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Short communication

Analysis of Y-chromosome STRs in Chile confirms an extensive introgression of European male lineages in urban populations

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ABSTRACT

We analyzed the Y chromosome haplotypes (Yfiler) of 978 non-related Chilean males grouped in five sampling regions (Iquique, Santiago de Chile, Concepción, Temuco and Punta Arenas) covering main geo-political regions. Overall, 803 different haplotypes and 688 singletons were observed. Molecular diversity was moderately lower than in other neighboring countries (e.g. Argentina); and AMOVA analysis on Y-STR haplotypes showed that among variation within Chile accounted for only 0.25% of the total variation. Punta Arenas, in the southern cone, showed the lowest haplotype diversity, and discrimination capacity, and also the highest matching probability of the five Chilean samples, probably reflecting its more marked geographic isolation compared to the other regions. Multidimensional scaling (MDS) analysis based on R_{ST} genetic distances suggested a close proximity of Chilean Y-chromosome profiles to European ones. Consistently, haplogroups inferred from Y-STR profiles revealed that the Native American component constituted only 8% of all the haplotypes, and this component ranged from 5% in the Centre of the country to 9–10% in the South and 13% in the North, which is in good agreement with the distribution of Native American communities in these regions. AMOVA computed on inferred haplogroups confirmed the very low among variation observed in Chilean populations. The present project provides the first Chilean dataset to the international Y-chromosome STR Haplotype Reference Database (YHRD) and it is also the first reference database for Y-chromosome forensic casework of the country.

1. Introduction

Chile has an unusual ribbon-like shape, with more than 4300 km long and on average 175 km wide. Climatic conditions varied from the world's driest desert in the Atacama region (North) to a Mediterranean-like climate in the centre. Chile has a multi-ethnic society that was mainly originated by the admixture between local Native populations and Europeans. The main proportion of Europeans were Spaniards that arrived in the country in the latter part of the XVI century, most of them males [1]. Other minor European groups arrived to Chile in more recent times such as Croatians and Germans.

The national census (INE; http://www.ine.cl) carried out in 1992 aimed to characterize the indigenous population of the country by recording self-reported ancestry of Chileans. The 1992 census considered only three indigenous populations (Mapuche, Aymara, and Rapanui; Ley N°19.523; "Ley Indígena"), but the posterior modification of this law in 1993 adopted the allocation of each person to one of the following eight population groups: Alacalufe (Kawaskar), Atacameño, Aymara, Colla, Mapuche, Quechua, Rapanui, and Yámana (Yagán). The last official census from 2002 indicated that only 4.6% of the Chileans (n = 692.192 persons in a total population of about 15 million) declared to belong to one of these eight ethnic groups; most of them belonging to the Mapuche (87.3%), and being the Aymara the second most important group (7%).

In contrast to other South American countries where Y-chromosome genetic studies have been very popular on different

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anthropological and forensic contexts, there are only a few studies carried out on the Y-chromosome variation in Chile, all of them related to small-scale sampling or the genotyping of a few Y-STRs. The pioneering study by Cifuentes et al. [2] based on autosomal STR markers, suggested that admixed Chileans had and aboriginal admixture of around 40%, but this percentage depended on the socioeconomic stratum, with less than 10% in their social upper strata and greater in social lower strata. Later, Cifuentes et al. [3] analyzed the DYS19 and DYS199 markers in two Chilean population samples of mixed ancestry and inferred admixture proportions from them. By comparing these results on the Y-chromosome with autosomal data the authors claimed an asymmetric pattern of genome admixture between the European and Native American components, a pattern that is also observed in many other regions of South America [4–6]. According to these authors the autosomal Native component was around 40%, while the Native Y-chromosome component was below 20%. More recently, Lardone et al. [7] analyzed the prevalence of Amerindian Y-chromosomes in Chilean patients with spermatogeneic failure and their association with classical and/or AZFc-partial Y-chromosome deletions; this study showed the presence of haplogroup Q1a3a in 9% of their main control group, but frequencies ranged from 8.2% to 11.8% in their various sub-control groups.

Chile is one of the very few larger unsampled countries (together with i.e. Canada and New Zealand) in the Y-Chromosome STR Haplotype Reference Database (YHRD), the international reference forensic database that currently contains Y-chromosome haplotypes sampled worldwide (e.g. 129 countries represented in a collection of >154,000 minimal haplotypes [MHT]; Release 49, 2015/Feb/27). The present study is therefore the first attempt in characterizing the Y-chromosome variation of Chile by way of genotyping one of the forensic Y-STR reference panel (namely, the Yfiler; see below). This project was prompted by the need of having population haplotype frequencies from randomly drawn samples of individuals in Chile for the purpose of human identification casework being done in the context of Human Rights Program of the Medico Legal Services of the country. In addition, the populations analyzed in the present project represent a large sampling effort that has a wide geographic coverage, representing the main regions from northern to southernmost Chile.

2. Material and methods

2.1. Sampling

Buccal swab from 978 unrelated males from urban populations were collected after written informed consent at five sample collection sites of Chile, namely, Iquique (n = 196; YHRD accession number: YA004059), Santiago de Chile (n = 196; YA004061), Concepción (n = 198; YA004058), Temuco (n = 194; YA004062) and Punta Arenas (n = 194; YA004060) (Fig. 1). Sampling was carried out at random without taking social strata into consideration; therefore, there is no evidence indicating that social strata were differently represented at the five recruitment sites.

The samples analyzed in the present study represent a subset of the samples genotyped for a panel of autosomal STRs in Toscanini et al. [8].

Beside the Chilean samples, various datasets from Purps et al. [9] were used for population comparisons, representing populations from (i) South America (n = 487): Bolivia ('Mestizo' and Native American), Peru, Brazil ('Admixed' from São Paulo and Río de Janeiro, São Gabriel de Cachoeira, Native Americans), (ii) Europe (n = 2168): Aragón, Asturias, Barcelona, Galicia, Madrid (all from Spain), Brescia, Calabria, Liguria, Marche, Milano, northeastern Italy, Puglia, Ravenna, Sicily, Tuscany (all from Italy), (iii) Asia: Yunnan (n = 101), and (iv) Africa (n = 136): Ibadan from Nigeria and Zimbabwe.

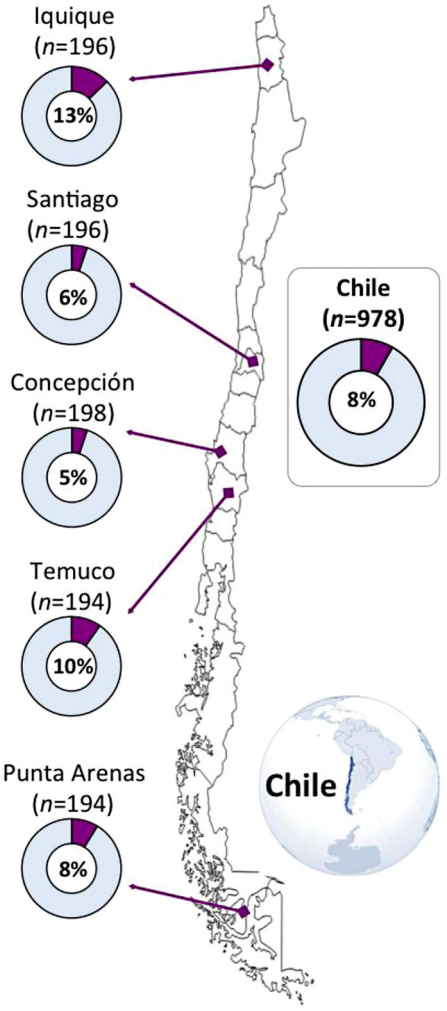

Fig. 1. Map of Chile showing the sampled regions in the present study. The pie charts indicate the frequency of the Native American haplogroup frequencies vs. the rest of the haplogroups merged in a single category.

In addition, further data from the neighboring populations of Argentina (bordering at the East of Chile in all its extension from North to South) analyzed in Toscanini et al. [10,11], were also added because its geographic proximity and comparable past and recent demographic history.

2.2. DNA extraction and genotyping

DNA was extracted from buccal swabs using the DNA IQTM System (Promega Corporation, Madison, USA) on the Tecan Freedom EVO1 100 (Tecan Group Ltd., Männedorf, Switzerland).

The seventeen Y-STR markers included in the AmpF/STR® Yfiler™ kit (Life Technologies, USA; DYS19, DYS385a/b, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS437, DYS438, DYS439, DYS448, DYS456, DYS458, DYS635, Y-GATA-H4), were analyzed following manufacturer instructions. All samples were subjected to electrophoresis on the ABI PRISM1 3130xl Genetic Analyzer (Applied Biosystems, USA). DNA extraction and Y-STR genotyping were carried out in the Laboratory of Human Identification of the University of North Texas (UNT) Health Science Center.

Amplified products were subsequently analyzed with the GeneMapper v3.2 software (Applied Biosystems). Only