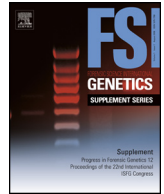




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# Analysis of the genetic structure of Santa Cruz province and its comparison with the other Southern Patagonian provinces of Argentina

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### ABSTRACT

Santa Cruz is the southernmost Patagonian province of Argentina. Its demographic history is complex and changing over time. Aiming to investigate the overall genetic features of this province a set of markers routinely used in forensic casework were analyzed. The study involved 645 from Santa Cruz ( $N=145$ ), Chubut ( $N=102$ ), Rio Negro ( $N=99$ ), Buenos Aires ( $N=93$ ) and potentially parental population samples from CEPH panel ( $N=206$ ). We analyzed 15 autosomal STRs, 23 Y-STRs and 6 Y-SNPs. Autosomal STRs displayed statistically significant genetic distances between Santa Cruz and Chubut, Rio Negro and Buenos Aires populations. Native American Y-specific haplogroup Q1a3a was present in around 15% of the Santa Cruz males and the haplogroup R1b was the most frequent European hg (45%). Structure analysis demonstrated that three ancestral contributors that build to Santa Cruz gene pool were European (50%), Native American (36%) and African (13%). Genetic characterization of this population allowed to further increase genetic knowledge of the country, where previous studies have identified regional differences associated with the complex demographic process that took place in South America.

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## 1. Introduction

Santa Cruz province is the southernmost Patagonian province (latitude ranging 46°00'S–52°23'S and longitude 65°43'W–73°35'W). The origin of the current population of Santa Cruz (SC) is based on successive internal and external migration events, constituting an interesting model from molecular demography point of view. Being the second larger province of Argentina, after Buenos Aires, with 243,945 km<sup>2</sup>, is one of the less populated (after Tierra del Fuego, Antártida and South Atlantic Islands) with a population density of 1.1 hab/km<sup>2</sup>. The actual population size is 263,974 inhabitants, mostly concentrated in Rio Gallegos, capital of the province.

The populations of Santa Cruz had experienced fluctuations across a short period. The oldest settlers were the Aónikenk, arriving around 12,000 years before present. The Mapuche penetration started from western Andes during the seventeenth and eighteenth centuries. Europeans arrived during nineteenth and early twentieth centuries. In the recent past, the region

experienced a constant demographic fluctuation due to internal migrations (mostly from Buenos Aires province) and external migrations from neighboring countries (Chile and in a lesser degree Bolivia) accompanying economic fluctuations related to oil exploitation.

Aboriginal communities living in Santa Cruz, as well as in the rest of Argentinean Patagonia, come mainly from two ethnic groups: Mapuche and Tehuelche [1].

The overall objective of this study was to characterize the genetic composition of this population by means of Mendelian and uniparentally transmitted markers. In addition, a comparison with other Argentinean provinces, geographically or demographically related was carried out.

## 2. Materials and methods

### 2.1. Sample donors

We analyzed 145 samples belonging to the most populated areas of Santa Cruz (SC) province. Population comparisons included Buenos Aires (BA,  $N=93$ ), Chubut (CHU,  $N=102$ ) and Rio Negro (RN,  $N=99$ ) (Fig. 1).

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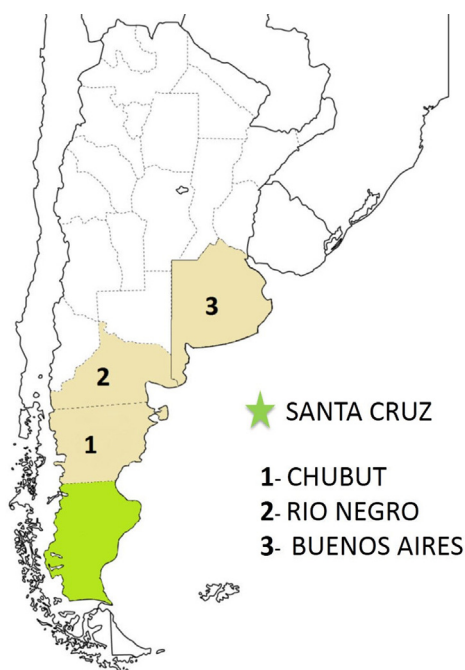


Fig. 1. Map of Argentina shown geographical localization of sampling places.

## 2.2. Genetic polymorphisms analyzed

15 autosomal STRs, included in Identifiler Plus kit (Applied Biosystems), 23 Y-STRs (Power Y-23, Promega) and six Y-SNPs, specific for Q1a3a, R1b, E1b, J, I2 and G2a Y-haplogroups (Hgs).

## 2.3. Statistical analysis

Population genetic parameters were determined by using the Software GenAlEx v6.501. Hardy Weinberg Equilibrium and interpopulation analysis were determined with Arlequin v3.5.1.2. Analysis of the population composition was carried out with Structure v2.3.4; parental populations (European, Native American and African) were selected from HGDP-CEPH panel ( $N=206$ ). Haplogroup Predictor Software (<http://www.hprg.com>) and YHRD-global tool database ([www.yhrd.org](http://www.yhrd.org), release 49) were used for determine Y Chromosome Hgs, confirmed with specific Y-SNPs typing by Real Time PCR followed by HRM.

## 3. Results and discussion

Allele frequencies were determined by counting method. None except one autosomal STR (D16S539) departed from HWE however, after Bonferroni correction, all system met equilibrium. The combined match probability was  $1.358 \times 10^{-17}$  and the combined power of exclusion was higher than 99,999999%. FGA, D18S51 and D21S11 systems shown the highest PD and PE (ranging from 0.9708 to 0.9545; and 0.7188 to 0.7031, respectively) and the lowest values were observed in TPOX (PD = 0.8296 and PE = 0.3164), in accordance with other Patagonian provinces [2].

Males were represented in 44/145 samples. All the haplotypes were unique, and only 2 were found in the worldwide Y-haplotype database (YHRD, release 49). The haplogroup distribution was R1b (45%), E1b1b (20%), E1b1a (2%), J1 (5%), J2a (5%), I1 (2%) and I2b (2%). Native American Y specific hg Q1a3a was observed in 15% of the male population, higher than general Argentinean population (5%) [3].

Population comparison between Santa Cruz and Buenos Aires population (selected due its demographic relationship) and two Patagonian provinces, Chubut and Río Negro (geographically

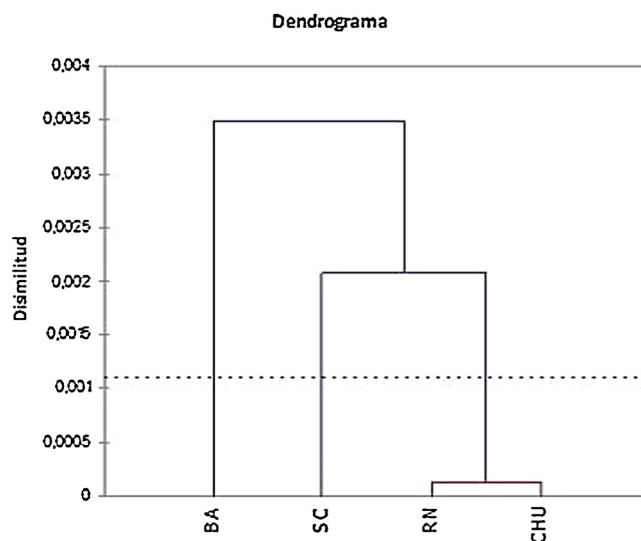


Fig. 2. Dendrogram representing the genetic distances between populations of Buenos Aires (BA); Chubut (CHU); Rio Negro (RN) and Santa Cruz (SC).

related to SC). Dendrogram from  $F_{st}$  genetic distances considering the 13 STRs Codis Core is shown in Fig. 2. All genetic distances, except RN-CHU, were significant ( $p < 0.05$ ). As can be observed, SC is located in between Buenos Aires and the cluster conformed by Chubut and Rio Negro.

Amova analysis considering three groups (BA, CHU+RN, and SC) shown no significant values for *Among groups* and *Among populations within groups* fixation indexes (FCT 0.00264,  $p=0.176+/-0.0098$ ; FSC 0.00011,  $p=0.411+/-0.0144$ , respectively).

A structure analysis was performed employing parental population's samples from HGDP-CEPH panel. Using biparentally transmitted markers, Santa Cruz population show a European ancestral component of 50.7%, Native-American component 6.3% and African component 13%, similar to those results obtained for populations included in the comparison (data not shown).

The results obtained could be correlated with the demographic history of Santa Cruz, reflecting in their genetic composition migratory movements of the recent past. This study let us to infer the ethnic composition of the studied population and generate a database for forensic purpose. Further analysis of mtDNA D-Loop sequence and Ancestral Informative Markers will complete the characterization of one of the most austral population of the planet.

## Conflict of interest

The authors report no declarations of interest.

## Acknowledgments

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