



Revealing latitudinal patterns of mitochondrial DNA diversity in Chileans



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ABSTRACT

The territory of Chile is particularly long and narrow, which combined with its mountainous terrain, makes it a unique scenario for human genetic studies. We obtained 995 control region mitochondrial DNA (mtDNA) sequences from Chileans representing populations living at different latitudes of the country from the North to the southernmost region. The majority of the mtDNA profiles are of Native American origin (~88%). The remaining haplotypes are mostly of recent European origin (~11%), and only a minor proportion is of recent African ancestry (~1%). While these proportions are relatively uniform across the country, more structured patterns of diversity emerge when examining the variation from a phylogeographic perspective. For instance, haplogroup A2 reaches ~9% in the North, and its frequency decreases gradually to ~1% in the southernmost populations, while the frequency of haplogroup D (sub-haplogroups D1 and D4) follows the opposite pattern: 36% in the southernmost region, gradually decreasing to 21% in the North. Furthermore, there are remarkable signatures of founder effects in specific sub-clades of Native American (e.g. haplogroups D1j and D4p) and European (e.g. haplogroups T2b3 and K1a4a1a+195) ancestry. We conclude that the magnitude of the latitudinal differences observed in the patterns of mtDNA variation might be relevant in forensic casework.

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1. Introduction

The territorial shape of Chile is particularly long (from North to South there are more than 4200 km) and comparatively narrow (from East to West there are about 445 km on average), and it has more than 6400 km of Pacific coast. These features, combined with the mountainous character of the country, make it a unique scenario for human and forensic genetic studies.

Historically, Chile was occupied by different Native American peoples at different latitudes, some of them even crossing the Andes mountains and reaching the Atlantic ocean [1]. Aymaras,

Atacameños and Diaguitas settled the North, while the region South of the Aconcagua river was inhabited by semi-nomad Mapuche. Several Native American communities inhabited also the southernmost end of Chile (e.g. Aonikenk, Caucahue, Chonos, Selknam, Kawésqar, etc). The Spaniards arrived in Chile in 1520, and several Spanish settlements were established after these initial expeditions.

The 1907 census estimated that more than 101,000 indigenous people (about 3% of the population of that time) lived in the country, most of them in the South provinces of Cautín and Valdivia (this census did not consider the populations from the North and the southernmost end of the country). In the 2002 census, Chile officially recognized the existence of nearly 700,000 Native Americans (about 4.6% of the total population), belonging to one of the following eight ethnic categories: Aymara, Quechua, Atacameño, Kola, Rapanui, Mapuche, Yagán, and Kawésqar. In 2008, the Diaguita were added to the list. By far,

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the most numerous group were the Mapuches (>87%), mostly living in Central Chile, in the provinces of Araucanía and Región Metropolitana.

Today, there are about 18 million people living in Chile, half of them in the central region of the country. Several official documents make use of the traditional and old-fashion ethnic nomenclature system to classify Chileans into main population categories. For instance, Medina-Lois and Kaempffer [2] considered the existence of the following main 'ethnic groups': 'blancos' ('whites') or 'caucásicos' ('Caucasians') (30%), 'mongoloid aborígenes' (5%), and 'mestizos' (65%). Moreover, these authors claimed that the 'mestizo' groups conformed an 'homogeneous' group; whereas 'blancos' originated from different Spanish regions, mainly from Castilla (Central Spain), Andalusia (Southern Spain), and the Basque Country (Northern Spain). These authors also mentioned a limited amount of sub-Saharan Africans arriving in the territories of Chile (1%), with a peak of 25,000 people during the colonial period.

Only a few genetic studies have been undertaken on Chileans, and most of them focused on uniparental markers. A few mtDNA

haplotypes were obtained in the early study by Horai et al. [3], one of the first attempts to reconstruct the initial peopling of the Americas. This study revealed for the first time the main Native American mtDNA composition of Chileans. Moraga et al. [4] analyzed Restriction Fragment Length Polymorphisms (RFLPs) and the mtDNA control region in Chilean aboriginal populations from the southernmost end of the country; their data showed the existence of haplogroups C and B at high frequency (43% and 47.7%, respectively) in the Pehuenche, Mapuche and Yaghan. García-Bour et al. [5] analyzed the mtDNA control region I (HVS-I) of 24 skeletal samples from 60 ancient individuals sampled in Patagonia-Tierra del Fuego. Their analysis indicated a Native American ancestry closely related to present-day Chileans and Argentineans but also the signatures of population bottlenecks and isolation. According to these authors, the DNA evidence suggested an early genetic diversification of the Fueginians right after their arrival into the southern cone. Moraga et al. [6] obtained RFLP and control region results for ancient remains found in archaeological cemeteries located in North Chile. Their analyses indicated a Native American haplogroup composition that differed from present-day Chileans

Table 1
Molecular diversity indices in Chilean mtDNA sequences, computed by recruitment sites and main haplogroups. "African-L" stands for haplotypes of recent sub-Saharan African ancestry, that is "L(xM,N)", while the category "Others" is an artificial polyphyletic cluster (paragroup). Codes are as follows: HG = haplogroup, n = sample size, h = number of different haplotypes, S = segregating sites, HD = haplotype diversity, M = mean number of nucleotide differences, π = nucleotide diversity.

Region/HG	n	h	S	HD	M	π
All Chile	995	256	125	0.9701 ± 0.0025	7.598573 ± 3.548369	0.022218 ± 0.011472
A2	57	21	23	0.9236 ± 0.0189	2.753798 ± 1.480738	0.008052 ± 0.004803
B2/B4b	257	62	60	0.8838 ± 0.0148	2.480027 ± 1.343928	0.007252 ± 0.004347
C1	309	63	58	0.8452 ± 0.0190	2.181326 ± 1.211527	0.006378 ± 0.003919
D1/D4	254	52	43	0.9035 ± 0.0143	3.323828 ± 1.712919	0.009719 ± 0.005541
African-L	11	10	30	0.9818 ± 0.0463	10.803550 ± 5.329739	0.031589 ± 0.017581
Others	107	49	57	0.9494 ± 0.0114	6.251282 ± 2.991164	0.018279 ± 0.009686
Iquique	200	92	76	0.9703 ± 0.0054	7.611104 ± 3.565444	0.022255 ± 0.011536
A2	18	11	15	0.9281 ± 0.0401	2.711694 ± 1.510317	0.007929 ± 0.004938
B2/B4b	61	23	30	0.8650 ± 0.0344	2.423539 ± 1.332800	0.007086 ± 0.004322
C1	68	29	36	0.8872 ± 0.0312	2.442144 ± 1.339191	0.007141 ± 0.004341
D1/D4	42	20	23	0.9141 ± 0.0309	4.058251 ± 2.065747	0.011866 ± 0.006708
African-L	2	-	-	-	-	-
Others	9	7	14	0.9167 ± 0.0920	4.132136 ± 2.268421	0.012082 ± 0.007525
Santiago	203	108	82	0.9722 ± 0.0058	7.569630 ± 3.547397	0.022133 ± 0.011478
A2	15	9	13	0.8476 ± 0.0878	2.389730 ± 1.375602	0.006988 ± 0.004510
B2/B4b	53	27	26	0.8846 ± 0.0372	2.537371 ± 1.386252	0.007419 ± 0.004497
C1	53	23	27	0.7779 ± 0.0615	2.091811 ± 1.187158	0.006116 ± 0.003851
D1/D4	46	21	26	0.9295 ± 0.0229	3.796062 ± 1.946776	0.011100 ± 0.006319
African-L	4	4	16	1.0000 ± 0.1768	10.544126 ± 6.108788	0.030831 ± 0.021329
Others	32	25	34	0.9657 ± 0.0236	4.556986 ± 2.299721	0.013325 ± 0.007480
Concepción	201	83	68	0.9674 ± 0.0052	7.716728 ± 3.610774	0.022564 ± 0.011683
A2	13	7	9	0.8333 ± 0.0861	2.826060 ± 1.592343	0.008263 ± 0.005234
B2/B4b	61	19	25	0.8732 ± 0.0287	2.413227 ± 1.328212	0.007056 ± 0.004307
C1	56	20	25	0.8104 ± 0.0422	1.634466 ± 0.979449	0.004779 ± 0.003177
D1/D4	48	21	26	0.9043 ± 0.0301	2.894508 ± 1.547273	0.008463 ± 0.005022
African-L	1	-	-	-	-	-
Others	22	15	31	0.9567 ± 0.0276	7.494902 ± 3.639522	0.021915 ± 0.011873
Temuco	194	87	80	0.9662 ± 0.0069	7.579295 ± 3.552223	0.022162 ± 0.011494
A2	9	7	10	0.9444 ± 0.0702	3.135884 ± 1.791189	0.009169 ± 0.005942
B2/B4b	53	22	27	0.9100 ± 0.0253	2.424790 ± 1.336115	0.007090 ± 0.004335
C1	60	21	27	0.7751 ± 0.0559	1.674811 ± 0.996841	0.004897 ± 0.003233
D1/D4	48	20	22	0.9184 ± 0.0270	3.369515 ± 1.757358	0.009852 ± 0.005703
African-L	3	3	15	1.0000 ± 0.2722	13.238315 ± 8.262198	0.038709 ± 0.030131
Others	21	14	29	0.9238 ± 0.0426	6.698225 ± 3.290897	0.019585 ± 0.010741
Punta Arenas	197	67	64	0.9543 ± 0.0067	7.264731 ± 3.416729	0.021242 ± 0.011055
A2	2	-	-	-	-	-
B2/B4b	29	13	15	0.8867 ± 0.0388	2.546178 ± 1.408614	0.007445 ± 0.004584
C1	72	19	24	0.8541 ± 0.0276	2.636081 ± 1.424201	0.007708 ± 0.004616
D1/D4	70	22	19	0.8422 ± 0.0356	2.758389 ± 1.478744	0.008065 ± 0.004793
African-L	1	-	-	-	-	-
Others	23	11	22	0.7747 ± 0.0901	5.636053 ± 2.805883	0.016480 ± 0.009150

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