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# Ancestry and admixture of a southernmost Chilean population: The reflection of a migratory history

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**Abstract**

**Objectives:** Punta Arenas is a Chilean city situated on ancestral Aönikenk territory. The city was founded by 19th- and 20th-century colonists from Chile (Chiloé) and Europe (Croatia). This work uses uniparental and ancestry-informative markers (AIMs) to explore the effects of historic migratory and admixture patterns on the current genetic composition of Punta Arenas.

**Methods:** We analyzed mitochondrial DNA (mtDNA), Y-chromosome single-nucleotide polymorphisms (SNPs), and 141 AIMs obtained from 129 DNA samples from male residents with regional ancestry. After characterizing uniparental lineages and ancestry proportions, multivariate analysis was used to explore relationships among the various types of data.

**Results:** Punta Arenas has an admixed population with three main genetic components: European (56.5%), northern Native (11.3%), and south-central Native (28.6%). The Native component is preponderant in the mtDNA (83.76%), while the foreign component predominates in the Y-chromosome (92.25%). Non-Native mtDNA lineages are associated with European genetic ancestry, and Native mtDNA lineages originated mainly in the southern and southernmost regions of Chile. Most non-Native Y-chromosome SNPs originated in Spain, and secondly, in Croatia.

**Conclusions:** The population of Punta Arenas is mainly of Chilote origin with south-central Native and Spanish ancestral components, as well as some Croatian components. The persistence of local Native lineages is notable, suggesting continuity with the ancestral populations of the region such as the Kawésqar, Aönikenk, Yámana, or Selknam peoples. This study contributes to our knowledge of local history and its links to national and global developments in genetic ancestry.

## 1 | INTRODUCTION

Punta Arenas, Chile, is located on the north bank of the Strait of Magellan at 53° 09′ South latitude. The historic migratory and admixture patterns of this city differ from those of central Chilean cities as it was founded relatively late in the country's history. Punta Areas was established in 1848 by the Chilean government to defend the territory adjacent to the strait (Martinić Beros, 1977, 2002; Ortega Perrier, 1980; Zamora, 1975), a hub for diverse colonists from various regions.

The city was populated by multiple waves of immigrants from the second half of the 19th century until the second half of the 20th century, with both Chilean and foreign immigrants drawn to the region by government incentives (Cussen, 2016). Prior to the city's foundation,

  

the area was inhabited by Tehuelche or Aönikenk communities and sporadically visited by Kawésqar and Yámana individuals. These Native groups were displaced and/or reduced during colonization (Gobierno Regional de Magallanes y de la Antártica Chilena, 2013; Harambour, 2018).

The first wave of immigrants was from central and southern Chile, especially the Chiloé Archipelago. Settlers from throughout the Greater Island of Chiloé were descendants of the admixture between Amerindians and the original Spanish colonists in the region (Ortega Perrier, 1980; Vázquez de Acuña, 1991). Since efforts at national immigration were not sufficiently fruitful, the government began offering incentives to overseas immigrants in 1873. Most of European immigrants were Croatians from the Island of Brač in the Dalmatian area, who arrived in the 1890s (Hernández et al., 1993; Martinić Beros, 1999a; Martinić Beros, 1999c; Martinić Beros, 2002; Ortega Perrier, 1980; Zamora, 1975). The local demography thereafter was characterized by periods of immigration and emigration, depending on economic activity. The last wave of migrants arrived from Chiloé between 1955 and 1970.

Demographic growth has been largely stagnant since that time (Vera Giusti, 2008). Of the total inhabitants recorded in the 2002 census, 43% were born in another region and 1.5% in another country (INE, CEPAL, & CELADE, 2014), and the corresponding figures from the 2017 census were 42 and 3% (INE, 2017). This considerable immigrant population makes it clear that both migration and admixture of the populations inhabiting the region are ongoing phenomena. Therefore, studying the characteristics and interactions of these human groups is likely to yield useful insights.

Admixture is a process in which two or more genetically and/or phenotypically-distinct populations of different continental ancestry mate to form new populations (Adhikari et al., 2016; Chacón-Duque et al., 2018; Ding et al., 2011; Enoch et al., 2006; Koehl & Long, 2018; Santos et al., 2010). Human populations are not panmictic. Instead, our species tends to form relatively isolated or endogamic reproductive communities that present genetic differences between them, that internally share a genealogical or ancestral history (Peter, 2016; Vanegas et al., 2008). Population structure occurs in populations with recent admixture among subpopulations, reflected as systematic differences in allele frequency associated with the different ancestries of origin (Hajiloo et al., 2013; Santos et al., 2010). This phenomenon is the result of historic divisions between populations, including social, migratory, mating, and demographic differences (Hajiloo et al., 2013; Relethford, 2019; Sevini et al., 2013).

Admixture and population structure can be studied by using ancestry-informative markers (AIMs) to explore the differential contributions of various ancestries to admixed populations. AIMs are genetic markers that present contrasting allelic frequencies between populations of different ancestry (Esposito et al., 2018; Huerta-Chagoya et al., 2019; Pfaff et al., 2001; Pritchard & Donnelly, 2001; Shriver et al., 1997). Moreover, lineage-based approaches, involving phylogeographic analysis of non-recombinant DNA through uniparental markers such as mitochondrial DNA (mtDNA) and the non-recombinant region of the Y-chromosome (NRY, Y-chromosome), allow for detailed examination of migratory processes (Oppenheimer, 2012). The use of both uniparental markers also allows for estimation of sex-biased contributions to the admixture, which are largely attributable to differential migration rates between males and females (Alfonso-Sánchez et al., 2019; Heyer et al., 2012; Jobling & Tyler-Smith, 2003; Kivisild, 2015; Vieira-Machado et al., 2016).

Our understandings of these molecular-based findings can be improved by examining culturally transmitted ancestry markers such as surnames. By examining the links between documented genealogical history and genetics, we can identify correlations between self-reported ethnic affiliation and genetic identity (Colantonio et al., 2003; Corach et al., 2010; Sevini et al., 2013; Shraga et al., 2017).

Despite abundant historical knowledge regarding the migratory dynamics of Punta Arenas, its historic admixture patterns have yet to be studied from a genetic perspective. It would be useful to understand how Native and immigrant populations have interacted over the years, elucidating the patterns of their admixture as well as the influence of this history on the current genetic variability of this city.

This work aims to analyze the effect of historic admixture and migration patterns in Punta Arenas on the genetic composition of its current population using uniparental markers —Y-chromosome single-nucleotide polymorphisms (SNPs) and mtDNA— and ancestry-informative markers (AIMs), in order to identify the geographic origins of the populations that gave rise to the city and better understand the admixture among these groups.

## 2 | MATERIALS AND METHODS

### 2.1 | Sample

This study was authorized by the Human Research Ethics Committee, Faculty of Medicine, Universidad de Chile

(Approval Number 166-2014, December 23, 2014). Study participants signed an informed consent form approved by the committee.

The study sample included 129 male adults born in Punta Arenas or the Magallanes region with at least one parent also born in the region. Participants provided saliva samples used to obtain DNA with an Oragene-DNA Self-Collection Kit OG-500 from DNA Genotek Inc. Participants also answered a genealogical survey.

Samples were collected using a stratified design, with strata size proportional to the population of the neighborhood unit per the 2002 Census (Figure 1; Ilustre Municipalidad de Punta Arenas, n.d.; INE Magallanes, n.d.; INE, 2003).

## 2.2 | Laboratory analysis

### 2.2.1 | Mitochondrial DNA

We sequenced the control region of human mtDNA from position 16 024 to 576 according to Parson and Bandelt (2007). Amplification conditions are described in Table S1 (de la Fuente et al., 2015; Handt et al., 1996; Moraga et al., 2000). Macrogen Inc. (South Korea) purified the PCR products and performed the sequencing. Sequences were aligned to the Revised Cambridge Reference Sequence (rCRS; Andrews et al., 1999) following standard control parameters (Bandelt & Parson, 2008). Polymorphism confirmation and sequence editing were carried out on Geneious Prime v.2019.2.3 (http://www.geneious.com, Kearse et al., 2012), and haplogroups were assigned using Haplogrep2 v.2.2 (https://haplogrep.i-med.ac.at/, Weissensteiner et al., 2016), based on PhyloTree v.17 (http://www.phylotree.org, van Oven & Kayser, 2009).

### 2.2.2 | Y-chromosome

We typified a set of 19 biallelic markers (SNPs) to assign Y-chromosome haplogroups. We followed a hierarchical approach according to (Karafet et al., 2008). The primer for R-M207 was designed using the online software Primer-BLAST (http://www.ncbi.nlm.nih.gov/tools/primer-blast, Ye et al., 2012), and the other 18 makers were taken from the literature (Table S2; Bailliet et al., 2012; de Saint Pierre, 2013; Hammer & Horai, 1995; Niederstätter et al., 2012; Su et al., 1999; P. A. Underhill et al., 1997). The allele state of each SNP was determined by amplifying the target sequence followed by digestion with restriction enzymes, using the PCR-RFLP technique. The annealing temperatures and restriction enzymes are available in Table S2. Due to the mutation defining haplogroup DE (Alu insertion), only a PCR was carried out. The resulting fragments were analyzed by agarose gel electrophoresis (2%).

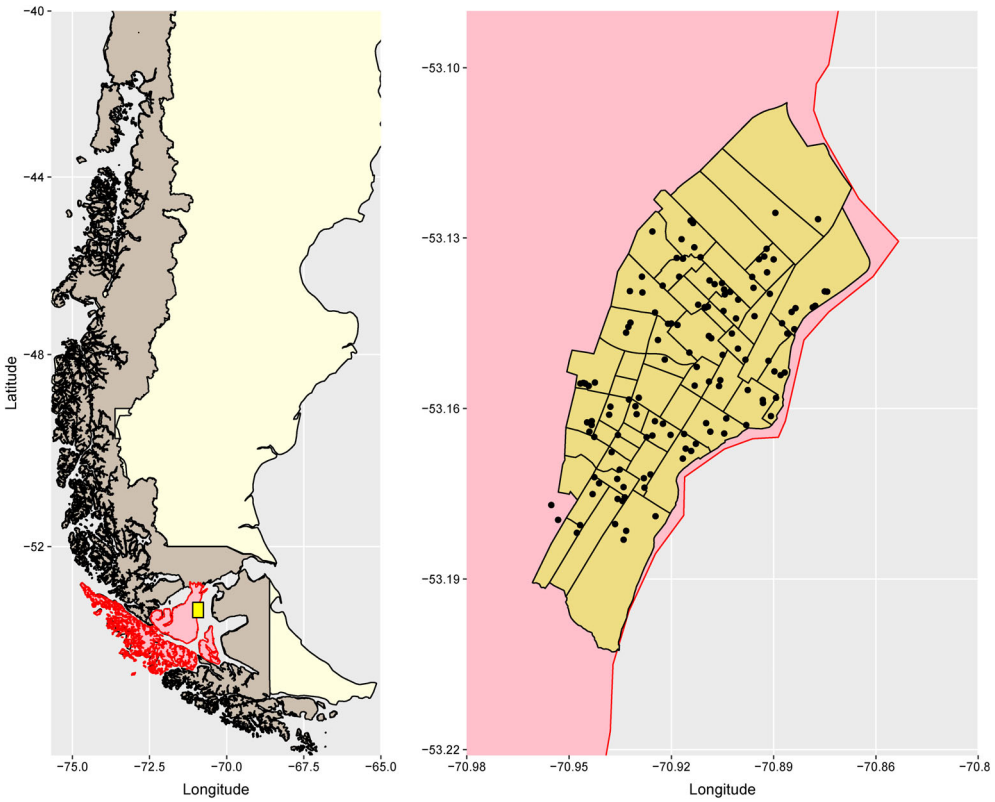


**FIGURE 1** Left: Map of the Southern Cone of South America, including Chilean and Argentinian Patagonia. Magallanes Province indicated in red and Punta Arenas City as yellow rectangle. Right: Map of Punta Arenas, including the 53 neighborhood units and saliva sample collection location

### 2.2.3 | Nuclear DNA

DNA samples were genotyped using a 150-AIMs panel in LGC Genomics with KASP™ (Kompetitive Allele Specific PCR). This panel allows to work under a continental differentiation model and to differentiate among European, African, and Native American (Amerindian) ancestry, as well as distinguishing within ancestral Native American populations, including those from the Chilean, Peruvian, and Bolivian highlands versus those from southern Chilean regions, hereafter referred to as northern Native or south-central Native (Verdugo et al., 2020).

### 2.2.4 | Statistical and bioinformatic analysis

The relative frequencies of the uniparental markers were estimated in the sample. DnaSP v.6.12.03 was used to estimate standard genetic diversity indices: number of segregating sites (S), nucleotide diversity ($\pi$), haplotype diversity (Hd), and mean number of pairwise differences (K). Arlequin 3.5.1.2 (http://cmpg.unibe.ch/software/arlequin35/, Excoffier & Lischer, 2010) was used to estimate fixation (Fst) index (Hartl & Clark, 1997; Wright, 1978), to account for differentiation between populations. Given the known history of the region, the mtDNA data were compared with reference frequencies for various Native populations: Aymara, Atacameño, Pehuenche, Huilliche, Argentinian Mapuche, Tehuelche, Kawésqar, and Yámana (de Saint Pierre et al., 2012), as well as for populations from other rural and urban parts of Chile: Azapa, Camarones (Apata et al., 2017), and Santiago (Gómez-Carballa et al., 2016). The Y-chromosome data was compared with that of reference samples from five urban Chilean populations (Toscanini et al., 2016). Neighbor-joining dendrograms were constructed from the Fst matrices using MEGA X (Kumar et al., 2018). Networks were constructed for the B, C, and D haplogroups using Network v.10.0.0.0 and Network Publisher v2.1.2.5 (http://fluxus-engineering.com) following the median-joining algorithm (Bandelt et al., 1999), and post-processing was performed using maximum parsimony (Polzin & Daneshmand, 2003). Positions 152, 309.1C, 315.1C 523d-524d, 16 182, 16 183, 16193.1C, and 16 519 were eliminated, and positions 146, 195, 16 189, and 16 311 were assigned a low mutation weight (de Saint Pierre et al., 2012).

PLINK v.1.90b6.9 (Purcell et al., 2007) was used for sample's quality control. SNPs with a low genotyping rate or in Hardy–Weinberg disequilibrium were discarded, resulting in a database with 129 individuals and 147 SNPs. Reference panels included African (YRI) and European (CEU; https://www.internationalgenome.org, 1000 Genomes Project Consortium, 2015) as well as northern (Aymará; Ioannidis et al., 2020) and south-central Amerindian ancestries (Pehuenche and Huilliche; de la Fuente et al., 2018). After merging and quality control, 141 SNPs remained. ADMIXTURE v.1.3.0 (Alexander et al., 2009) was used to infer individual ancestry proportions, running from $K = 2$ to $K = 8$ with default parameters, estimating the cross-validation error for each $K$ value, and choosing the $K$ value with the lowest cross-validation error.

Genealogical survey results, haplogroup typification data, and individual ancestry proportions were entered into a database and analyzed using R statistical software (R Core Team, 2020). Frequencies and associations were evaluated using contingency tables and Fisher's exact test. A principal component analysis (PCA) was performed on the SNPs using the EIGENSOFT v.7.2.1 Perl package smartpca function (Patterson et al., 2006). Hierarchical clustering for principal components was performed on the ancestry results to identify natural clusters in Punta Arenas, compared to reference populations. Since the data contained both categorical and continuous variables, a factorial analysis of mixed data (FAMD) was carried out to evaluate associations between Y-chromosome and mtDNA haplogroup origin and ancestry proportions. Both multivariate analyses were performed using the FactoMineR R package (Lê et al., 2008).

## 3 | RESULTS

### 3.1 | Self-declared ancestry

We obtained the self-declared geographic origins of each participant's maternal and paternal families from the genealogic survey. While self-declared maternal and paternal origin were independent (chi-square = 22.5; $p = .3140$), paternal last name and paternal ancestral geographic origin did show a significant association (chi-square = 160.79; $p = .0000$).

Self-declared paternal geographic ancestry was mainly Spanish, Latin American, or Chilean (including Chilote; 51.16%, $n = 66$). Most surnames originated in Spain (76.74%, $n = 99$). A total of 14 individuals had both Croatian paternal surname and self-declared Croatian paternal origin (10.85% for both variables), while paternal surnames and self-declared ancestry corresponded to other European nations in 19 (14.72%) and 15 (11.63%) individuals, respectively.

Self-declared maternal ancestry was also primarily Spanish (62.79%, $n = 81$). Another 5.43% ($n = 7$) and 6.2% ($n = 8$) of the sample was of self-declared Croatian

or other European origin, respectively. Native American origin represented only a small proportion of surnames and self-declared geographic origins.

## 3.2 | Mitochondrial DNA

Four samples could not be sequenced. Of the sample analyzed, mitochondrial lineage frequencies (Table S3, Figure S1) showed a predominance of Native American macrohaplogroup C (36.43%, $n = 47$), followed closely by macrohaplogroup D (30.23%, $n = 39$) and then macrohaplogroup B (15.5%, $n = 20$), while macrohaplogroup A accounted for only 1.6% ($n = 2$) of this sample from Punta Arenas. When Native American ancestry was explored at higher resolution, B2i2 (11.6%, $n = 15$), C1b13 (20.93%, $n = 27$), and D1g 17.83%, $n = 23$) were the predominant subhaplogroups in their respective macrohaplogroups. Non-Amerindian haplogroups represented 15.5% ($n = 20$) of the lineages: D4p (14.29%), H* (50%), I1* (14.29%), K1a4a1a + 195 (14.29%), L3d1b2 (7.14%), T2* (14.29%), U* (21.42%), and X2 (7.14%). The overall genetic diversity of the sample was 0.987 (Table S4).

The neighbor-joining dendrogram (Figure 2) contained three main clusters: northern Native and rural; south-central Native; and southernmost populations. The sample from Punta Arenas landed between the south-central and southernmost clusters, closest to the Kawésqar, Yámana, and Tehuelche populations (Native communities with a moderate presence in the geographic area near Punta Arenas) and the Santiago population, the only other urban city on the analysis. The next-closest population was the Huilliche, a Native community with a strong presence on the Chiloé archipelago.

The haplotype networks for the B2, C1, and D haplogroups indicated that the population of Punta Arenas shared more haplotypes with south-central Native populations and the population of Santiago than with northern Native or northern rural populations (Figures S2–S4). Most of the subhaplogroups within B2 corresponded to B2i2a, B2i2b, and B2i2b1. In the C1 network (Figure 3), haplotype C1b13 + 16311C + 16343G is shared by the population of Punta Arenas and the Yámana population. Haplotype C1b + 152 + 200G was common to Punta Arenas, Santiago, and Huilliche, while C1b + 16189C + 185A + 16093C + 16126C + 16 344 T was found in both Punta Arenas and Kawésqar individuals. Finally, haplotype C1d + 194 + 146 + 16 274 + 15 311 + 16 533 was common to Punta Arenas, groups of Argentinian Mapuche descent, and one Kawésqar individual. Within haplotype D1g in the D network (Figure 3), D1g + 234G + 140 T was shared by the populations of Punta Arenas and Santiago, while haplotype D1g + 16086C + 16189C + 16 286 T was shared by

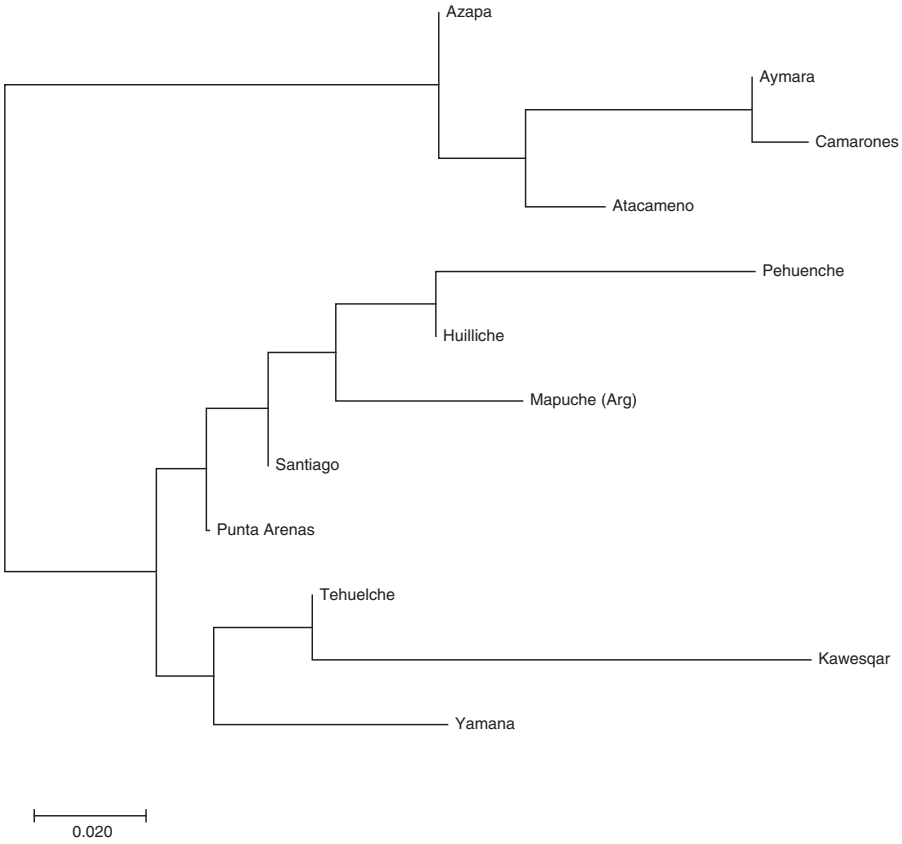


**FIGURE 2** Neighbor-joining dendrogram of Punta Arenas sample and Native, rural, and urban Chilean reference samples, constructed from Fst distance matrix

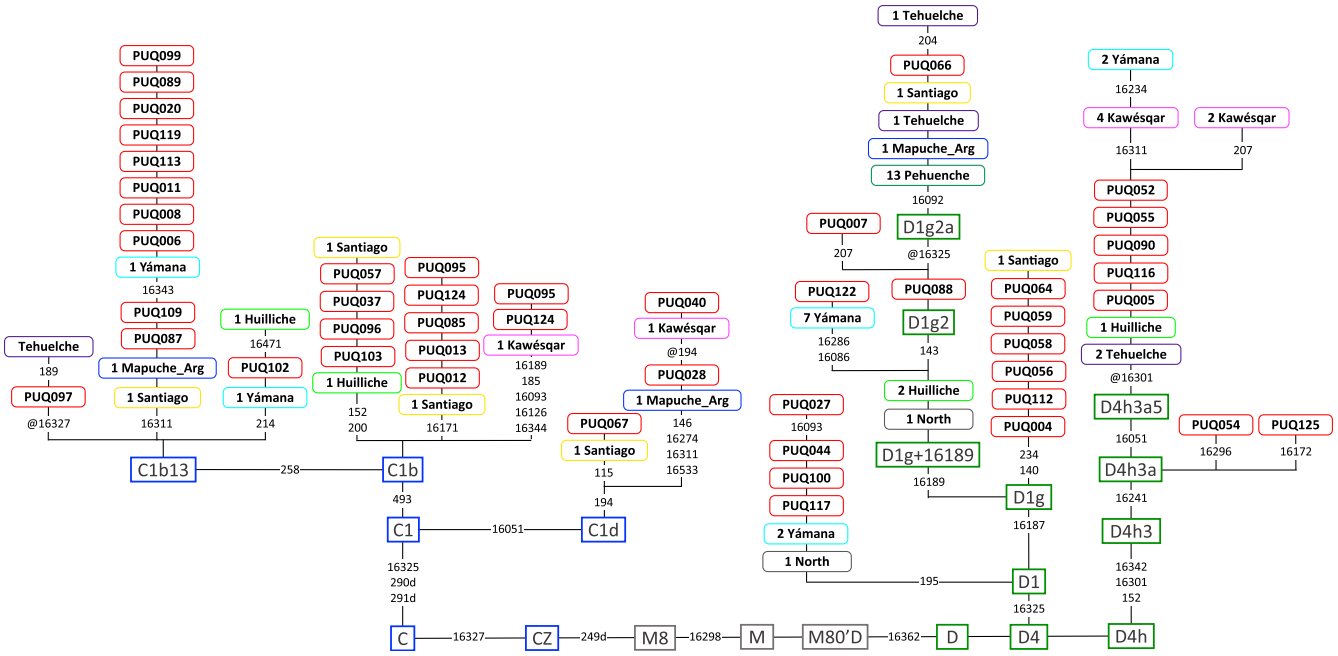


**FIGURE 3** Network representing most noteworthy relationships between haplotypes in haplogroups C and D (for complete networks, see Supporting Figures)

the Punta Arenas sample and the Yámana population. In addition, haplotype D1 + 195C in the D network was common to the population of Punta Arenas, two Yámana individuals, and one sample from northern Chile. Haplogroup D4h3a5 was found in the population of Punta Arenas as well as the Tehuelche and Huilliche populations. Finally, derived haplotype D4h3a5 + @16 301 + 16311C was present in the Kawésqar and Yámana population and D4h3a5 + @16 301 + 207A in two Kawésqar individuals.

## 3.3 | Y-chromosome

Y-chromosome data (Table S5) for the Punta Arenas sample reflects a predominance of the European haplogroup R. R1b ($n = 61$) accounted for 47.29% of the sample. R1a1, highly prevalent in Croatia, was present in 4.65% of cases ($n = 6$). The second-most common haplogroup was I (8.53%, $n = 11$), with 4 individuals characterized as I2a1 (3.1%), which is another haplotype common in Croatian populations. Haplogroups J (8.53%, $n = 11$), containing J2 (4.65%, $n = 6$), and E (7.75%, $n = 10$) were also identified in the sample. In terms of Native American haplogroups, 10 individuals belonged to the Q-M3 haplogroup (7.75%), and 3 carried the mutation Q-M242 (2.3%), which has been found at a low frequency in Croatian populations. Genetic diversity within the sample was 0.7438.

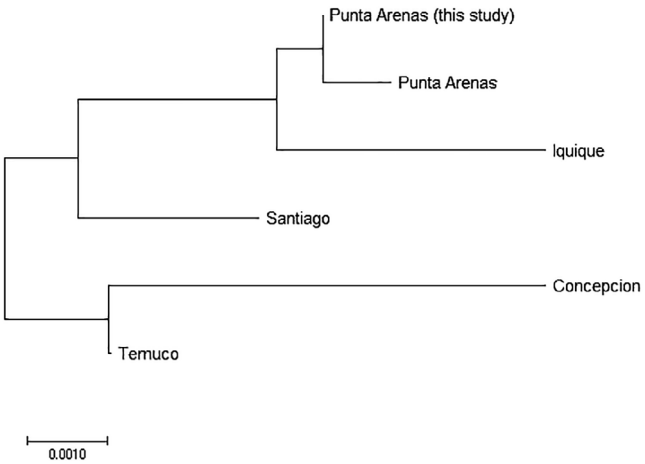


**FIGURE 4** Neighbor-joining dendrogram of Punta Arenas and other Chilean samples, constructed from Fst distance matrix

The neighbor-joining dendrogram (Figure 4) showed an upper cluster that includes the northern and central Chilean populations and a lower cluster that grouped the southern populations. The location of the Punta Arenas sample indicates genetic proximity to other Chilean samples, but no statistically significant relationship with any of these populations. The closest relationship was between the two samples from Punta Arenas (this study and Toscanini et al., 2016), followed by the relationship between the sample studied here and the population of Santiago.

### 3.4 | Nuclear DNA data

The cross-validation error in ADMIXTURE was lowest for $K = 4$ (CVe = 0.48374), with African, European, northern Native and south-central Native genetic components at relative contributions of 3.6, 56.5, 11.3, and 28.6% (Figure 5, Figure S5).

A PCA was performed on the SNPs present on each individual in the Punta Arenas sample and the reference populations (Figure 6); the first component explained 38.14% of the variance and separated African ancestry from the other groups. The second principal component explained 14.8% of the variance and distinguished European from Native American ancestry; the third component explained 2.9% of the variance and separated south-central from northern Native ancestry. Punta Arenas fell between the European and Native American populations, closer to the south-central than the northern Native populations.

A HCPC analysis was performed on the results of a second PCA, built from the ancestry proportions obtained from ADMIXTURE with $K = 4$. The analysis identified five natural clusters, with the population of Punta Arenas belonging to a fifth cluster that was separate from the reference populations, except for the 15.5% ($n = 20$) individuals that fell into the European cluster (Figure S6). Among these individuals, 40% had a Y-chromosome haplogroup associated with Croatian origins, and 45% had a non-Amerindian mtDNA haplogroup.

### 3.5 | Mixed data analysis

The mtDNA and Y-chromosome analyses revealed marked differences in terms of Amerindian and non-Amerindian (European) origins, reflecting a clear sex bias. While 16.27% of lineages were non-Native according to the mtDNA analysis, the Y-chromosome studies indicated only 7.75% Native American ancestry. Most individuals in the sample (77.52%, $n = 100$), therefore, had Amerindian mtDNA and non-Amerindian Y chromosomes.

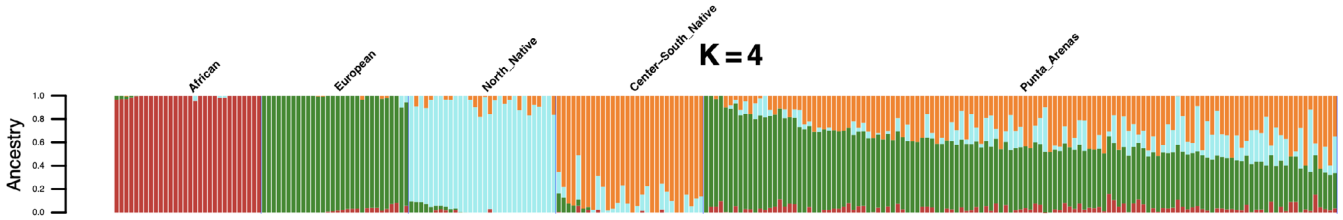


**FIGURE 5** Admixture, $K = 4$. Figure shows the four main genetic components in the Punta Arenas sample (African, European, northern Native, and south-central Native) and their relative frequency

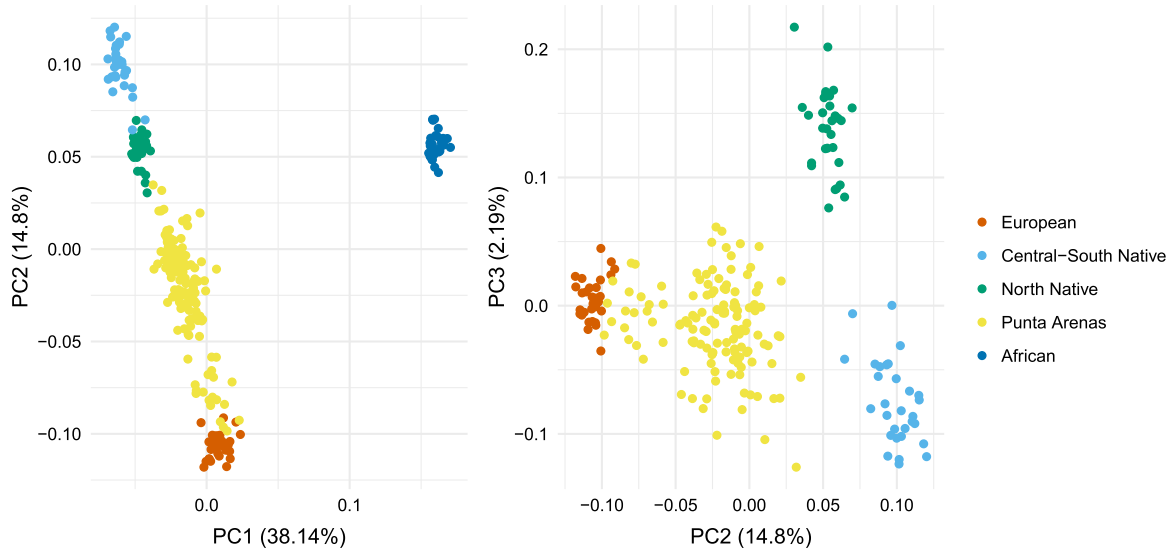


**FIGURE 6** Principal component analysis of individual SNPs. Left: First and second principal components. Right: Second and third principal components

A FAMD (Figure 7; Figure S7) was carried out to determine whether the sex biases evident in these uniparental markers were associated with individual ancestry proportions. The first dimension of the analysis explained 32.4% of the inertia and was associated with a greater degree of European ancestry toward the right and south-central Native American ancestry toward the left. Meanwhile, non-Amerindian mtDNA haplogroups clustered on the positive side of the axis, associated with European ancestry, and Amerindian mtDNA haplogroups mostly on the negative side, associated with south-central Native ancestry. The second dimension explained 22.3% of the inertia and discriminated between northern (on the positive side) and south-central Native ancestry. The third dimension, explaining 19% of the inertia, was associated with African ancestry and also separated Amerindian and non-Amerindian Y-chromosome haplogroups.

## 4 | DISCUSSION

The results show that the population of Punta Arenas is predominantly composed of individuals of mixed ancestry, with European ancestry being, on average, higher Native. The multivariate analysis identified African, European, northern Native, and southern Native populations clusters and characterized individuals of mixed ancestry as a fifth cluster, distinct from these other groups. This mixed-lineage cluster was located between the European and Native reference populations on the resulting. However, a substantial group of Punta Arenas sample belonged to the European rather than the Mestizo cluster, in contrast with the reference populations. This finding indicates the presence of groups which, despite multiple generations in the area, have remained unmixed with the local population.

European ancestry was more prevalent in the Punta Arenas sample than has been observed in the general Chilean population. European ancestry was also more common in this sample than in southernmost Chilean populations studied previously, with the exception of one study performed in Chillán. Correspondingly, the overall Amerindian component was smaller than observed for other Chilean samples (Cifuentes, 2015, 2016; Eyheramendy et al., 2015; Verdugo et al., 2020). When Native ancestry was examined at a higher resolution, this

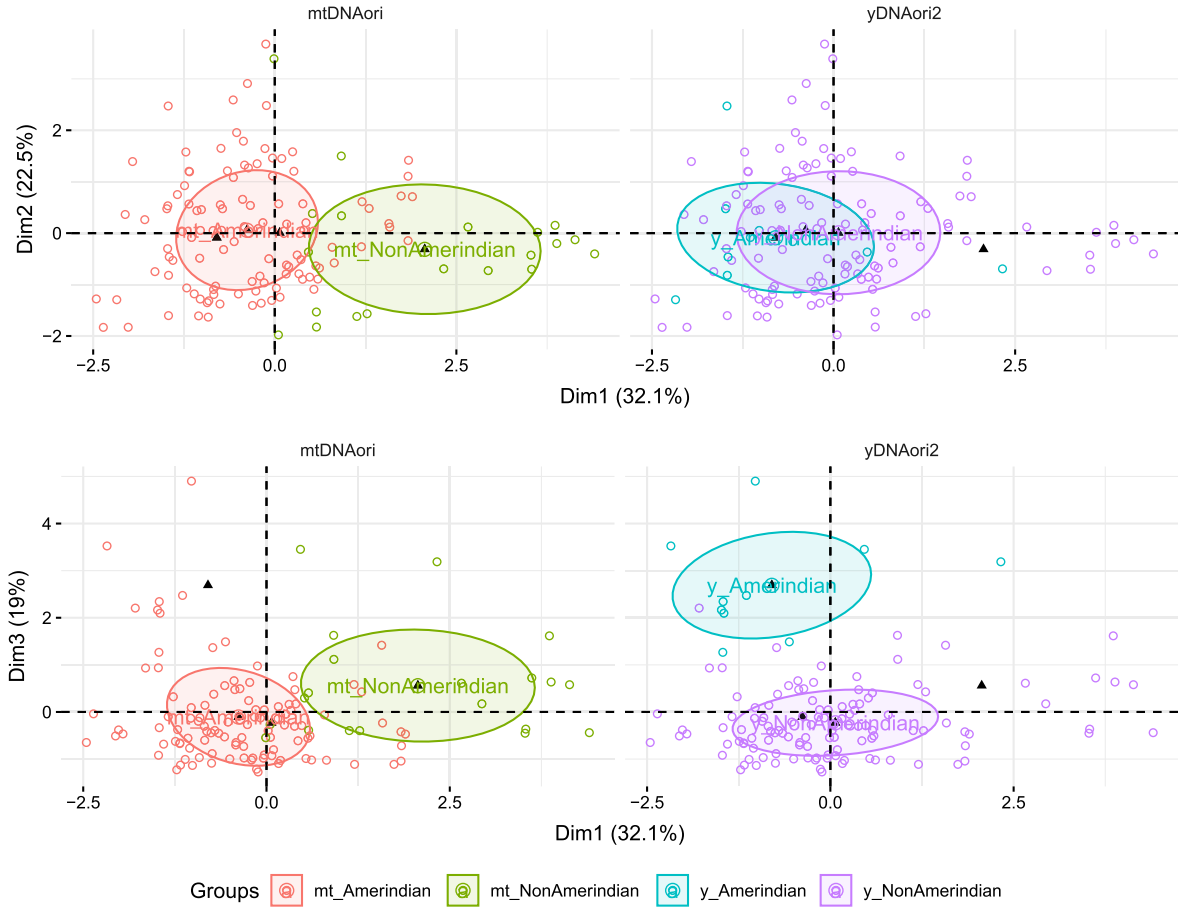


FIGURE 7 Factorial analysis of mixed data for Amerindian and non-Amerindian groups. Upper row: First and second principal components; bottom row: First and third principal components. Right: mtDNA data; left: Y-chromosome data

sample showed a much greater proportion of southern than northern Amerindian markers. The proportion of southern Native markers observed in this study was higher than in any previous Chilean sample studied, with the exception of studies performed in Temuco and Puerto Montt. This finding is consistent with historic migration patterns, as a large proportion of settlers to Punta Arenas arrived from Chiloé, whose Native population is of Huilliche origin (Duquesnoy, 2012; Martinić Beros, 1999b). This study identified 10 individuals with a European ancestry greater than 80%, whose self-declared ancestry was Croatian in four of these cases and Spanish, Chilote, and other European in the rest. The Y-chromosome and mtDNA haplogroups were non-Amerindian in 100% and 80% of these individuals, respectively. These results likely indicate endogamous behavior among European immigrants. These patterns were previously observed by Nock (1990) in a study of marriage records, corroborated by the presence of 19 individuals with European Y-chromosomes and mtDNA in the sample.

The uniparental markers analyzed suggest a gender bias in admixture, with a predominance of paternal lineages of European origin but maternal lineages of Native origin. A total of 77.52% of individuals showed this combination, a finding consistent with observations in other Chilean and Latin American populations. This pattern is a result of predominantly male European colonization (Gómez-Carballa et al., 2016; Rocco et al., 2002; Toscanini et al., 2016; Valenzuela, 2011). In other words, paternal lineage was mainly related to the history of foreign colonization, while maternal lineage was associated with the permanence of the Native population and internal national migration in a large proportion of the sample. Furthermore, mtDNA haplogroups of foreign origin were associated with a high proportion of European autosomal ancestry, on average 68.21%, also suggesting a directionality in the admixture that may also be indicative of endogamous behavior among foreign migrants.

Analysis of the Y-chromosome haplogroups showed low population differentiation between Punta Arenas and other Chilean populations. The high frequency of foreign Y-chromosome markers as compared to Native haplogroups is consistent with results from other urban locations in Chile (Toscanini et al., 2016).

Interestingly, 90% of individuals with a Y-chromosome haplogroup of Native origin had a Spanish surname. However, the self-declared geographic origins of the sample included Spain, other European nations, and Chiloé, reflecting a disparity between family history and self-declared genetic ancestry. Within this same group, 80% had a Native mtDNA haplogroup, lower than figures for the general population of the city. This finding is striking given the origins of the paternal lineages. Furthermore, given that the proportion of European ancestry was lower and the proportion of south-central Native lineages higher than in the general population of the city (50.82 and 32.56%, respectively), one would expect a proportion of Native maternal lineages equal to or even higher than that of the general population.

Among the European haplogroups, the frequency of R1b stands out. This haplogroup is present highly prevalent in Western Europe and its arrival and significant presence in Chile has been associated with Spanish colonization beginning in the sixteenth century (Cifuentes, 2015; Francalacci & Sanna, 2008; Underhill et al., 2014), and its presence in Punta Arenas may attributable primarily to migration from Chiloé after 1845 (Martinić Beros, 1999b; Riveros Quinteros & Fernández Génova, 2018). It is also interesting to note the presence of haplogroups common in Croatia, such as R1a1. The frequency of this haplogroup varies from 20.5 to 38.4% in different areas of that country but is always present in higher proportions than R1b. Furthermore, the Punta Arenas sample included individuals with haplogroup I2a1, which has been found at frequencies of up to 65.9% in Croatia (Barać et al., 2003; Francalacci & Sanna, 2008; Mršić et al., 2012; Pericić et al., 2005; Primorac et al., 2011). These haplogroups have been identified at similar frequencies in previous studies of the city (Toscanini et al., 2016), and are consistent with the regional demographic history, which has included important waves of immigration from Croatia since the 1890s (Martinić Beros, 1977, 1999c; Martinić Beros, 1999b; Mesarić Žabčić & Perić, 2006; Nock, 1990; Ortega Perrier, 1980). Half of the individuals with these latter two haplogroups had a self-declared Croatian geographic origin and surname, while four of the individuals reported that their families immigrated from other parts of Europe and one from Chiloé. Overall, their average ancestry was 72.45% European and 18.14% south-central Native in this subgroup. The mitochondrial haplogroup was of non-Native origin in only two cases.

It is noteworthy to point out that three individuals had a haplogroup corresponding to Q-M242, but not the Q-M3 haplogroup distinctive to America. These three individuals had both a Croatian surname and self-declared Croatian origin, consistent with the presence of markers from the corresponding area of Europe, albeit at a lower frequency (Francalacci & Sanna, 2008; Mršić et al., 2012). Two of these individuals had mitochondrial haplogroups of European origin and overall European ancestry greater than 95%. In addition, 100% of the individuals with a Croatian surname were of self-declared Croatian origin. The average European ancestry was 73.31% in this group, but only 28.57% of these individuals had a non-Amerindian mitochondrial haplogroup.

The analysis of mtDNA sequences allowed us to elucidate the relationship between the current population of Punta Arenas and Native populations from this area and other parts of the country, which may have arrived in national migratory waves. The Punta Arenas sample did not differ significantly from the Santiago reference sample, due to a similar contribution of foreign lineages in both urban populations (Rocco et al., 2002). Noteworthy foreign linages present in the current sample include I1a1, T2, and X2, all of which have been observed in Croatian populations (Barbarić et al., 2020; Cvjetan et al., 2004; Pericić et al., 2005). This finding is therefore consistent historical sources that indicate an influx of Croatian men and women to Punta Arenas since the 1890s (Martinić Beros, 1999c; Nock, 1990). However, these lineages cannot be attributed exclusively to Croatian migration. These uniparental markers are also likely associated with the arrival of other Europeans since 1873, as well as immigration from Chiloé since the middle of the 19th century. The above European lineages are often found in the Mestizo population of Chiloé, which has received European migrants since the sixteenth century (García et al., 2006; Huber et al., 2018; Parson & Dür, 2007).

In terms of comparisons with Native populations, the Punta Arenas sample is most closely related to the Huilliche, Tehuelche, and Yámana reference panels. The first group is native to south-central Chile, and part of its population is located in Chiloé. These lineages may have migrated to Punta Arenas, carried by Native or Mestizo women from the archipelago (Martinić Beros, 1999b; Saldívar Arellano, 2018). The few remaining decedents from the latter two groups inhabit small communities in Chilean and Argentine Patagonia (, 2018; INE, 2017). There may be Mestizo descendants of the Tehuelche and Yámana populations in the current urban population, as is also suggested by the shared lineages observed in the networks. A large proportion of the lineages found in the population of Punta Arenas are shared with south-central Chilean indigenous populations, including subhaplogroups B2i2, C1b13, and D1g as described above (Bodner et al., 2012; de Saint Pierre et al., 2012). The presence of these haplogroups in Punta Arenas is consistent with the hypothesis of migration originating in the Chiloé archipelago. There is also evidence of shared lineages with local indigenous populations within haplogroups C and D, including haplogroups shared between the population of Punta Arenas and contemporary as well as ancient Yámana and Kawésqar populations (de la Fuente et al., 2015; de la Fuente et al., 2018). These communities have inhabited the territory since before the occupation of Mestizo and foreign populations (Harambour, 2018). These findings suggest continuity between the current population and descendants of the original populations.

While the relatively small number of AIMs available allowed for comparisons by continental ancestry with other populations in the national territory, this limitation prevented more precise comparisons. For example, it was difficult to delve more precisely into the structure of the European component, identifying, for instance, specific Croatian components in the populations (Xing et al., 2009). Some comparisons with national reference populations were also challenging, as not all of these samples were characterized at the same resolution using the three types of markers analyzed here. Therefore, additional research regarding the population of Chiloé, especially Y-chromosome data, higher-resolution mitochondrial lineage studies, and collection of nuclear markers from a larger sample, could help to elucidate genealogical ramifications of migrations from Chiloé to Punta Arenas.

This is the first study to genetically characterize the city of Punta Arenas using uniparental and AIMs. This approach allows for analysis of the phenomena of genetic structuring, national and international migration, and admixture from various complementary perspectives, contributing to our understanding of local history in the context of national and global issues in genetic ancestry.

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## AUTHOR CONTRIBUTIONS

**Sandra Flores-Alvarado:** Conceptualization; data curation; formal analysis; investigation; methodology; visualization; writing-original draft; writing-review & editing. **Michael Orellana-Soto:** Data curation; formal analysis; investigation; methodology; visualization; writing-review & editing. **Mauricio Moraga:** Conceptualization; data curation; funding acquisition; investigation; project administration; supervision; writing-review & editing.

## DATA AVAILABILITY STATEMENT

The data that supports the findings of this study are available in the supplementary material of this article.

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## SUPPORTING INFORMATION

Additional supporting information may be found online in the Supporting Information section at the end of this article.

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