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Y-chromosome lineages in native South American population

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ABSTRACT

The present work tries to investigate the population structure and variation of the Amerindian indigenous populations living in Argentina. A total of 134 individuals from three ethnic groups (Kolla, Mapuche and Diaguitas) living in four different regions were collected and analysed for 26 Y-SNPs and 11 Y-STRs.

Intra-population variability was analysed, looking for population substructure and neighbour populations were considered for genetic comparative analysis, in order to estimate the contribution of the Amerindian and the European pool, to the current population.

We observe a high frequency of R1b1 and Q1a3a* Y-chromosome haplogroups, in the ethnic groups Mapuche, Diaguita and Kolla, characteristic of European and Native American populations, respectively. When we compare our native Argentinean population with other from the South America we also observe that frequency values for Amerindian lineages are relatively lower in our population.

These results show a clear Amerindian genetic component but we observe a predominant European influence too, suggesting that typically European male lineages have given rise to the displacement of genuinely Amerindian male lineages in our South American population.

1. Introduction

Population genetics historically concerns about the population subdivision and population structure, considering the allele frequency distribution and change under the influence of the four evolutionary forces: natural selection, genetic drift, mutation and gene flow. South America, and especially Argentina, is one of the human world locations where the genetic information still continue not clear at all. Argentina population harbours many different Amerindian ethnic groups, and many of them with a very low number of individuals (≤1000). The largest variety of population groups is found in the North, where the most representative ones are the Kollas, Diaguitas, Huarpe, Guaraní, Ava guarani, Wichi, Toba, Mocovi and Guarani-mbya. While in the centre and south of the country we can find other groups as the Tehuelche, Rankulche, Comechingon, Tonocote, Ona and Mapuche population (INDEC, Instituto Nacional De Estadística y Censos from Argentina, http://www.indec.gov.ar).

During the late 19th and the early 20th centuries, Argentina received lots of immigrants from different European countries, as Spain and Italy. However, immigrants settled mainly in the city areas, where is concentrated most of the inhabitants, giving rise to a diverse urban population with a highly diluted Amerindian component.

Indigenous population represent only about 1% of the total Argentina population (INDEC 2006), with around 403,000 individuals nowadays distributed all around the country, which shows a western area, with a rugged landscape corresponding to the andinian edge, and a landscape softened towards the eastern area, with high plateaus and plateaus. Among the indigenous groups, Kolla, Diaguita and Mapuche populations are current representatives following a North to South distribution (Fig. 1).

The Kolla group is represented by nearly 71,000 individuals settled in the provinces of Jujuy, Salta and Catamarca. Their languages are Aymara and Quechua.

Mapuche group is the most extensively represented, with almost 114,000 individuals, established along the Neuquen, Rio Negro and Chubut region. Their original language is Mapudungun.

A more reduced indigenous group, are the Diaguitas settled in the provinces of Catamarca and Tucuman, which accounts with close to 32,000 individuals. They have a original language too, known with different names "kaka", "chaka", "yacampis", and "calchaqui".

Although a large amount of data on autosomal markers has became available from different regions of South America [1–8], until now, few Y-chromosome populational studies have focused on this world region, and especially on Argentina [9–11]. Some of these studies include Y-STRs characterization, but only a few

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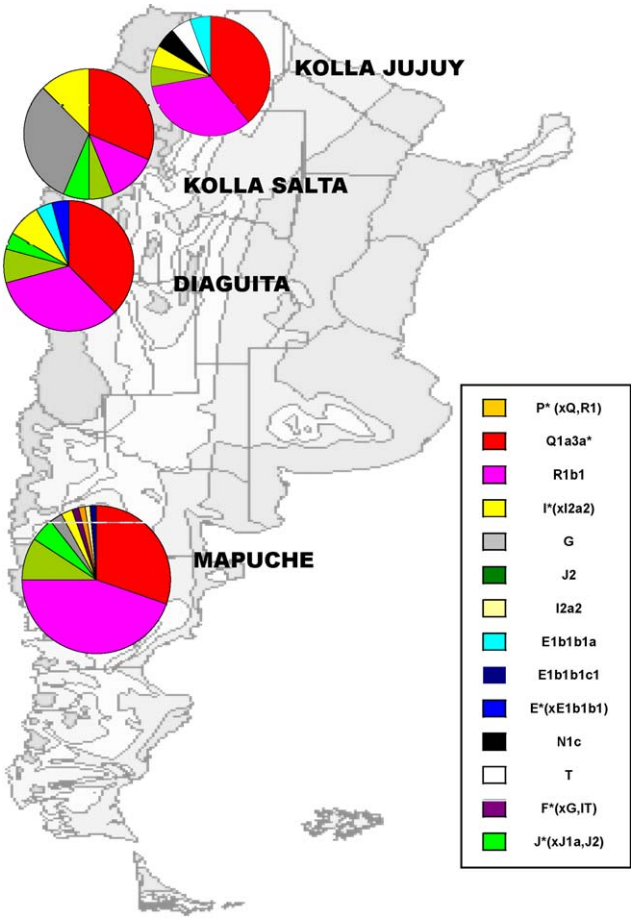

Fig. 1. Map of Argentina showing haplogroup frequencies in each population and their geographic location.

examples have made use of Y-chromosome SNPs [12–16]. In this work we have joined Y-STRs and Y-SNPs because they are a very useful tool in forensic DNA testing and human evolutionary studies, particularly as concerns male demographic history and migration patterns, furthermore Y-SNPs are especially interesting to find the American Y-chromosome founder haplogroups and to allow prediction of the geographic or ethnic origin of unknown casework samples [17]. There are two very important Y-SNPs which identify Amerindian populations: one of them is the M242 (C > T) [18], this mutation is located close in time to the entry of the first modern human into the American continent, the other one is M3 (C > T) [19], this polymorphism was described to be occurred in Beringia when the first modern human entered into the American continent [12,20,21]. However, the close phylogenetic position of both mutation in the Y-chromosome haplogroup tree open to debate the possible origin of M3 mutation before the first human settlement in America [22–26].

In order to gain insights into the genetic diversity present in the male lineages of the Argentinian indigenous populations, in this paper we have analysed the Y-chromosome lineages of the Mapuche, Kolla and Diaguita indigenous groups. Migration, admixture and antiquity patterns were evaluated, by the analysis of 26 Y-chromosome single nucleotide polymorphisms (Y-SNPs) and 11 Y-chromosome short tandem repeats (Y-STRs). We have looked for the genetic differences among those three indigenous populations and moreover, other neighbour populations were considered to evaluate Amerindian and European genetic contribution.

2. Materials and methods

2.1. Populations

We obtained 134 unrelated male blood samples on FTA® Whatman paper, under informed consent. The individual ethnic origin was considered in function of ethnic group with which the individual felt identified. DNA was extracted using a standard phenol chloroform method. Extracted products were quantified on a NanoDrop® ND-1000 Spectrophotometer. Among the 134 samples collected, 34 belong to the Kolla ethnic group, 24 to the Diaguitas ethnic group, and 76 samples were Mapuches. Samples from Mapuche indigenous group were collected from Chubut (latitude: 42°54′46.61″S, longitude: 71°18′43.22″O), a province in the South of the country. Kolla individuals were collected from two well-differentiated locations, 18 samples from Jujuy (latitude: 24°10′57.04″S, longitude: 65°17′41.17″O) and 16 samples from Salta province (latitude: 24°43′25.9″S, longitude: 65°52′28.36″O), since now called Kolla Jujuy and Kolla Salta, respectively, both in Northwest Argentina. Finally, Diaguita samples were collected without differentiation from Catamarca and La Rioja provinces in the Northwest Argentina (latitude: 28°20′02.49″S, longitude: 67°43′32.40″O).

2.2. Y-chromosome typing

In order to carry out our analyses, we used multiplex reactions, both for the Y-SNPs and for the Y-STRs.

The Y-STRs were amplified by means of the PowerPlex Y® System (Promega) multiplex kit, containing 11 STRs (DYS391, DYS389I, DYS439, DYS389II, DYS438, DYS437, DYS19, DYS392, DYS393, DYS390, DYS385). Some samples were further analysed with AmpFlSTR® Yfiler™ multiplex kit (Applied Biosystems). In both cases manufacturer recommendations were followed.

Y-chromosome SNPs were analysed by using the SNaPshot™ Multiplex kit (Applied Biosystems). The 26 Y-SNPs were amplified in the form of three multiplex reactions previously described by Brión et al. [27], and a novel multiplex reaction containing the SNPs M242, M3, M19, M194 and M199. Design of the amplification primers of the new multiplex was carried out using Primer3 Software. The possible occurrence of unspecific primer dimer was subsequently explored by means of the AutoDimer Version 1.0

Table 1
Y-chromosome markers and primer sequences for the multiplex PCR amplification.

SNP	Mutation	Primer (5′–3′)		Amplicon size (bp)	Conc. (μM)
		Forward	Reverse		
M242	C > T	atagaaagtttgtgcaaaaaggtga	accttacctagaacaactctgaagc	137	0.4
M3	C > T	ctgccagggctttcaaatag	aagggcatctttcattttaggt	93	0.4
M19	T > A	ctggtcataacactggaaatc	agctgaccacaaactgatgtaga	170	0.4
M194	T > C	gcctggatgaggaagtgag	atacagtcgttgccttctcg	127	0.4
M199	>Ins G	cctggttggattctggtctt	tgatttcaaggatttgttagtctt	197	0.4