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Genetic Diversity in the Dominican Republic: Implications for the Population and Demographic History of Hispaniola

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Genetic Diversity in the Dominican Republic: Implications for the Population and Demographic History of Hispaniola

ELIZABETH R. OAKLEY¹, ROBERT PAULINO-RAMIREZ^{2,3}, BERNARDO VEGA⁴, MIGUEL G. VILAR⁵, AIDA MENCIA-RIPLEY³, SUZANA GUERRERO-MARTINEZ^{2,3}, ARISMENDY BENITEZ^{2,3} and THEODORE G. SCHURR¹

¹Anthropology, University of Pennsylvania, ²School of Medicine, Universidad Iberoamericana (UNIBE), ³Research Department, UNIBE, ⁴Office of the President, Dominican Academy of History, ⁵Science and Exploration, National Geographic Society

Purpose

By sequencing mtDNAs from 472 individuals living in the Dominican Republic, we aimed to elucidate the following aspects of the population and demographic history of Hispaniola:

- 1) Describe genetic variation in the country today and link it to historical settlement patterns;
- 2) Identify indigenous haplotypes and their affinities with those from other Caribbean islands;
- 3) Describe the history of the Trans-Atlantic slave trade in the Dominican Republic; and
- 4) Assess the impact of European colonization of the Dominican Republic

Background

Geography

The Dominican Republic occupies the eastern two-thirds of the island of Hispaniola, which is situated centrally in the Caribbean basin and is the second largest country in the Caribbean (Figure 1). The country plays host to several different ecological zones including the fertile Cibao Valley, the semi arid San Juan Valley, the hot and arid Neiba Valley, the fertile Caribbean coastal plain, and four mountain ranges that make up 80% of the country.

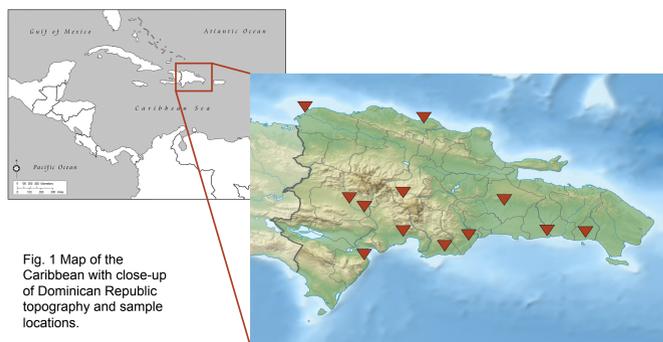


Fig. 1 Map of the Caribbean with close-up of Dominican Republic topography and sample locations.

Demographic History

The Dominican Republic has been called the "Island of firsts," including the first permanent European settlement, the first massacre of Amerindians (Taíno), the first importation of slaves from Africa, and the first revolt against Spanish authority (Sagas and Inoa 2015). In addition, the Caribbean has played host to migrants from other Caribbean islands, West and East Asia, and Jewish populations from Europe throughout the twentieth century. Unstable rule during the island's history, including periods of authoritarian dictatorship and ethnic cleansing have led to competing claims of racial purity and the contemporary deportation and stripping of rights from those Dominicans who are of Haitian descent. The complicated demographic history of the Dominican Republic has real social consequences for those inhabiting the country today. The current study thus hopes to demonstrate how this history enriches the country's great genetic diversity, which should be embraced over any claims of genetic purity. Outlined below are highlighted demographic events that contribute to this diversity (Pons 1998; Keegan 2013; Sagas and Inoa 2015; Wilson 1997).

5000 B.C.

Hispaniola was first occupied during the Mesoinidian Period. The Taíno are hypothesized to be part of a second wave of migration into the Caribbean, called the Saladoid period (500 BC-600AD).

1492

The Santa Maria runs aground December 6, 1492 on Hispaniola and the settlement of La Navidad is created. The capital is moved to Santo Domingo in 1497. Throughout this time members of the Sephardic Jewish population were exiled to Hispaniola.

1503

The encomienda system is implemented to force the Taíno to labor for the Spanish colonists.

1520

By this time African slaves have become the primary source of labor in Hispaniola and are brought in from diverse areas of the African continent.

1542

The population of indigenous Taíno has fallen to an estimated 26,000 due to disease and demands of the encomienda and repartimiento systems. Original population estimates vary widely between several thousand to one million.

1603

The remaining "few thousand colonists and their slaves" inhabited remote coves and beaches far from Santo Domingo and at this time were ordered to relocate to Santo Domingo.

1844

The Dominican Republic wins War of Independence from Haiti.

1900s

After a brief period of rule by Heucreux, during which laborers from SE Asia and other islands arrive to supply the sugar economy, the US intervenes. This begins a chain of unstable authoritarian governments.

1930

From 1930-1961 Trujillo ruled the Dominican Republic and ordered the massacre of any Haitians living in the country, although many black Dominicans were also murdered.

2010

The 2010 census records the following population breakdown: 73% mixed (African-European); 16% white European (Spanish and French); 11% black African

Acknowledgements and References

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Results

mtDNA Haplogroup Frequencies

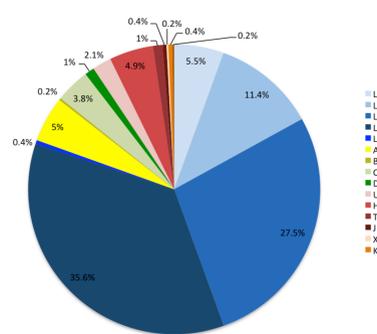


Fig. 2 Distribution of mtDNA haplogroups in the Dominican Republic. L types are shown in shades of blue, Indigenous types in yellow and green, and European types in shades of red and orange.

To understand the complicated demographic history across the Dominican Republic, we undertook a detailed mtDNA haplotype analysis. This analysis included individuals from 12 locations to obtain a total of 472 individuals. The distribution in Figure 2 shows that L types make up 80.4% of the population, with L2 and L3 being the most frequent. Indigenous A, B, C, and D types make up only 10% of the population, with A and C being the most frequent. This pattern is similar to that observed in Puerto Rico with higher frequencies of A2 and C1 types (Vilar et al 2014). In addition, less than 10% of the population displayed European haplotypes.

In each sample location (n=12), samples were collected from an average of 41 individuals. In order to assess the genetic diversity across the country, haplogroup frequencies were also compared across sample locations (Figure 4). Although the influences on the diversity at the sample locations is not yet clear, they could reflect long- and short-term historical settlement patterns on the island, as well as pre-contact settlement by Taíno populations.

Network Analysis

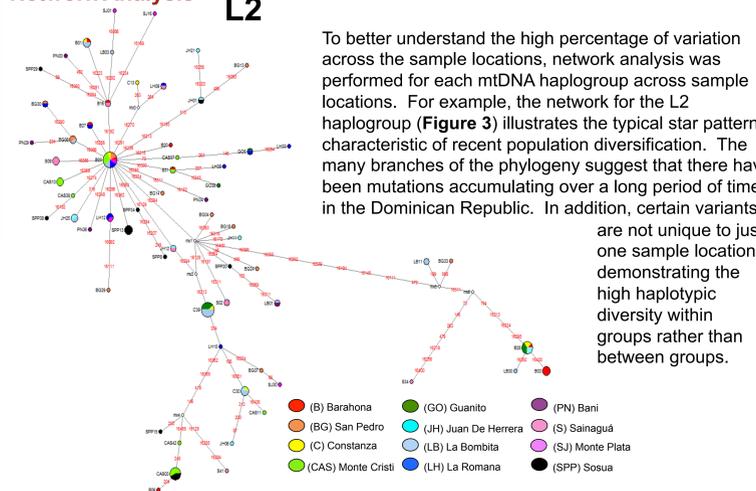


Fig. 3 (Above) Median Joining Network for Haplogroup L2 showing 62 distinct haplotypes.

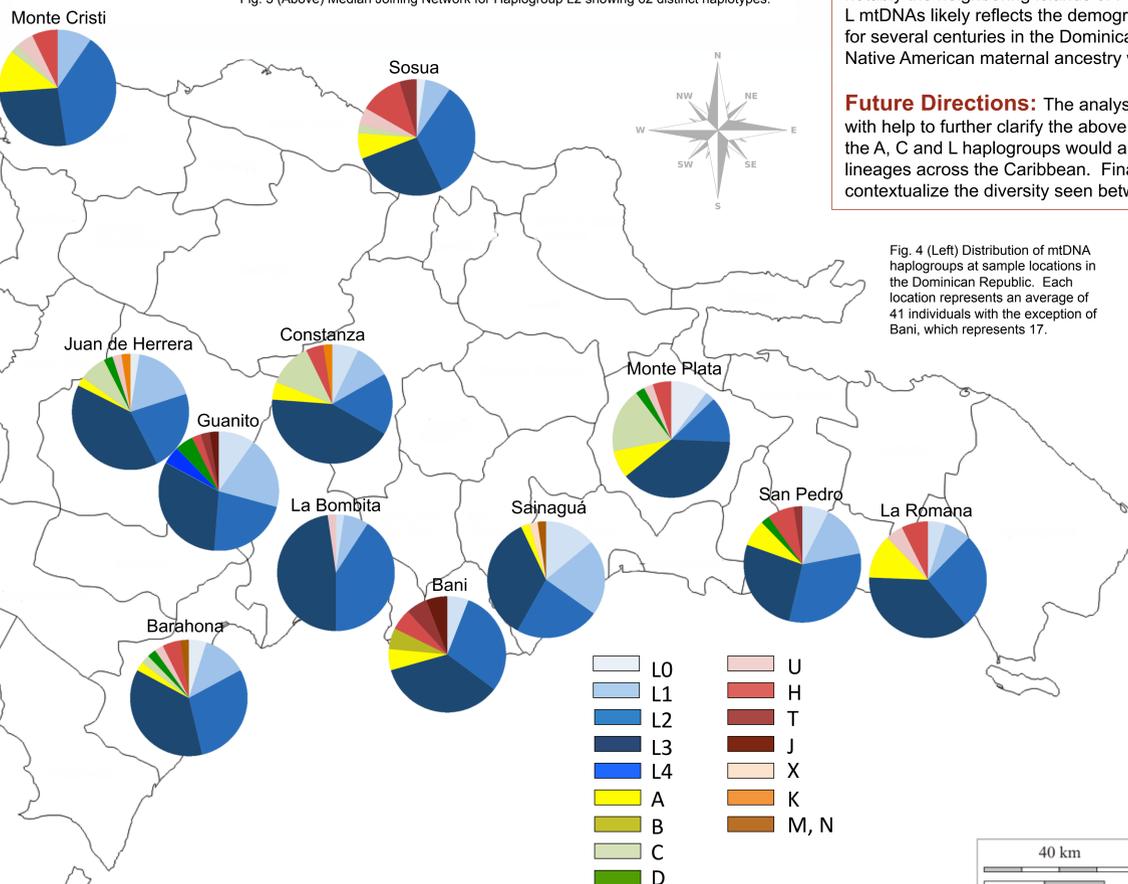


Fig. 4 (Left) Distribution of mtDNA haplogroups at sample locations in the Dominican Republic. Each location represents an average of 41 individuals with the exception of Bani, which represents 17.

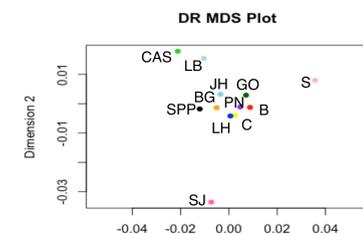
Results Continued

Genetic Distance Analysis

The F_{ST} plot in Figure 5 demonstrates the genetic distance between sample locations in the Dominican Republic based on mtDNA haplotype sequence data. Monte Cristi, La Bombita, Sainaguá, and Monte Plata individuals do not cluster with the rest of the sample locations, suggesting some genetic differences possibly related to the settlement history of each location. For example, Sainaguá is a small community that holds an annual music festival to celebrate Afro-Caribbean music and cultural identity. This fact suggests that the high frequency of L haplogroups in this neighborhood could be due to the tight-knit nature of the community (African ancestry).

To further explore the diversity across the Dominican Republic, an AMOVA was also performed on the sequence data from the sample locations (Figure 5). The results demonstrate that the observed differences within groups (98.53%) was higher than that between groups (1.47%). This result is statistically significant and does not allow us to reject the null hypothesis that there is no difference among the populations.

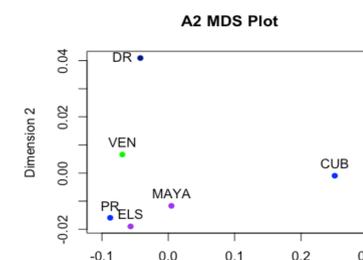
To better understand the genetic affinity between the DR and other populations in the Caribbean in terms of indigenous haplotypes, an MDS plot was generated based on A2 sequence data (Figure 6). Since the Taíno population in the Dominican Republic is thought to have derived from populations from Puerto Rico the results are different than expected. The Dominican Republic sample shares only 3 haplotypes with the Puerto Rican sample and no haplotypes with Cuba. mtDNA haplotyping of the remaining Dominican Republic samples may alter these results. In addition, the AMOVA analysis (Figure 6) demonstrates more variation within groups rather than between groups.



Source of Variation	d.f.	Sum of Squares	Variance Components	Percentage of Variation
Among Populations	11	78.628	0.06740 Va	1.47
Within Populations	457	2066.010	4.52081 Vb	98.53
Total	468	2144.638	4.58821	

Fixation Index: $F_{ST} = 0.01469$
 $P(\text{rand. value} > \text{obs. value}) = 0.00000$
 $P(\text{rand. value} = \text{obs. value}) = 0.00000$
 $P\text{-value} = 0.00000 \pm 0.00000$

Fig. 5 (Above) MDS Plot of F_{ST} values between locations in the Dominican Republic; AMOVA analysis results between sample locations in the Dominican Republic.



Source of Variation	d.f.	Sum of Squares	Variance Components	Percentage of Variation
Among Populations	5	73.616	0.19007 Va	13.75
Within Populations	451	537.808	1.19248 Vb	86.25
Total	456	611.569	1.38255	

Fixation Index: $F_{ST} = 0.13748$
 $P(\text{rand. value} > \text{obs. value}) = 0.00000$
 $P(\text{rand. value} = \text{obs. value}) = 0.00000$
 $P\text{-value} = 0.00000 \pm 0.00000$

Fig. 6 MDS Plot of F_{ST} values between the Dominican Republic and other locations across the Caribbean, South America, and Central America; AMOVA analysis results between the Dominican Republic and the above locations.

Conclusions and Future Directions

Conclusions: Based on the Dominican Republic mtDNA sequence data, it is clear that there is an incredible degree of genetic variation within each sample location. The presence of A2 and C1 haplotypes in each community reflects a pattern seen in other Caribbean islands, most notably the neighboring islands of Puerto Rico and Cuba. In addition, the diversity of haplogroup L mtDNAs likely reflects the demographic impact of the trans-Atlantic slave trade, which occurred for several centuries in the Dominican Republic. Overall, the pattern of African, European and Native American maternal ancestry was consistent across all locations.

Future Directions: The analysis of the remaining 538 individual mtDNA haplogroup data with help to further clarify the above observations. Further, a detailed and geographical study of the A, C and L haplogroups would allow us to better understand the timing and history of these lineages across the Caribbean. Finally, the addition of historical data should help to contextualize the diversity seen between sample locations in the Dominican Republic.

Methods

Sample Collection: From 2015-16, a total of 1,010 individuals were sampled from 25 locations across the DR. Buccal cell samples were taken using mouthwash after obtaining informed consent. DNA extraction and characterization took place in the Lab of Molecular Anthropology at the University of Pennsylvania using the Qiagen QIAmp DNA Mini Kits, following the manufacturer's protocol.

Sample Sequencing and Characterization: For each sample, the entire control region of the mitochondrial DNA (mtDNA) was sequenced, including hypervariable segments I and II (Zhadanov et al., 2010; Gaieski et al., 2011). Each sample was run on an ABI 3130XL Gene Analyzer, and read using the Sequencer 5.0 software tool (Gene Codes Corporation). Sequences were then compared to the revised Cambridge Reference Sequence to identify variant sites (Anderson et al., 1981; Andrews et al., 1999). Sequence data were assigned to haplotypes in consultation with Phylotree Build 17 and Haplogrep 2.0 (van Oven and Kayser, 2009; Weissensteiner, Pacher, Kloss-Brandstätter et al. 2016).

Statistical Analysis: Phylogenetic trees were generated using the Network 5.0 software tool and DNA aligner (Bandelt et al. 1999). Sites were weighed relative to their mutability, following Bandelt et al. (2002). F_{ST} genetic distance estimates were obtained using Arlequin ver. 3.1.2 (Excoffier et al., 2005). The pairwise F_{ST} matrix was then imported into RStudio to generate MDS plots. Comparative haplogroup frequency data were taken from the following groups: Cubans (Mendizabal et al., 2008); Colombians (Bryc et al., 2010); Trinidadians (Benn Torres et al., 2015); Vincentians (Benn Torres et al., 2015); Venezuelans (Lander et al., 2008); Northern Brazilians (Alvez-Silva et al., 2000); Puerto Ricans (Vilar et al. 2014); Salvadorians (Salas et al., 2009), and Yucatec Mayans (González-Martín et al. 2015).