa province) and 61 Amerindians from the Central-East region of the Republic of Bolivia (Lecos, Moxeños and Mosetenes) (Fig. 1). In all cases the samples were collected with the informed consent or derived from judicial requirements, and were treated anonymously. For comparison purposes a data set, previously published [15], including 1407 unrelated males from nine different populations of Spain was computed.

DNA was extracted from blood or buccal swab specimens by conventional organic extraction methods or by using FTA classic cards as described by the manufacturer (Whatman Bioscience, MA, USA).

2.2. Y-STRs typing

The nine loci were amplified in two multiplex PCRs, a triplex reaction including: DYS19, DYS390 and DYS391, and a tetraplex reaction included: DYS385a/b, DYS389I/II, DYS392 and DYS393. Forward primers were labeled with fluorescent dyes and the amplicons were mixed and loaded in a single injection in an automated sequencer (ABI 310/ABI3100-*Avant*, Applied Biosystems, Foster City, USA).

2.3. Statistic analysis

The number of haplotypes, haplotype diversity, R_{st} values and genetic structure, based on molecular variance (AMOVA), were computed with Arlequin 2.0 [16].

The statistical significance of the variance components and R_{st} values were assessed by 10,000 permutations.

A pairwise genetic distance matrix was generated in order to construct a neighbor joining tree using Mega 3.1 [17].

3. Results

Within the urban populations analyzed, 654 different haplotypes were found out of 991 individuals (66%). Two of them denoted relatively high frequency (∼3%), also observed in Latin America, North America and Europe populations included in the YHRD database (Table 1). The most frequent haplotype is the modal for the European haplogroup R1b and the remaining haplotype in this table are one step derived. This supports its European origin [18].

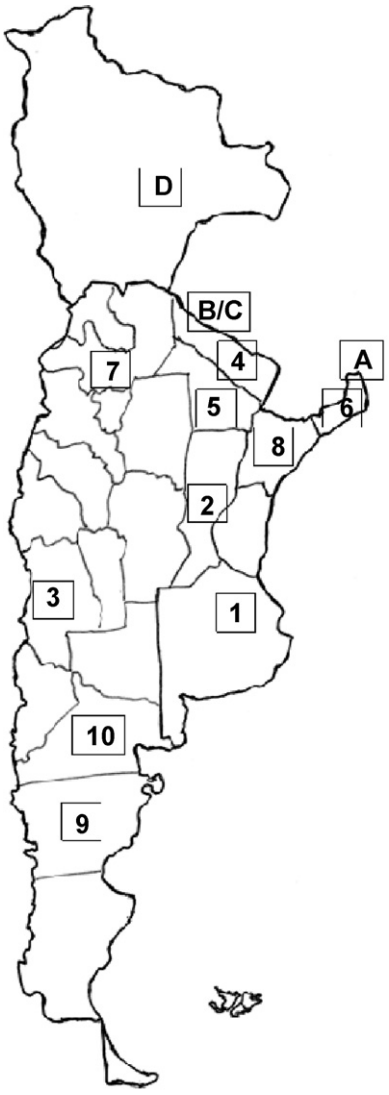

Fig. 1. Map of Argentina and Bolivia indicating the 10 provinces from Argentina: (1) Buenos Aires; (2) Santa Fe; (3) Mendoza; (4) Formosa; (5) Chaco; (6) Misiones; (7) Salta; (8) Corrientes; (9) Chubut; (10) Rio Negro; and the three Amerindian populations: (A) Guaranies (Misiones Province); (B and C) Tobas and Pilagas (Formosa Province); (D) Bolivian Amerindian.

The investigated urban population exhibited a considerable high level of haplotype diversity (0.9967), similar to that depicted by the European populations [19,20]. However, when the population of each of the 10 provinces were considered separately, average haplotype diversity values ranged from 0.994 to 1.000.

In average, haplotype diversities are significantly higher in urban than in Amerindian populations (Table 2), however the Amerindian tribes, included in this study, denoted a wider haplotype diversity range (from 0.86 to 0.99).

Genetic distance determined by R_{st}, showed that most urban populations are well separated from the Amerindian groups.

Table 1
Haplotype frequency comparisons with worldwide database (with 37.884 haplotypes at the moment of comparisons)

Minimal haplotype	Argentina	YHRD worldwide	Latin America	North America	Europe	Asia	Africa
14-13-29-24-11-13-13-11,14	0.031	0.020	0.033	0.032	0.026	0.0004	0.01
14-13-29-24-10-13-13-11,14	0.026	0.010	0.014	0.015	0.013	0.0004	0.005
14-13-30-24-11-13-13-11,14	0.018	0.004	0.01	0.009	0.004	0.0001	0.002
14-14-30-24-11-13-13-11,14	0.017	0.006	0.016	0.013	0.007	0	0.001

Table 2
Sample size, number of haplotypes and haplotype diversity

Region/province	Sample size	Number of haplotypes	Haplotype diversity (±S.E.)
Centre	439	316	0.9963 ± 0.0007
Buenos Aires	237	165	0.9951 ± 0.0011
Santa Fe	73	64	0.9943 ± 0.0042
Mendoza	129	113	0.9972 ± 0.0016
North	220	182	0.9969 ± 0.0011
Formosa	54	48	0.9944 ± 0.0052
Chaco	33	30	0.9943 ± 0.0090
Misiones	70	62	0.9967 ± 0.0030
Salta	35	35	1.0000 ± 0.0068
Corrientes	28	28	1.0000 ± 0.0095
South	332	250	0.9965 ± 0.0009
Chubut	108	92	0.9960 ± 0.0022
Rio Negro	224	179	0.9962 ± 0.0013
Total	991	654	0.9967 ± 0.0005
Amerindian populations	206	120	0.9807 ± 0.0049
Guaranies	68	26	0.8635 ± 0.0372
Bolivian Amerindians	61	57	0.9973 ± 0.0038
Tobas	32	20	0.9556 ± 0.0202
Pilagas	45	26	0.9586 ± 0.0157

Nevertheless, the urban population of Salta province exhibited an intermediate genetic distance between the two defined clusters (Fig. 2). On the other hand, no significant differences were detected between the Spanish data set and the Eastern provinces including Corrientes, Misiones, Formosa and Chaco, in addition to Mendoza provinces (Fig. 2).

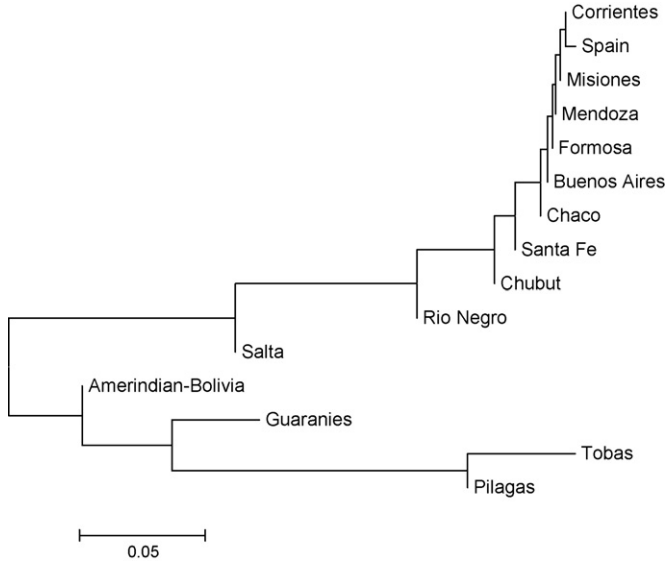

Fig. 2. *Neighbor-joining* tree based on the R_{st} values between pairs populations.

The genetic distance (R_{st}) values between each pair of populations (Table 3) are further reinforced by AMOVA results.

The degree and significance of differentiation between groups were assessed by AMOVA (Table 4).

The results indicate that the Y-STR haplotypes differ significantly between Centre, North, South and Amerindian populations; ~11% of the genetic variance reflects differences among groups, whereas 2% reflects differences among populations within groups, and 87% reflects the genetic variance within populations.

When each group is analyzed separately the Centre and South populations are not significantly different regarding Y-STR haplotypes, whereas both Northern and Amerindian groups exhibit significant differences among populations.

To investigate further the cause of the significant heterogeneity of the population inhabiting the Northern region, Salta province population was removed and the AMOVA repeated. Removal of Salta reduced the heterogeneity among the remaining populations to non-significant levels (R_{st} value among populations was −1.07 and within populations 101.07), whereas removal of any other northern population resulted in R_{st} values that were still statistically significant.

When the analysis of molecular variance is repeated using all the urban populations, except the sample of Salta province, the differences become non-significant among groups and among populations within groups.

Table 3
R_{st} (below the diagonal) and P values (above the diagonal) between pairs populations

Santa Fe		0.2095	0.0917	0.1370	0.2299	0.10138	0.3944	**0.0294**	0.2567	0.3361	**0.0000**	**0.0000**	**0.0000**	**0.0000**	**0.0006**
Formosa	0.00599		0.78032	0.9728	0.8758	0.7382	0.6088	**0.0105**	0.5826	0.0748	**0.0000**	**0.0000**	**0.0000**	**0.0000**	0.2371
Buenos Aires	0.00908	−0.00623		0.6169	0.9366	0.3388	0.4047	**0.0095**	0.4467	**0.0095**	**0.0000**	**0.0000**	**0.0000**	**0.0000**	**0.0006**
Misiones	0.00959	−0.01309	−0.00337		0.8022	0.8069	0.2321	**0.0034**	0.6535	**0.0183**	**0.0000**	**0.0000**	**0.0000**	**0.0000**	0.4373
Chaco	0.00638	−0.01531	−0.01273	−0.01193		0.5588	0.6798	0.0530	0.3988	0.2500	**0.0000**	**0.0000**	**0.0000**	**0.0000**	0.2198
Mendoza	0.01020	−0.00666	0.00014	−0.00639	−0.00553		0.1275	**0.0034**	0.8963	**0.0032**	**0.0000**	**0.0000**	**0.0000**	**0.0000**	0.0586
Chubut	−0.00068	−0.00470	−0.00045	0.00365	−0.00840	0.00668		0.0646	0.1941	0.3874	**0.0000**	**0.0000**	**0.0000**	**0.0000**	**0.0003**
Salta	**0.04001**	**0.05817**	**0.04695**	**0.07263**	0.04010	**0.07296**	0.02466		**0.0073**	0.1985	**0.0000**	**0.0000**	**0.0000**	**0.0211**	**0.0000**
Corrientes	0.00580	−0.00767	−0.00288	−0.00945	−0.00096	−0.01497	0.00955	**0.08503**		0.0879	**0.0000**	**0.0000**	**0.0000**	**0.0000**	0.5689
Rio Negro	0.00045	0.01291	**0.01248**	**0.02165**	0.00449	**0.02463**	−0.00035	0.00740	0.02185		**0.0000**	**0.0000**	**0.0000**	**0.0000**	**0.0000**
Tobas	**0.40477**	**0.45915**	**0.41092**	**0.46443**	**0.45288**	**0.44365**	**0.39037**	**0.24829**	**0.49249**	**0.31473**		0.5015	**0.0000**	**0.0005**	**0.0000**
Pilagas	**0.32049**	**0.35851**	**0.33633**	**0.37304**	**0.33689**	**0.36778**	**0.30648**	**0.15310**	**0.38558**	**0.24705**	−0.00781		**0.0001**	**0.0099**	**0.0000**
Guaranies	**0.29187**	**0.30337**	**0.27011**	**0.32252**	**0.29055**	**0.30740**	**0.25323**	**0.14640**	**0.36122**	**0.21149**	**0.19611**	**0.10904**		**0.0000**	**0.0000**
Amerindians-Bolivia	**0.19527**	**0.21431**	**0.19389**	**0.23460**	**0.19796**	**0.22987**	**0.16628**	**0.04637**	**0.25515**	**0.12587**	**0.12855**	**0.05934**	**0.08773**		**0.0000**
Spain	**0.03952**	0.00261	**0.01325**	−0.00093	0.00515	0.00555	**0.03168**	**0.12654**	−0.00558	**0.05735**	**0.50899**	**0.44546**	**0.36281**	**0.29261**	

Bolded number, $P < 0.05$.

4. Discussion

A considerable amount of information has been published concerning population structure based on Y-STR haplotypes worldwide [5,19,20,22], but doesn't for Argentina population. In this work we presented the first study of population structuring of Argentina based on Y-STR haplotypes.

The populations analyzed denoted remarkable haplotype diversity in all the urban populations as well as the geographic regions to which they belong. The Amerindian population group has more reduced haplotype diversity, and within this group the Guarani population depicts the smallest value (Table 2). This is consistent with a long history of genetic drift, endogamy, isolation and founding effect of the Native American populations [21,23,24].

The determination of the genetic distance, based on R_{st} values, between the different urban populations, denoted a great homogeneity in all the populations tested. Presumably this homogeneity is determined by a relevant European male contribution in all the country. In particular, the absence of significant differences in R_{st} values ($P < 0.05$) between the Spanish data set and the Eastern provinces might reflect the relevant influence of the Jesuit settlement on this region that started in 1639.

The analysis of molecular variance showed significant differences among groups and among populations within groups, when all the urban and Amerindian were considered. In contrast, when each urban population was computed, the difference among groups becomes non-significant; and when each group is analyzed separately in the Center and the Southern regions significant differences were not detected. This is not the case for the Amerindian populations and the Northern region. Within this group, Salta province is the only one that belongs to the Northwest of the country. This region is known to have been strongly influenced by Quechuas and Aymaras population prior to the Spanish conquest and where a strong cultural and ethnical contribution still persists nowadays. When this population is removed from the group, and the analysis of molecular variance repeated, the differences become non-significant for this region.

In contrast to the situation depicted by the Y markers, the mtDNA showed a major Amerindian contribution in Argentina, being the Northern and Southern regions those with the most preponderant Native American component [25]. This situation was also reflected in the study of other Latin American populations, and in the Hispanic-American population of the United States [20,22]. These results are consistent with the history of submission undergone by the native populations during the conquest of the Americas. The admixture process started in 1536, when the first Spanish conquerors settled in the territory that is, at present, occupied by Argentina; in addition to the great migratory waves that entered during the last four centuries.

On the other hand, although significant genetic distances (R_{st}) exist between Salta province and the Native American

Table 4
Analysis of molecular variance results

Region/province	Number of haplotypes	Number of populations	Number of groups	% variance		
				Among groups	Among populations within groups	Within populations
Centre	439	3	1	–	0.54	99.46
North	220	5	1	–	**1.67**	**98.33**
South	332	2	1	–	0.03	99.97
All urban populations	991	10	3	0.23	**0.68**	**99.09**
All urban populations [without Salta]	956	9	3	0.76	−0.01	**99.26**
Amerindian populations	206	4	1	–	**7.78**	**92.22**
All populations	1197	14	4	**10.71**	**2.12**	**87.16**

Bolded number, $P < 0.05$.

populations (Table 3) they are lower than those found for other non-Amerindian populations. These observations might suggest a major Amerindian contribution in Salta province.

The results presented herein underscore the impact of the European male genetic contribution to the aboriginal populations. Previously, our group demonstrated, by DYS-199 typing, that only about 17% of the male lineages of an Argentinean-wide sample set of 325 unrelated males were of Amerindian ancestry [25]. Accordingly, it becomes apparent that a more informative platform might be required for either anthropological or forensic purposes as that offered by the combined use of Y-STRs and Y-SNPs, as may be provided by the YHRD in a near future.

The observation in the North region population of a certain degree of genetic sub-structure, and the high degree of differentiation of the Native American populations, underscore the necessity to increase the studies in this region and the Argentinean Amerindian populations that inhabit them.

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