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**PAPER**

**CRIMINALISTICS**

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# Forensic Analysis of mtDNA Haplotypes from Two Rural Communities in Haiti Reflects Their Population History

**ABSTRACT:** Very little genetic data exist on Haitians, an estimated 1.2 million of whom, not including illegal immigrants, reside in the United States. The absence of genetic data on a population of this size reduces the discriminatory power of criminal and missing-person DNA databases in the United States and Caribbean. We present a forensic population study that provides the first genetic data set for Haiti. This study uses hypervariable segment one (HVS-1) mitochondrial DNA (mtDNA) nucleotide sequences from 291 subjects primarily from rural areas of northern and southern Haiti, where admixture would be minimal. Our results showed that the African maternal genetic component of Haitians had slightly higher West-Central African admixture than African-Americans and Dominicans, but considerably less than Afro-Brazilians. These results lay the foundation for further forensic genetics studies in the Haitian population and serve as a model for forensic mtDNA identification of individuals in other isolated or rural communities.

**KEYWORDS:** forensic science, DNA typing, forensic genetics, human mitochondrial DNA, hypervariable region, haplotypes, population genetics, Haiti

Haiti resides on the western third of the island of Hispaniola in the Caribbean Sea. With a population of more than 8 million people, it contains the 4th largest African Diaspora population in the Americas and the largest in the Caribbean. The reasons for choosing Haiti to study African Diaspora genetics are twofold. First, it is composed of a large, predominantly African-derived population and also it has a unique history as the first black independent nation in the Americas. Furthermore, as much of Haiti's population live in isolated rural areas, the challenges and lessons drawn from this study could be applied to forensic studies of other populations.

Before the arrival of Spanish settlers in 1492, the island was populated by various indigenous groups, namely the Ciboneys, Tainos, and Caribs. The Tainos were the most prevalent group when the Europeans arrived. While the Caribs, who were not native to the island, were known to periodically attack Taino villages on the island. Both Tainos and Caribs are thought to be Arawak descendants, but their exact linguistic and genetic relationships to each other are still not clear. Last, the Ciboneys were nomadic people who lived in small communities. Today, most of the indigenous population is thought to be extinct as a result of enslavement, disease, and fighting against European colonists. However, intermixing of indigenous people with Africans and Europeans has left a unique genetic imprint on modern-day people living on the island (1–4).

The Spanish were the first Europeans to settle on the island, which they named Hispaniola. They brought the first wave of Africans to the colony, many of whom had lived in and spoke the languages of Spain and Portugal. As the indigenous population on the island dwindled in size, more enslaved Africans were brought to the colony for mining and domestic work. The origins of this first wave of Africans appear to be from the Gulf of Guinea and Congo region (5–7). By 1520, large numbers of captured Africans were imported into Hispaniola for sugarcane cultivation. This became possible as the Portuguese established trade centers in Elmina Ghana, Mpinda Congo, and a settlement beyond the Cape of Good Hope (8–11). Enslaved people coming directly from Africa were preferred over the previous "westernized" ones who were considered prone to uprising (12).

The second wave of Africans occurred as the French began to dominate the western portion of the island, which they named Saint-Domingue. This wave of enslaved Africans came to the colony from Senegambia, Sierra Leone, Guinea, Dahomey, Nigeria, Congo, Angola, and Mozambique to serve French masters at sugar and coffee plantations (13–15). According to the census reports, in 1789, there were 465,429 enslaved people, 25,000 free blacks/mulattos, and 32,000 whites in Saint-Domingue (16). The real numbers of enslaved people may not have been reported because of the head taxes. In this regard, the elderly, invalids, and children who were not able to work often were excluded from census tabulations. The Haitian historian Thomas Madiou estimates the number of enslaved people at 709,642 (17). A good portion of the enslaved people in Saint-Domingue was sequestered in the north, where the majority of sugar plantations were located. There were

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also multiple maroon colonies made up of escaped slaves who settled in the mountains. On the other hand, the southern portion of the colony was less densely populated. It also had higher numbers of free mulattos who, like the free blacks, were able to buy land and enslaved people.

By the end of the 18th century, the Haitian revolution had begun and France lost control of its colony. This important event marked both the end of slavery and the importation of enslaved Africans into Haiti. At the time of the Haitian revolution, more than 50% of the blacks in Haiti were African-born, with the highest numbers coming from Central Africa and the second highest from Bight of Benin (18,19). This was probably a consequence of the horrific conditions in which enslaved Africans were forced to work, where many passed away without bearing offspring.

The large number of African-born people in Haiti at the time of independence meant that a good deal African culture was retained, such as voodoo practices, which nowadays are nearly identical to their African counterparts. French Creole, the language of Haiti today, is a fusion of French with influences from Wolof, Fon, and Ewe languages from Africa (20,21). After the independence, Haiti welcomed a small number of immigrants, including Polish soldiers, some African-Americans, Germans, French, and other Europeans (22). Later, after 1880, some Syrians and other Middle-Easterners also came to Haiti as merchants (23). Until the present time, many people have emigrated out of Haiti for various reasons, but very few besides those mentioned above have immigrated there. This makes Haiti unique in that its population today is mostly a reflection of those African populations who were brought to the colony just prior to Haitian independence.

The transatlantic slave trade has made it difficult for those in the African Diaspora to trace the exact genealogies (paternal or maternal) to the regions of Africa from which they emanate. The advent of DNA technology has raised the possibility of determining genetic origin from an ancestral population using haploid loci such as Y chromosome or mitochondrial DNA (mtDNA). The advantages of these loci are their lack of recombination, predictable linear inheritance pattern (directly paternal in the Y and directly maternal in mtDNA), and polymorphic display across world populations. In particular, many studies have demonstrated that continental origin can be readily inferred by classification into known mtDNA haplogroups (24–26). In Africa, these haplogroups appear to be differentially distributed across the continent (27–37). Some recent mtDNA studies have shown differential regional African admixture in African Diaspora from North America, Central/Caribbean America, and South America, with the first two showing higher west African percentages and the latter higher West-Central African (26,38,39). In these studies, most of the Central/Caribbean populations were small and were not from populations that are of predominantly African descent (i.e., Mexicans, Caribs, Dominicans).

Here, we present hypervariable segment one (HVS-1) mtDNA sequences from 291 individuals from the nation of Haiti. This is the first DNA study of the Haitian population and their ancestry. The objective of the study is to determine whether the patterns of mtDNA sequences correlate with known historical data. More specifically, do those mtDNA patterns reflect the African regional admixture expected from historical records? Is there substructuring of the Haitian population based on geography? Are there non-African mtDNA lineages, such as Native American or European, in the predominately black Haitian population and to what extent? Also, studying mtDNA lineages in Haiti may help fill in the gaps in the phylogenetics of African mtDNA lineages. According to Curtin (18), the largest proportion of slave shipments from Africa to Saint-Domingue came from Angola (45%) and Bight of Benin (28%). As many regions in Africa such as the Gold Coast, Bight of Benin, and Congo have not been sufficiently sampled, we may find some of these mtDNA lineages in Haiti, where historically many of Haiti's inhabitant's lineages originate. Last, what relevance does this study have for future human identification efforts in Haiti or other challenging rural areas?

## Materials and Methods

### *Sampling*

Buccal swabs were collected in Haiti from 291 individuals who self-identified as "black." These collections took place in the rural mountains of Leogane, Jeremie, and Cap Haitien. At these collection sites, samples were collected at local church meetings where individuals volunteered DNA for the purpose of understanding their genealogical origins. A map of our collection sites in Haiti is shown in Fig. 1. All individual participants were asked to identify the town in which their last known direct maternal ancestor was born. In most cases, individuals only knew their mother or maternal grandmother as their last known direct maternal ancestor. It was assumed that all individuals volunteering samples were unrelated. However, because of hardships in these areas, where mortality and orphans are common, many people were not certain of their exact genealogy in relation to others. All individuals gave proper consent, and research performed was within the guidelines of the institutionally approved International Review Board consent forms (University of Massachusetts Lowell).

### *PCR Amplification and mtDNA Sequencing*

Total buccal cell DNA was extracted from the swabs using the BuccalAmp™ (Epicentre, Madison, WI) DNA Extraction kit according to the manufacturer's specifications. Amplifications of HVS-1 mtDNA control region from each subject were performed using the Platinum *Taq* Polymerase kit (Invitrogen, Carlsbad, CA) with primer pairs L15926 (5′-TCAAAGCTTACACCAGTCTTGTAAACC-3′) and H16498 (5′-CCTGAAGTAGGAACCAGATG-3′). The mtDNA haplotypes of all subjects were determined by analyzing the nucleotide sequence of the HVS-1. Cycle sequencing was performed using BigDye Terminator chemistry (Applied Biosystems, Foster City, CA) in an ABI PRISM 3730 DNA sequencer in both L and H directions. SEQUENCHER 4.7 Forensic Edition

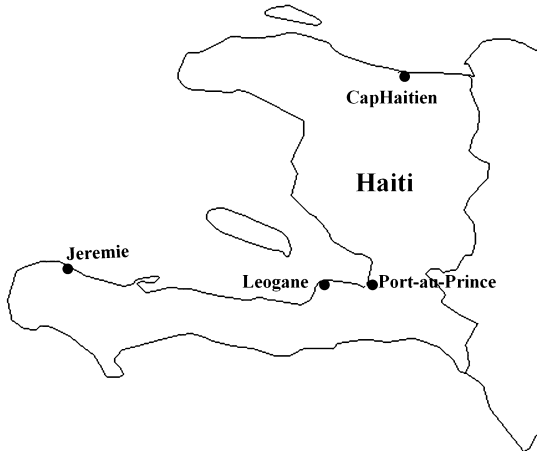


FIG. 1—*Map of Haiti. Map of Haiti showing locations sampled in this study.*

software (Gene Codes Corp, Ann Arbor, MI) was used to edit HVS-1 DNA sequences from both directions and align with the Cambridge Reference Sequence [CRS (40,41); GenBank accession number J01415] to ensure the identification of unambiguous polymorphisms. Transitions in HVS-1 are indicated in all figures and tables by the CRS position minus 16,000. Transversions contain the suffix of the variant base. From the observed polymorphisms, haplogroup assignments for each sample were made based on nomenclature in Salas et al. (37) and revisions in both Kivisild et al. (33) and Gonzalez et al. (31).

### *Data Analysis*

Molecular diversity indices were performed using Arlequin 3.1 software package (42). Calculation of sequence diversities and standard errors were made according to the study of Nei (43). Pairwise $F_{ST}$ distance calculations were made with 1000 permutations and significance level of 0.05. Analysis of molecular variance (AMOVA) calculations were applied to populations based on geography and were made with 1000 permutations using the Arlequin 3.1 software package (42). For all these calculations, mtDNA nucleotide positions 16,000–16,400 were considered. Length variations, such as the cytosine stretch region of HVS-1 (16,182–16,185), were not taken into account. When performing genetic comparisons with other published data, a shorter range was used to accommodate those data sets. The following African mtDNA data were used for comparisons: a total of 1661 from West Africa that include 240 from Senegal (44–46), 204 from Mali (31,47), 372 from Guinea-Bissau (48), 393 from Sierra Leone (32,49), 160 from Nigeria/Niger (46,50), and 292 from Cabo Verde (51); a total of 1132 from West-Central Africa includes 673 from Cameroon (29,30,52), 74 from Central African Republic (46,50,53), 153 from Sao Tome (54,55), 55 from Equatorial Guinea (54,56), 23 from Democratic Republic of Congo (39,46,50), 110 from Cabinda (27), and 44 from Angola (36); a total of 659 from East Africa include 65 from Sudan (57), 289 from Ethiopia/Eritrea (33,58), 161 from Kenya (28,46), 27 from Somalia (46), and 117 from Tanzania (34,50); a total of 416 from Southeast Africa mostly from Mozambique (35,37). Also, published data from African Diaspora included 1065 African-Americans (59), 73 Gullah-speaking African-Americans (47), 168 Dominicans (3,4), eight Mexicans (60), 25 Belize (61), 41 Choco from Columbia, 37 Garifuna from Panama/Belize (62), and 298 Brazilians (24,39,63). Many of these populations were then pooled together into larger groups based on nationality or geographic affiliation. Principal component (PC) analysis based on relative mtDNA haplogroup frequencies of each population was performed using POPSTR software (http://harpend.dsl.xmission.com/popstr/). Maternal admixture in the Haitian population and African Diaspora was estimated using the LEADMIX program (64). Estimates of the admixture coefficient and standard deviations were calculated using the Roberts and Hiorns method (65), the Bertorelle and Excoffier method (66), and the Wang (64) method for admixture estimation. These methods will be referred to hereon as RH, BE, and W, respectively.

## Results

### *HVS-1 Molecular Diversity*

A total of 291 Haitian HVS-1 mtDNA sequences are summarized in Table 1. Figure 1 is a map indicating the primary locations in Haiti where samples were collected. Haitian individuals were categorized by the birthplace of their mother or maternal grandmother. Except for one sample, all individuals from the north had a last known maternal ancestor from Cap Haitien. In the south, 166 were from Leogane, 25 were from Jeremie, and nine were from other southern towns (Port-au-Prince, Jacmel, Les Cayes).

One hundred and two different haplotypes were obtained belonging to 32 mtDNA-derived haplogroups based on HVS-1 polymorphism motifs (Table 2). Although most haplogroup assignments were obvious based on their HVS-1 polymorphism motifs, 45 samples were confirmed by typing HVS-2 polymorphisms. In these cases, the HVS-2 polymorphisms confirmed the haplogroup assignment (data not shown). Most of these haplogroups are well documented in sub-Saharan African populations (37), the exception being a single European mtDNA haplogroup I. The most frequent haplogroups are L2a1, L1b1, and L2c2 found in 22%, 7%, and 8% of the individuals, respectively. It is interesting to note that no Native American haplogroups were observed in the sample (i.e., A, B, C, D, or X). Therefore, more than 99% of the sampled individuals had a sub-Saharan African–derived maternal lineage.

The overall HVS-1 mtDNA sequence diversity values for northern Haiti and southern Haiti were similar, 0.962 and 0.969, respectively (Table 2), even though the northern Haitian subgroup had a slightly higher percentage of different sequences (northern Haiti $K/n = 46.2\%$, southern Haiti $K/n = 32.5\%$) compared with the southern Haitian subgroup (Table 2). The average number of pairwise differences ($M$), percentage of segregating sites ($S/L$), and distributions of pairwise differences based on raggedness index (northern Haiti $r = 0.0145$, southern Haiti $r = 0.0074$) (67) are also similar between the northern and southern subgroups (Table 2).

### *Genetic Differentiation in Haiti*

There appears to be little haplotype sharing between the northern and southern Haitian subgroups, as only five different haplotypes are shared. AMOVA revealed that 3.27% of the variance is attributed to differences in Haitian geography (Table 3). Pairwise $F_{ST}$ ($F_{ST} = 0.033$, $p = 0.000$) based on haplotypes revealed significant differences between northern and southern Haiti (Table 3). Given the high diversity of mtDNA haplotypes, differentiation was also assessed at the haplogroup level. Nineteen of the 32 Haitian mtDNA haplogroups are shared between the northern and southern Haitian subgroups. Of the 13 haplogroups not shared, 10 were only in the southern Haitian subgroup. When $F_{ST}$ pairwise differences were analyzed based on 17 generalized haplogroups found in the Haitian sample (L0a, L1b, L1c1, L1c2, L1c3, L2a, L2a1, L2b, L2c, L3b, L3d, L3e1, L3e2, L3e3, L3f, L4g, I), they continued to show significant differences between northern and southern Haiti ($F_{ST} = 0.022$, $p = 0.001$).

### *Comparisons with Other African and African-Derived Populations*

A database of HVS-1 sequences from published data on African and African Diaspora mtDNAs was assembled. HVS-1 sequences that were found to contain potentially spurious polymorphisms were excluded from the database. Forty-two of the 99 different African-derived haplotypes in the Haitian data set had no matches in the assembled database. When we compared the Haitian haplotypes with only continental African mtDNAs in the database, four more haplotypes (total 46 of 99) did not have a match. The distribution of these haplotypes without a match did not appear specific to any particular haplogroups.

TABLE 1—*Haitian HVS-1 mtDNA sequences.*

| Ht* | Hg† | Differences in HVS-1‡ | North§ | South¶ | Total |
|---|---|---|---|---|---|
| Ht1 | L0a1 | 126 129 148 168 172 187 188G 189 223 230 311 320 | | 1 | 1 |
| Ht2 | L0a1 | 129 148 168 172 187 188G 189 223 230 311 320 | 1 | | 1 |
| Ht3 | L0a1a | 093 129 148 168 172 187 188G 189 223 230 278 293 311 320 | 1 | | 1 |
| Ht4 | L0a1a | 129 148 168 172 187 188G 189 223 230 256 278 293 311 320 | 1 | | 1 |
| Ht5 | L1b | 126 172 187 189 223 264 270 278 311 368 | 2 | | 2 |
| Ht6 | L1b | 126 187 189 223 264 270 274 278 311 | 12 | | 12 |
| Ht7 | L1b | 126 187 189 223 270 278 311 | | 4 | 4 |
| Ht8 | L1b1 | 093 126 187 189 223 264 270 278 293 311 | | 1 | 1 |
| Ht9 | L1b1 | 111 126 187 189 223 239 270 278 293 311 | | 1 | 1 |
| Ht10 | L1b1 | 126 166 187 189 193 223 264 270 278 293 311 | | 1 | 1 |
| Ht11 | L1b1 | 126 187 189 223 239 264 270 278 293 311 | | 7 | 7 |
| Ht12 | L1b1 | 126 187 189 223 239 264 278 293 311 | | 2 | 2 |
| Ht13 | L1b1 | 126 187 189 223 264 270 278 293 311 | 4 | 3 | 7 |
| Ht14 | L1c | 086 129 154 187 189 223 241 278 294 311 360 | 1 | | 1 |
| Ht15 | L1c | 102 129 184 187 189 223 278 294 301 311 360 | | 1 | 1 |
| Ht16 | L1c | 129 184 187 189 223 278 294 301 311 360 | 1 | | 1 |
| Ht17 | L1c | 129 187 189 223 261 278 311 360 | | 2 | 2 |
| Ht18 | L1c | 129 187 189 223 278 294 311 360 | | 1 | 1 |
| Ht19 | L1c1 | 038 093 129 187 189 223 271 278 293 294 311 360 | | 6 | 6 |
| Ht20 | L1c1 | 086 129 163 187 189 223 278 293 294 311 360 | | 1 | 1 |
| Ht21 | L1c1 | 093 129 187 189 223 263 278 293 294 311 360 368 | | 1 | 1 |
| Ht22 | L1c1 | 129 187 189 223 278 292 293 294 311 360 | | 3 | 3 |
| Ht23 | L1c1a | 066 129 189 223 274 278 293 294 311 360 | | 1 | 1 |
| Ht24 | L1c1a | 129 172 184 189 223 261 274 278 290 293 311 360 | | 1 | 1 |
| Ht25 | L1c1a | 129 187 189 223 274 278 292 293 294 311 360 | 1 | | 1 |
| Ht26 | L1c1a | 129 187 189 223 274 278 293 294 311 360 | | 1 | 1 |
| Ht27 | L1c1a1 | 051 129 187 189 214 234 249 258 274 278 293 294 311 360 | | 4 | 4 |
| Ht28 | L1c1a1 | 129 187 189 214 234 249 278 293 294 311 360 | | 1 | 1 |
| Ht29 | L1c1a1 | 129 187 189 214 234 278 293 294 311 360 368 | | 1 | 1 |
| Ht30 | L1c2 | 33.1TCTCTGTTCTTTCAT 129 187 189 223 265C 286G 294 311 360 | 1 | | 1 |
| Ht31 | L1c2 | 071 129 145 187 189 213 223 234 265C 278 286G 294 311 360 | 5 | | 5 |
| Ht32 | L1c2 | 129 187 189 223 265C 278 286A 292 294 311 360 | | 1 | 1 |
| Ht33 | L1c3a1 | 129 183C 189 215 223 278 294 311 327 355 360 390 | | 1 | 1 |
| Ht34 | L1c3a1 | 129 183C 189 215 223 278 294 311 360 | 5 | | 5 |
| Ht35 | L1c3b1 | 017 129 163 187 189 223 278 293 294 311 360 | | 1 | 1 |
| Ht36 | L2a | 051 114A 189 192 223 278 294 362 390 | | 4 | 4 |
| Ht37 | L2a | 189 192 223 278 294 362 390 | 1 | | 1 |
| Ht38 | L2a | 189 223 234 249 278 294 295 390 | 1 | | 1 |
| Ht39 | L2a | 223 234 249 278 294 295 390 | 1 | | 1 |
| Ht40 | L2a | 223 266 278 294 390 | 1 | | 1 |
| Ht41 | L2a1 | 037 129 223 278 294 309 390 | 2 | | 2 |
| Ht42 | L2a1 | 086 223 278 294 309 390 | | 7 | 7 |
| Ht43 | L2a1 | 124 189 192 223 278 294 309 390 | 3 | | 3 |
| Ht44 | L2a1 | 126 189 192 223 245 278 294 309 390 | 2 | | 2 |
| Ht45 | L2a1 | 126 192 223 255 278 309 390 | | 18 | 18 |
| Ht46 | L2a1 | 183C 189 223 224 278 294 309 390 | | 13 | 13 |
| Ht47 | L2a1 | 183C 189 223 269 278 294 309 390 | | 4 | 4 |
| Ht48 | L2a1 | 185 213 223 278 294 309 390 | | 1 | 1 |
| Ht49 | L2a1 | 189 192 223 278 294 309 352 390 | | 4 | 4 |
| Ht50 | L2a1 | 189 192 223 278 294 309 390 | 1 | 1 | 2 |
| Ht51 | L2a1 | 223 278 294 309 390 | 6 | 3 | 9 |
| Ht52 | L2a1a | 223 278 286 294 309 390 | 1 | | 1 |
| Ht53 | L2b | 114A 129 212 213 223 278 390 | 6 | | 6 |
| Ht54 | L2b | 114A 129 213 223 278 354 390 | | 5 | 5 |
| Ht55 | L2b | 114A 129 213 223 278 390 | | 6 | 6 |
| Ht56 | L2b1 | 114A 129 213 223 278 355 362 390 | | 5 | 5 |
| Ht57 | L2c | 223 278 390 | 2 | | 2 |
| Ht58 | L2c1 | 223 278 318 390 | | 3 | 3 |
| Ht59 | L2c2 | 223 264 278 311 390 | | 1 | 1 |
| Ht60 | L2c2 | 223 264 278 390 | | 1 | 1 |
| Ht61 | L2c2 | 264 278 390 | | 20 | 20 |
| Ht62 | L3b | 048 124 223 278 362 390 | | 5 | 5 |
| Ht63 | L3b | 124 189 223 278 362 | 1 | | 1 |
| Ht64 | L3d | 086 124 223 | | 1 | 1 |
| Ht65 | L3d1 | 124 223 319 | 7 | | 7 |
| Ht66 | L3d2 | 124 194T 223 256 324 368 | | 3 | 3 |
| Ht67 | L3d2 | 124 223 256 | 1 | | 1 |
| Ht68 | L3e1 | 086 223 327 | | 1 | 1 |
| Ht69 | L3e1 | 093 176 223 295 327 | 2 | | 2 |
| Ht70 | L3e1 | 176 223 327 | | 1 | 1 |
| Ht71 | L3e1 | 179 223 327 | 1 | | 1 |

*Continued.*

TABLE 1—*Continued.*

| Ht* | Hg† | Differences in HVS-1‡ | North§ | South¶ | Total |
|---|---|---|---|---|---|
| Ht72 | L3e1 | 183C 189 223 260 327 | 2 | | 2 |
| Ht73 | L3e1 | 207 223 327 | 1 | | 1 |
| Ht74 | L3e1 | 223 327 | | 3 | 3 |
| Ht75 | L3e1a | 185 223 311 327 | | 1 | 1 |
| Ht76 | L3e2 | 072 223 311 320 | | 5 | 5 |
| Ht77 | L3e2 | 124 223 320 | | 1 | 1 |
| Ht78 | L3e2 | 179 182C 183C 189 223 239 311 320 362 | 2 | | 2 |
| Ht79 | L3e2 | 223 258T 278 320 | | 3 | 3 |
| Ht80 | L3e2 | 223 320 | 1 | | 1 |
| Ht81 | L3e2 | 223 320 399 | 1 | 5 | 6 |
| Ht82 | L3e2b | 051 172 183C 189 223 320 | | 1 | 1 |
| Ht83 | L3e2b | 172 182C 183C 189 223 320 | 3 | | 3 |
| Ht84 | L3e2b | 172 183C 189 223 320 | 1 | 1 | 2 |
| Ht85 | L3e2b | 172 183C 189 223 250 301 320 | 1 | | 1 |
| Ht86 | L3e2b | 172 183C 189 223 292 320 | | 5 | 5 |
| Ht87 | L3e2b | 172 183C 187.1T 189 223 290 320 | 1 | | 1 |
| Ht88 | L3e2b | 172 189 223 320 | | 1 | 1 |
| Ht89 | L3e3 | 093 148 223 265T | | 1 | 1 |
| Ht90 | L3f | 172 209 223 263 311 368 | | 2 | 2 |
| Ht91 | L3f | 209 223 241 311 | | 3 | 3 |
| Ht92 | L3f | 209 223 295 311 | | 1 | 1 |
| Ht93 | L3f | 209 223 311 | 1 | | 1 |
| Ht94 | L3f1 | 051 209 223 292 295 311 362 | 1 | | 1 |
| Ht95 | L3f1 | 093 129 209 223 292 295 311 320 | | 1 | 1 |
| Ht96 | L3f1 | 129 209 223 292 311 | | 1 | 1 |
| Ht97 | L3f1 | 209 223 292 295 311 362 | | 5 | 5 |
| Ht98 | L3f1 | 209 223 292 311 | | 4 | 4 |
| Ht99 | L4g | 051 114 189 192 223 293T 311 316 355 362 399 | | 1 | 1 |
| Ht100 | L1c? | 086 129 148 172 184 187 189 223 243 261 278 311 355 360 398 | 1 | | 1 |
| Ht101 | I | 129 223 362 391 | 1 | | 1 |
| Ht102 | I | 129 223 391 | | 1 | 1 |
| | | | 91 | 200 | 291 |

*Ht are haplotypes.
†Hg are haplogroups.
‡All mtDNA sequences minus 16,000.
§North = 90 Cap Haitien and one other.
¶South = 166 Leogane, 25 Jeremie, and nine others.

TABLE 2—*HVS-1 sequence diversity in Haiti.*

| Population | *n* | *K* (*K*/*n*) | *S* (*S*/*L*) | *H* (SE) | *M* |
|---|---|---|---|---|---|
| North Haiti | 91 | 42 (46.2) | 63 (15.8) | 0.962 (0.009) | 8.93 |
| South Haiti | 200 | 65 (32.5) | 74 (18.5) | 0.969 (0.005) | 8.51 |
| Total Haiti | 291 | 102 (35.1) | 88 (22.0) | 0.981 (0.002) | 8.73 |

*n*, sample size; *K*, number of different sequences (haplotypes) and percentage of sample size in brackets; *S*, number of segregating sites and percentage of all sites in brackets (*L* = sequence length); *H*, sequence diversity; SE, standard error; *M*, average number of pairwise differences.

TABLE 3—*Analysis of molecular variance (AMOVA) Among Haitian samples.*

| Source of Variation | d.f. | Sum of Squares | Variance Components | Percentage of Variation |
|---|---|---|---|---|
| Among populations | 1 | 2.530 | 0.01636 Va | 3.27 |
| Within populations | 289 | 139.656 | 0.48324 Vb | 96.73 |
| Totals | 290 | 142.186 | 0.49960 | |

Fixation Index $F_{ST}$: 0.03275.
Va and $F_{ST}$: P(random value ≥ observed value) = 0.00000 + −0.00000.

The haplotypes that did have matches are summarized in Table 4. Thirteen different haplotypes are pan-African or have a wide distribution across the continent. These haplotypes are uninformative in regard to intracontinental or regional origin of those maternal lineages. Two belong to European haplogroup I and may reflect a female European founder to those maternal lineages in Haiti. Both northern and southern Haiti each share one of these related, but different haplogroup I lineages. The other matches were grouped as West Africa, West-Central Africa/Southeastern Africa, or East Africa. West-Central and Southeastern Africa were grouped together because many of the haplotypes had matches to both regions. This is probably a reflection of Bantu migrations that carried lineages from Cameroon/Nigeria to more southern regions such as Angola and Mozambique (27,35,36). West-Central/Southeastern Africa followed by West Africa showed the highest number of region-specific matches, while east Africa was negligible. It is also noted that the ratio of West-Central African matches to West African matches is higher in northern than in southern Haiti. (Table 4, Fig. 2).

TABLE 4—*Distribution of Haitian mtDNA haplotype matches.*

| Population | WA | WC/SE | EA | EU | Pan-Af | No Match |
|---|---|---|---|---|---|---|
| Northern | 4 (10) | 10 (20) | 1 (1) | 1 (1) | 11 (27) | 15 (32) |
| Southern | 10 (31) | 14 (33) | 0 | 1 (1) | 8 (19) | 32 (116) |
| Total | 14 (41) | 23 (53) | 1 (1) | 2 (2) | 15 (46) | 47 (148) |

Boxes contain number of unique haplotypes; numbers in parentheses represent number of haplotypes out of the total. WA, West Africa; WC/SE, West-Central and Southeast Africa; EA, East Africa; EU, European; Pan-Af, Pan-African.

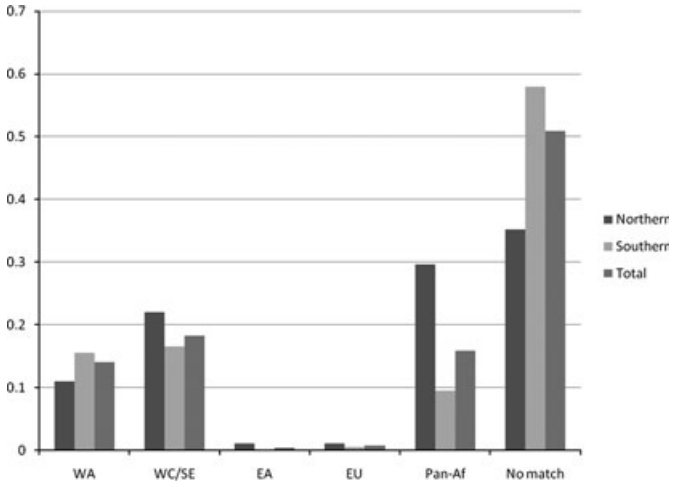


FIG. 2—*Percentage of Haitians having exact matches to particular ancestral geographic regions. WA, West Africa; WC/SE, West-Central/Southeast Africa; Pan-Af, Pan African; EA, East African; EU, European. Bars represent percentage of respective populations having HVS-1 matches to regions in Africa.*

### *Estimates of Maternal Admixture*

To ascertain the African intracontinental admixture found within the Haitian population, all the HVS-1 sequences for African and African Diaspora populations were put into 42 haplogroups. The frequency of each haplogroup in each population was calculated, and their distributions were compared between populations (Table 5). Haplogroup I was excluded in this analysis because it is not found in sub-Saharan Africa and it represented a negligible percentage of the total Haitian population. The PC plot using the haplogroup distributions is shown in Fig. 3. Thirty-five percent of the variance is attributable to PC1, and it separates Mozambique from the rest of the populations. The closest to Mozambique are the West-Central populations and then the African Diaspora, and the most distant are the West African populations. PC2 accounts for 16% of the variation and separates West African and West-Central African populations, with Diaspora, except for Dominicans, clustering near West African populations.

The distributions of haplogroups were also used to estimate maternal African admixture coefficients for the African Diaspora populations. For this analysis, only the West African and West-Central African populations were used as parental populations. The maternal West African admixture estimates using three different methods, and 95% confidence intervals (CI) are listed for each African Diaspora population in Table 6. The highest maternal West African admixture appears to be in the Dominican population and the lowest in Brazilians. The rest of the African Diaspora, African-Americans, and Haiti had values intermediate of the Dominican and Brazil. South Haitians have a slightly higher maternal West African admixture than north Haitians, although in all three methods the values are not significantly different.

## Discussion

In this study, we present genetic data on Haitians living in rural areas in the northern and southern Haiti. These populations were chosen because they were more likely to have remained in these isolated areas over many generations and unlikely to have interacted extensively with other ethnic groups. Also because of Haiti's unique history, it was possible that there could be some genetic differentiation between northern and southern Haitians because of the timing and African ethnic makeup of the different waves of importation. The historical data would suggest that northern Haitians are more derived from the second wave of African enslaved people who were mostly from West-Central Africa, while southern Haitians contain a lesser component from that second wave of African enslaved people. The reason is because most of the population lived in the north and that is where majority of Africans were brought. Also, many of the enslaved Africans did not live long enough to propagate naturally; so much of the first wave's genetic imprint has been lost. However, those who were free blacks or mulattos tended to live in the south and were more likely to propagate naturally and continue their maternal lineages containing genetic imprints from the first wave. This is why the southern Haitians are more likely to retain genetic imprints from the first wave of enslaved Africans who were mostly from upper West Africa. This can also be seen in the African-derived maternal lineages from the Dominican Republic which display elevated West African maternal ancestry. These lineages are probably indicative of the first wave of Africans that the Spanish brought to the island of Hispaniola. Once the French took control of the western portion of the island, which became Haiti, more enslaved people from West-Central Africa were brought over, slightly changing the ethnic and genetic makeup of the population. Just before the revolution, the number of enslaved people imported from Central Africa peaked at 65% (18). Our data based on unique haplotypes and haplogroup distributions between northern and southern Haitians do support the idea that genetic differentiation exists between their maternal lineages. This means that the genetic data are in line with what would be derived from historical data.

Not only do the Haitian genetic data suggest genetic differentiation between north and south Haitians, they also support the predicted historical African intracontinental origins of the maternal lineages in those regions as well. Previous studies on African Diaspora have demonstrated that African intracontinental admixture estimates can be determined, which give values close to what would be expected based on historical data (26,38,39). When looking at the haplotypic level, the ratios of West African to West-Central African mtDNA exact matches was higher in southern Haitians than in northern Haitians (Table 4, Fig. 2), and at the haplogroup distribution level, the estimate of West African maternal ancestry was also higher in southern Haitians (Table 6). On the whole, the total Haitian sample shows a West African maternal ancestry estimate similar to the African-American sample, less than the Dominican Republic sample, but more than Brazil. This is also agrees with what is indicated by historical data.

We also observed only a small number of maternal lineages of non-African origin in this study. This small group consisted of two individuals, both having different lineages belonging to the European haplogroup I. The small number of European-derived maternal lineages is supported by historical data, reflecting that very few European female immigrants came to Haiti after the revolution. It is also possible that a person of mixed ancestry moved to Haiti from neighboring Dominican Republic or another nearby island. It is important to note that neither these two individuals nor anyone else in this study were aware of any non-black ancestors. Moreover, absent were indigenous maternal lineages reflective of the native population of Hispaniola. This observation is also supported by the historical data that suggest the indigenous population began dissolving after European colonization. However, there may still be a small genetic imprint in Haitians today, which was not discovered in this study. Further study in areas that are historically known to have preserved indigenous ancestry, such as in the southern townships Bainet and Petit Groave, may yet reveal this genetic imprint.

TABLE 5—*African and African Diaspora haplogroup frequencies for comparative analysis.*

| Hg | Haiti (T) | Haiti (N) | Haiti (S) | DomRep | U.S.A. | Gullah | Sen | Mali | GB | SL | NiN | WA |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| L0a* | 0 | 0 | 0 | 0 | 3 | 0 | 0 | 1 | 0 | 0 | 0 | 1 |
| L0a1 | 2 | 1 | 1 | 0 | 20 | 2 | 2 | 2 | 19 | 9 | 2 | 34 |
| L0a1a | 2 | 2 | 0 | 1 | 14 | 0 | 0 | 0 | 0 | 0 | 1 | 1 |
| L0a2 | 0 | 0 | 0 | 6 | 4 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| L0d/k/f | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| L1b | 37 | 18 | 19 | 7 | 104 | 6 | 45 | 26 | 38 | 40 | 22 | 171 |
| L1c* | 6 | 2 | 4 | 3 | 29 | 1 | 3 | 0 | 3 | 5 | 1 | 11 |
| L1c1 | 11 | 0 | 11 | 5 | 32 | 0 | 0 | 0 | 0 | 9 | 1 | 18 |
| L1c1a | 10 | 1 | 9 | 1 | 4 | 1 | 0 | 1 | 2 | 0 | 0 | 3 |
| L1c2 | 7 | 6 | 1 | 1 | 44 | 3 | 0 | 0 | 0 | 0 | 0 | 0 |
| L1c3a | 6 | 5 | 1 | 2 | 16 | 0 | 5 | 3 | 5 | 0 | 1 | 14 |
| L1c3b | 1 | 0 | 1 | 0 | 1 | 0 | 1 | 2 | 2 | 2 | 1 | 8 |
| L5 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| L2a | 8 | 4 | 4 | 2 | 41 | 3 | 11 | 7 | 15 | 26 | 10 | 69 |
| L2a1 | 66 | 15 | 51 | 28 | 176 | 4 | 28 | 39 | 46 | 26 | 36 | 175 |
| L2b | 17 | 6 | 11 | 5 | 22 | 4 | 6 | 3 | 8 | 3 | 0 | 20 |
| L2b1 | 5 | 0 | 5 | 5 | 32 | 0 | 13 | 10 | 21 | 3 | 1 | 48 |
| L2c | 2 | 2 | 0 | 18 | 49 | 7 | 38 | 19 | 52 | 25 | 5 | 139 |
| L2c1 | 3 | 0 | 3 | 0 | 9 | 0 | 8 | 2 | 3 | 2 | 1 | 16 |
| L2c2 | 22 | 0 | 22 | 6 | 13 | 1 | 4 | 10 | 6 | 8 | 0 | 28 |
| L2d1 | 0 | 0 | 0 | 1 | 5 | 1 | 0 | 2 | 4 | 3 | 1 | 10 |
| L2d2 | 0 | 0 | 0 | 6 | 7 | 1 | 4 | 2 | 3 | 9 | 1 | 19 |
| L3* | 1 | 1 | 0 | 0 | 0 | 0 | 2 | 1 | 0 | 4 | 2 | 9 |
| L3b | 6 | 1 | 5 | 16 | 68 | 4 | 13 | 17 | 18 | 27 | 18 | 93 |
| L3b1 | 0 | 0 | 0 | 2 | 16 | 0 | 10 | 10 | 14 | 5 | 7 | 46 |
| L3b2 | 0 | 0 | 0 | 3 | 9 | 2 | 0 | 0 | 0 | 0 | 1 | 1 |
| L3d/d2 | 5 | 1 | 4 | 14 | 52 | 3 | 18 | 12 | 28 | 25 | 8 | 91 |
| L3d1 | 7 | 7 | 0 | 0 | 11 | 0 | 0 | 0 | 5 | 0 | 3 | 8 |
| L3d3 | 0 | 0 | 0 | 4 | 6 | 0 | 2 | 1 | 2 | 0 | 0 | 5 |
| L3e1* | 11 | 6 | 5 | 3 | 19 | 5 | 0 | 1 | 0 | 2 | 2 | 5 |
| L3e1a | 1 | 0 | 1 | 1 | 33 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| L3e1b | 0 | 0 | 0 | 0 | 4 | 0 | 1 | 0 | 0 | 0 | 0 | 1 |
| L3e2 | 18 | 4 | 14 | 3 | 38 | 5 | 3 | 11 | 10 | 3 | 7 | 34 |
| L3e2b | 14 | 6 | 8 | 13 | 65 | 9 | 1 | 9 | 6 | 11 | 8 | 35 |
| L3e3 | 1 | 0 | 1 | 2 | 27 | 4 | 1 | 2 | 0 | 0 | 2 | 5 |
| L3e4 | 0 | 0 | 0 | 4 | 9 | 1 | 6 | 1 | 11 | 5 | 0 | 23 |
| L3f | 7 | 1 | 6 | 1 | 3 | 1 | 0 | 1 | 0 | 0 | 2 | 3 |
| L3f1 | 12 | 1 | 11 | 2 | 50 | 5 | 4 | 6 | 9 | 13 | 8 | 40 |
| L4g | 1 | 0 | 1 | 1 | 4 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| L3h | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 13 | 2 | 1 | 16 |
| M | 0 | 0 | 0 | 0 | 11 | 0 | 1 | 0 | 4 | 0 | 0 | 5 |
| U6/U5 | 0 | 0 | 0 | 2 | 15 | 0 | 5 | 2 | 18 | 4 | 6 | 35 |
| | 289 | 90 | 199 | 168 | 1065 | 73 | 235 | 203 | 372 | 271 | 159 | 1240 |

| Hg | Cam | Angola | Cabinda | WCA | MZ | EGuinea | BZ |
|---|---|---|---|---|---|---|---|
| L0a* | 14 | 0 | 0 | 14 | 8 | 0 | 0 |
| L0a1 | 10 | 2 | 4 | 17 | 6 | 1 | 15 |
| L0a1a | 12 | 1 | 7 | 20 | 34 | 0 | 9 |
| L0a2 | 12 | 3 | 3 | 22 | 55 | 4 | 6 |
| L0d/k/f | 0 | 0 | 0 | 0 | 21 | 0 | 1 |
| L1b | 29 | 2 | 3 | 36 | 4 | 2 | 17 |
| L1c* | 2 | 0 | 3 | 6 | 1 | 1 | 4 |
| L1c1 | 3 | 2 | 4 | 9 | 2 | 0 | 15 |
| L1c1a | 15 | 0 | 3 | 18 | 3 | 0 | 2 |
| L1c2 | 21 | 4 | 13 | 39 | 8 | 1 | 23 |
| L1c3a | 8 | 1 | 1 | 20 | 5 | 10 | 4 |
| L1c3b | 5 | 0 | 3 | 9 | 3 | 1 | 1 |
| L5 | 2 | 1 | 0 | 3 | 2 | 0 | 0 |
| L2a | 42 | 2 | 2 | 46 | 9 | 0 | 8 |
| L2a1 | 60 | 9 | 3 | 80 | 131 | 8 | 50 |
| L2b | 6 | 2 | 1 | 10 | 6 | 1 | 15 |
| L2b1 | 8 | 0 | 6 | 14 | 0 | 0 | 4 |
| L2c | 11 | 0 | 0 | 11 | 0 | 0 | 1 |
| L2c1 | 1 | 0 | 0 | 1 | 0 | 0 | 0 |
| L2c2 | 2 | 0 | 2 | 4 | 3 | 0 | 5 |
| L2d1 | 8 | 0 | 0 | 15 | 3 | 7 | 0 |
| L2d2 | 4 | 0 | 0 | 4 | 0 | 0 | 1 |
| L3* | 21 | 0 | 2 | 24 | 1 | 1 | 2 |
| L3b | 26 | 1 | 3 | 30 | 7 | 0 | 6 |
| L3b1 | 7 | 0 | 0 | 7 | 0 | 0 | 3 |
| L3b2 | 10 | 0 | 1 | 11 | 5 | 0 | 3 |

*Continued.*

TABLE 5—*Continued.*

| Hg | Cam | Angola | Cabinda | WCA | MZ | EGuinea | BZ |
|---|---|---|---|---|---|---|---|
| L3d/d2 | 23 | 0 | 2 | 25 | 8 | 0 | 11 |
| L3d1 | 13 | 1 | 1 | 15 | 14 | 0 | 4 |
| L3d3 | 4 | 1 | 3 | 8 | 1 | 0 | 1 |
| L3e1* | 11 | 3 | 5 | 22 | 14 | 3 | 14 |
| L3e1a | 7 | 2 | 4 | 14 | 16 | 1 | 12 |
| L3e1b | 8 | 0 | 4 | 12 | 13 | 0 | 1 |
| L3e2 | 26 | 0 | 0 | 35 | 4 | 9 | 6 |
| L3e2b | 27 | 1 | 8 | 37 | 4 | 1 | 16 |
| L3e3 | 8 | 3 | 2 | 14 | 14 | 1 | 13 |
| L3e4 | 7 | 0 | 1 | 8 | 1 | 0 | 0 |
| L3f | 9 | 1 | 8 | 21 | 7 | 3 | 5 |
| L3f1 | 14 | 0 | 6 | 20 | 3 | 0 | 9 |
| L4g | 15 | 2 | 1 | 18 | 0 | 0 | 7 |
| L3h | 3 | 0 | 0 | 3 | 0 | 0 | 0 |
| M | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| U6/U5 | 7 | 0 | 0 | 7 | 0 | 0 | 4 |
| | 521 | 44 | 109 | 729 | 416 | 55 | 298 |

Hg, haplogroups; Haiti (T), Total Haiti; Haiti (N), North Haiti; Haiti (S), South Haiti; DomRep, Dominican Republic; U.S.A., African-Americans; Gullah, Gullah-speaking African-Americans; Sen, Senegal; Mali, Mali; GB, Guinea-Bissau; SL, Sierra Leone; NiN, Niger/Nigeria; WA, West Africa; Cam, Cameroon; WCA, West-Central Africa; MZ, Mozambique; EGuinea, Equatorial Guinea; BZ, Brazil.

*Underived haplogroups.

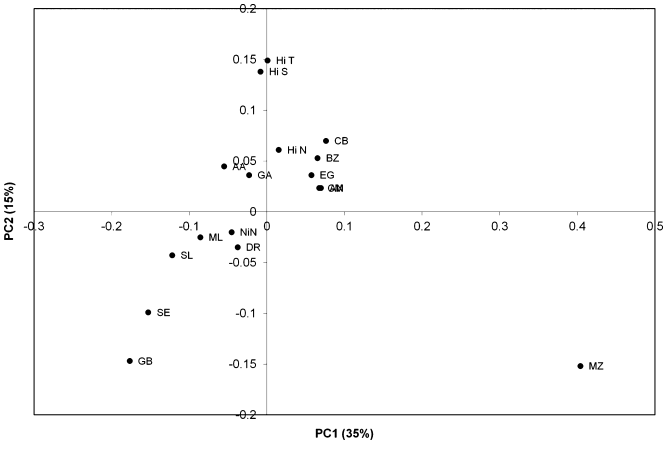


FIG. 3—*Principal component (PC) graph of Haiti, other African Diaspora, and African populations. PC graph of the first two PCs based on haplogroup frequencies of African Diaspora and African populations. West Africa are GB (Guinea-Bissau), SE (Senegal), SL (Sierra Leone), ML (Mali), NIN (Nigeria/Niger). West-Central Africa are CM (Cameroon), CB (Cabinda), AN (Angola), EG (Equatorial Guinea). Southeast Africa is only represented by MZ (Mozambique). Diaspora are HiT (Total Haiti), HiS (South Haiti), HiN (North Haiti), AA (African-Americans), BZ (Afro-Brazil), DR (Dominicans).*

TABLE 6—*Estimates of West African maternal admixture in Haitians and Other African Diaspora populations.*

| Population | RH (95% CI) | BE (95% CI) | W (95% CI) |
|---|---|---|---|
| Total Haiti | 0.45 (0.07, 0.97) | 0.70 (−2.63, 3.38) | 0.38 (0.00, 0.71) |
| North Haiti | 0.43 (−0.18, 1.05) | 0.69 (−3.00, 4.18) | 0.16 (0.00, 0.59) |
| South Haiti | 0.46 (0.10, 1.10) | 0.71 (−3.32, 4.22) | 0.25 (0.00, 0.76) |
| Dominican Republic | 0.67 (0.31, 1.04) | 0.67 (−1.76, 3.16) | 0.66 (0.40, 0.86) |
| Afro-Brazil | 0.07 (−0.09, 0.39) | 0.15 (−1.97, 2.34) | 0.00 (0.00, 0.23) |
| African-American | 0.50 (0.38, 0.71) | 0.61 (−0.88, 1.83) | 0.48 (0.28, 0.68) |

West African and West-Central African populations were used as parental populations. RH is Roberts and Hiorns (65), BE is Bertorelle and Excoffier (66), and W is Wang (64) method for admixture estimation.

Also, further studies in Cazale may reveal more European ancestry where the descendants of Polish soldiers who served in the French army still live today.

Another finding in this study is the high number of unique mtDNA sequences found in our Haitian sample (51%), which had no match to any previously published public mtDNA HVS-1 haplotypes. This is consistent with the findings of Ely et al. (47), which showed that a large number of African Diaspora HVS-1 sequences do not have matches to current public African mtDNA data. This is quite problematic for tracing the origin of a specific mtDNA lineage. An explanation for the high percentage of no matches could be due to the relatively few samples from areas that may have contributed greatly to the Haitian populations such as Bight of Benin and Congo region. However, these mtDNA sequences may allow us to fill in the gaps in the phylogenetics of African mtDNA lineages.

The diversity found in this study is likely reflective of isolated rural populations found in Haiti. However, it cannot be ruled out that high frequencies of certain nonshared haplotypes (i.e., Ht6, Ht45, Ht46, and Ht61) could be the result of inadvertent sampling of maternally related individuals. Even if this is true, it is not likely to alter the conclusions because three of the four haplotypes belong to fairly common haplogroups L1b or L2a1. These haplogroups would be expected to have relatively high frequencies anyway. The urban areas probably contain much more diversity as they are destinations for economic or academic opportunities. Further studies in urban areas might provide useful mtDNA haplotypic diversity that would be very useful for resolving African mtDNA phylogenetics. Other studies have found extreme founder effects islands in the Caribbean (62), and it remains to be seen whether these effects will be observed in Haiti as well.

The study of Haitian people has important implications toward understanding of the African Diaspora because it demonstrates that historical data may be corroborated with genetic data. This is important because many historical documents relating to the African Diaspora, such as shipping records, may be limited, may be missing or may contain errors. Haiti is the 4th largest African Diaspora population in the Americas and contains a wealth of African genetic variation, which could help reconstruct the origins of the African Diaspora.

Of importance to forensic DNA applications, this study presents the first mtDNA data set for Haiti. These samples were collected

3 years before the massive earthquake that devastated the nation. In the aftermath of the earthquake, given the unique challenges in Haiti, forensic DNA identification of victims would have been difficult. It is hoped that this study may offer some insights into the Haitian population's genetic structure and a reference mtDNA database from which future comparisons can be made. This study would also be useful as a reference database for other forensic laboratories in other parts of the Caribbean as well.

The high number of shared lineages is another problem when dealing with isolated, rural populations. While we demonstrated that many maternal lineages were shared among individuals within a location, it is reasonable to expect that the Y chromosome paternal lineages would be similar or less diverse. It is also likely that the paternal non-African genetic component of the Haitian population will be higher than the maternal non-African contribution. In addition to Y chromosome, nonhaploid markers (autosomes plus X chromosome) might be more useful when necessary to discern individuals from rural areas especially when genealogy is not well known. Future studies looking at nonhaploid markers across these populations could reveal more insights into the Haitian population structure.

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