

# Analysis of Y chromosome lineages in native South American population

A. Blanco-Verea, M. Brion <sup>\*</sup>, P. Sanchez-Diz, J.C. Jaime,  
M.V. Lareu, A. Carracedo

*Institute of Legal Medicine, Faculty of Medicine, University of Santiago de Compostela, Spain*

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**Abstract.** The present work tries to identify both the evolutionary footprints and the origin of native population of Argentina using 26 SNPs and 11 STRs of the Y chromosome. A total of 34 samples from 3 different native population (Kolla, Mapuche, Diaguitas) from 5 different regions were analysed. © 2006 Elsevier B.V. All rights reserved.

*Keywords:* SNP; STR; Genetic population; Y chromosome

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## 1. Introduction

Argentina harbours many different Amerindian ethnic groups. Here we have studied, at the Y chromosome level, three populations (Kolla, Mapuche and Diaguita) from 5 different regions throughout Argentina.

In order to gain information on the genetic diversity and similarity present in the male lineages of these populations, we typed 26 Y-SNPs and 11 Y-STRs in 134 samples.

## 2. Materials and methods

To carry out our analyses, we used multiplex reactions for both the Y-SNPs and Y-STRs. The Y-STRs were amplified by means of the PowerPlex Y<sup>®</sup> System (Promega), containing 11 STRs. The 26 Y-SNPs were amplified in the form of three multiplex reactions previously described [1], and one novel multiplex reaction with the SNPs M242, M3, M19, M194, M199 [2,3] (Fig. 1). A QUIAGEN<sup>®</sup> Multiplex PCR kit was used in order to optimize the functioning of this newly created multiplex. Multiplex single base

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<sup>\*</sup> Corresponding author. Institute of Legal Medicine, San Francisco s/n, 15782 Santiago de Compostela, Spain. Tel.: +34981582327; fax: +34981580336.

*E-mail address:* brioniml@usc.es (M. Brion).

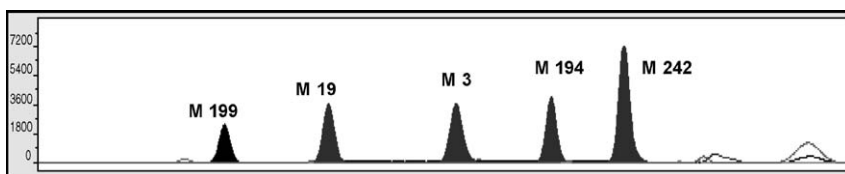


Fig. 1. Electropherogram of the new multiplex for haplogroup Q.

extension reactions were carried out following the Y-SNP amplifications by means of the SNaPshot™ Multiplex kit (Applied Biosystems). The products of both the Y-STR amplifications and the Y-SNP single base extensions were run on a ABI PRISM® 3100 Genetic Analyzer (Applied Biosystems) and analyzed using GeneScan® Analysis Software version 3.7 (Applied Biosystems). Statistical calculations were carried out using the Arlequin® Software [4].

### 3. Results and discussion

Our analysis reveal the presence of 14 different Y chromosome haplogroups (Fig. 2A). The two main haplogroups found in each of the 5 regions are R1b and Q3. In Kolla Salta

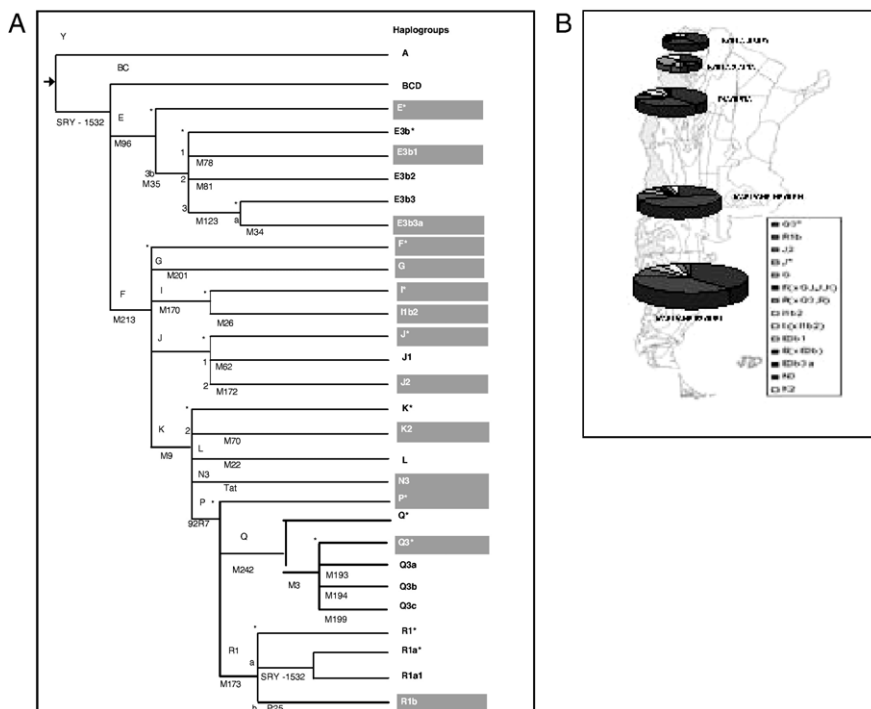


Fig. 2. A) Maximum parsimony phylogeny of the Y-SNPs analysed. Haplogroups found in the populations studied are represented with different colours. B) Haplogroup frequencies of the 5 regions analysed. Area of the circles is proportional sample size.

Table 1  
Haplogroup and haplotype diversity values

	Mapuche Esquel	Kolla Jujuy	Kolla Salta	Diaguita	Mapuche Nequen
Sample size	42	18	16	24	34
SNPs Haplogroup diversity	0.7077 ± 0.0684	0.7647 ± 0.0735	0.8167 ± 0.0628	0.7609 ± 0.0596	0.7077 ± 0.0685
STRs Haplotype diversity	0.9977 ± 0.0058	1.0000 ± 0.0185	0.9917 ± 0.0254	0.9964 ± 0.0133	0.9929 ± 0.0090

Table 2  
Variation among and within populations

	% of variation in SNPs		% of variation in STRs	
	5 regions	3 populations	5 regions	3 populations
Among population	2	−0.68	5.40	1.40
Within population	98	98.17	94.6	94.27
Within population among groups	–	2.51	–	4.33

population is different because haplogroup G frequency is higher than R1b (Fig. 2B). On the other hand, looking at the Y-STRs results a total of 112 different haplotypes were found. We can see haplogroup and haplotype diversity values in Table 1.

The percentage of variation observed at the Y-SNPs level for the five different regions shows that most of the variation is found within populations and not among populations. Furthermore the *P* value ( $P=0.09971$ ) associated to the latter variation is non-significant, suggesting this value is not real. However at the Y-STRs level we found a small percentage of variation among populations which according to its associated *P* value ( $P=0.00098$ ), seems to be real. When the five geographical regions are grouped into three populations, similar results are observed (Table 2). Only two STRs haplotypes were shared among populations, one between Mapuches and Kollas and the other one between Mapuches and Diaguitas. All of them belonging to hapogroup R1b. By contrast, the number of shared haplotypes among regions within populations rise to 12 in the case of Mapuches.

All of the populations show a significant influence of European lineages. We think that the high frequency of haplogroup R1b (typically European) implies a rapid dissemination of this haplogroup in the Amerindian population after the European colonization of South America.

## References

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