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Analysis of ethnic origin asymmetries in uniparental lineages in Argentinian population

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ABSTRACT

With the aim to study Argentinian ethnic structure and its miscegenation, we analyzed the mitochondrial DNA (mtDNA) and Y-chromosome Short Tandem Repeats (Y-STR) of 1580 blood samples from male donors. Our results showed that the population has a greater presence of Native American lineages on the maternal side (68.73 %) regarding the paternal line (4.18 %), alongside a majority of Eurasian origins in the paternal line (92.47 %). From the independence analysis, a significant difference was observed when compared with that expected by random crossings. The results are in agreement with a model where the miscegenation of native women with European men prevails, which has been widely observed in Argentina's migratory currents and population history.

1. Introduction

Argentinian ethnic composition is the result of the contribution of three main elements: Native American population, whose Asian origin has already been confirmed [1], the European contingents -mainly male- during both the Spanish conquest and the overseas immigration as a result of European wars and famines, and finally the Africans brought as slave labor.

Miscegenation processes in Argentina have been based on asymmetric male and female gene flow and migration, resulting in the conservation of native maternal lineages in high proportions, contrary to what happened with native paternal lineages. This is due to a very frequent interbreeding, both voluntary and forced, between native women and migrant men. This process is common to all Latin American region and began during the Spanish conquest, with the arrival of single European men, continuing later on during the colonial period. Overall, it lasted until the end of the 19th century and the beginning of the 20th, coinciding with waves of immigration from Europe with a higher ratio of males.

The study and analysis of mitochondrial DNA (mtDNA) and Y-chromosome Short Tandem Repeats (Y-STR) haplotypes allows to study the genetic composition of both the female and male contribution to the populations as well as the different haplogroups frequencies distribution.

The present study aims to analyze Argentinian ethnic structure and its miscegenation.

2. Materials and methods

We analyzed the mtDNA Control Region and the Y-STR of 1580 blood samples from unrelated male Argentinian donors. All the samples provided were analyzed in the BNDG within Law 26.548 and accompanied by a signed consent form accepting the use of their genetic profiles for population studies.

DNA from blood samples collected on FTA cards was extracted using *QIAmp DNA Mini Kit* in a *QIAcube System (Qiagen)* or *Chelex-100 (Promega)*.

Full mtDNA control region sequences (16024-576) were generated by Sanger method using *BigDye Terminator v1.1 (Applied Biosystems)*. Separation and detection were achieved by capillary electrophoresis on an *ABI 3500*. MtDNA sequences were analyzed using *Sequencing Analysis Software v6.0*, compared to the revised Cambridge Reference Sequence [2] using *SeqScape v3.0* software, and the haplotype classification was carried out following the guidelines proposed by the International Society for Forensic Genetics [3]. Haplogroups were attributed and submitted for quality control via EMPOP [4] and will be available under the accession number EMP00842. Mitochondrial haplogroups A2, B2, C1 and D1+D4h3a were considered to be of Native American origin.

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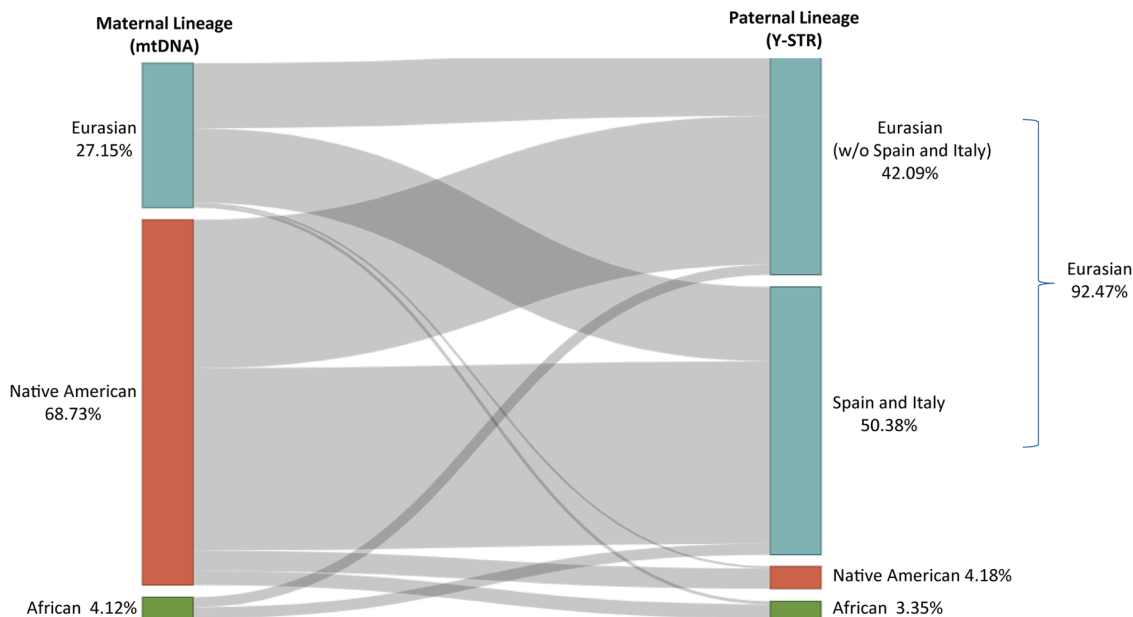


Fig. 1. Sankey diagram, representing the correspondence between maternal lineage and paternal lineage for each individual analyzed (N = 1580). The connector's width reflects the number of individuals with the respective combination. The values show the percentage of the population analyzed for each associated continental origin.

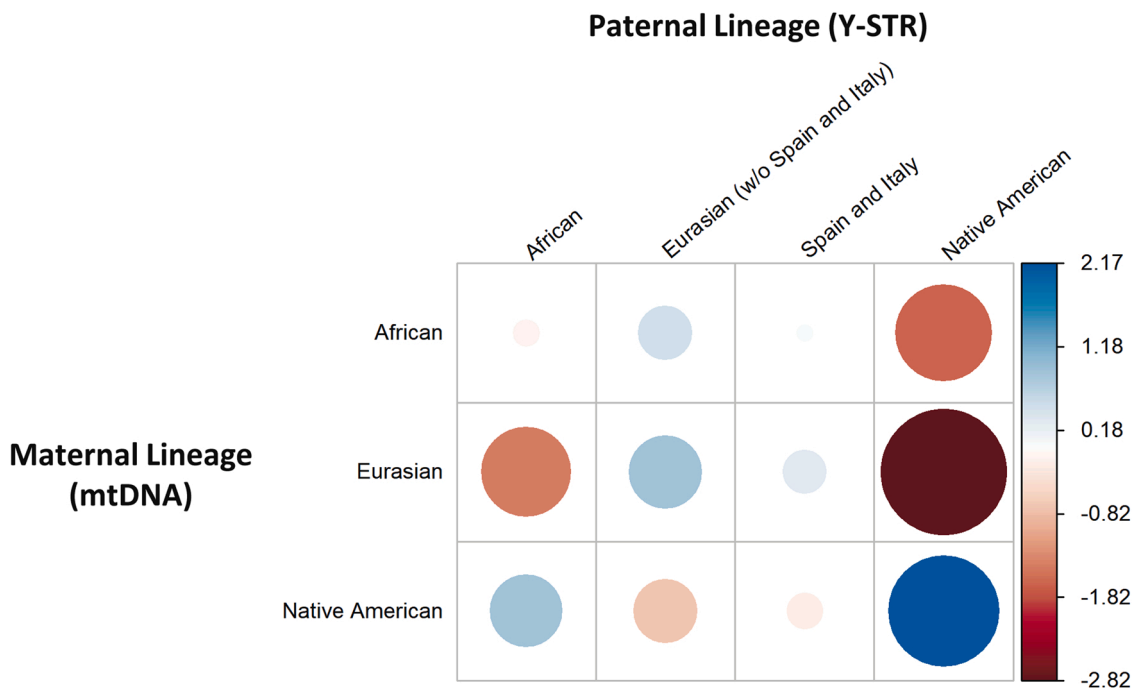


Fig. 2. Deviations were detected for the observed interbreeding between maternal and paternal origin, compared to the expected deviation in the case of independent origin. Smaller circle size and intensity of colour represent a lower deviation. The red and blue colour range indicates underrepresented and overrepresented associations respectively. (For interpretation of the references to colour in this figure legend, the reader is referred to the web version of this article.)

However, haplogroups L, M1 and U6 were considered as African contributions, while the remaining haplogroups from macrohaplogroups M and N were assumed to have a Eurasian origin [5].

Y-STRs profiles were obtained with Applied Biosystems kits: *Yfiler™ PCR Amplification* (293 samples) or *Yfiler Plus Yfiler™ Plus PCR Amplification* (1287 samples). Capillary electrophoresis was done in an *ABI 3500 Genetic Analyzer* and data analyzed with *GeneMapper X-ID* software. Y-STR data was included and is available in the YSTR database (<http://www.yhrd.org>) under the accession number YA004634.

Haplogroups were assigned using Haplogroup Predictor (<http://www.nevgen.org>). Haplotypes belonging to haplogroup Q were considered to be of Native American origin and haplogroups E1b1a and T of African origin. The remaining haplogroups were considered to come from Eurasia and particularly haplogroup R1b as the main contribution from Spain and Italy [6].

3. Results and discussion

3.1. Analysis of the origin of mtDNA and Y-STR haplogroups

A clear asymmetric contribution was observed when the maternal (mtDNA) and paternal (Y-STR) lineages origins were analyzed. A prevalence of Native American maternal lineages (68.73 %) was observed compared to the paternal contribution, associated with haplogroup Q (4.18 %). Interestingly, the paternal lineages showed a high proportion of Eurasian origins (92.47 %), mainly from Spain and Italy (50.38 %). Regarding the contribution of African origin, very similar values were obtained for both lineages (Fig. 1).

3.2. Independence analysis between maternal and paternal associated origins

The independence analysis showed a significant difference between the observed associations of maternal and paternal origins and the expected ones by chance (p -value < 0.05 ; Freeman-Halton test).

When analyzing the deviances from the independence model (Fig. 2), we noticed that the most significant one was given by a greater association of the Native American paternal lineage with a maternal lineage of the same origin.

3.3. Analysis of Native American origin mtDNA haplogroups

All four major Native American haplogroups in Argentinian maternal lineages showed similar representation and miscegenation with the different paternal lineage origins analyzed (Supplementary Fig. 1).

4. Conclusions

The higher presence of Native American maternal origin lineages, in

marked contrast to the predominantly Spanish and Italian paternal origin, showed a strong asymmetry in the distribution of origins in the Argentinian population.

These results are in agreement with a model where there is a majority of miscegenation of Native American women with European men, which has been widely observed during the American conquest and the migratory currents throughout the history of Argentine populating.

Conflict of interest

None.

Appendix A. Supporting information

Supplementary data associated with this article can be found in the online version at [doi:10.1016/j.fsigs.2022.10.007](https://doi.org/10.1016/j.fsigs.2022.10.007).

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