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Paternal genetic structure of the Dominican Republic: Y-STR haplotype diversity and comparison with Caribbean populations

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Research Article

Keywords: Y-STR, PowerPlex Y23, Dominican Republic, haplotype diversity, forensic genetics, Caribbean populations

Posted Date: November 26th, 2026

DOI: https://doi.org/10.21203/rs.3.rs-11668542/v1

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Additional Declarations: No competing interests reported.

Abstract

This study presents the first comprehensive Y-STR haplotype dataset for the Dominican Republic generated using the PowerPlex® Y23 System (PPY23). A total of 412 unrelated Dominican males were analyzed, yielding 392 distinct haplotypes, including 373 singletons (90.53%), with a discrimination capacity (DC) of 0.9515, haplotype diversity (HD) of 0.99975, and haplotype match probability (HMP) of 0.00268. DYS385a/b showed the highest gene diversity (GD = 0.9019), followed by DYS481 (GD = 0.8137), DYS570 (GD = 0.8117), and DYS576 (GD = 0.8052). Interpopulation comparisons with published datasets from Puerto Rico (n = 116), Jamaica (n = 140), and Haiti (n = 122) were performed using 15 shared single-copy Y-STR loci to ensure comparability across populations. AMOVA attributed 7.66% of the total paternal genetic variation to differences among populations (global RST = 0.0766, P < 0.001). Pairwise comparisons revealed no significant differentiation between the Dominican Republic and Puerto Rico (RST = 0.0004, P = 0.332), whereas significant differentiation was observed between the Dominican Republic and Jamaica (RST = 0.0820, P < 0.001) and Haiti (RST = 0.1244, P < 0.001). These findings highlight the high discriminatory potential of PPY23 in the Dominican population and underscore the importance of population-specific Y-STR reference databases for forensic applications in the Caribbean.

1 Introduction

The analysis of Y-chromosome short tandem repeats (Y-STRs) is a well-established tool in forensic genetics and in the reconstruction of paternal lineages [1, 2]. Because Y-STRs reside in the non-recombining portion of the Y chromosome, the complete haplotype is transmitted as a single paternal unit from father to son. This linkage limits individualization capacity relative to autosomal STR profiling, since an entire Y-STR profile is treated as one event under the product rule, but it makes Y-STRs particularly valuable for resolving male lineage contributions in male-female DNA mixtures, for paternity testing in deficiency cases, and for tracing paternal ancestry in population studies [1, 3]. Population-specific reference databases of sufficient size are therefore essential for both forensic and demographic applications; at the global level their coordination and quality control are managed by the Y Chromosome Haplotype Reference Database (YHRD) [4].

The PowerPlex® Y23 System (PPY23; Promega Corporation) extended the previously standard Yfiler® 15-locus panel to 23 Y-STR loci, substantially improving discrimination among individuals who share the same 15-locus haplotype. The panel includes several highly polymorphic markers (notably DYS481, DYS570, DYS576, and DYS643) absent from earlier kits, and an international collaborative study confirmed its utility across diverse global populations [5]. As a consequence, PPY23 has been progressively adopted in forensic laboratories, and population-reference datasets based on this panel are increasingly required for accreditation and casework interpretation in many jurisdictions.

The Caribbean presents a complex paternal genetic landscape shaped by pre-Columbian Amerindian settlement, Iberian colonization from the late fifteenth century, and the forced migration of West and

Central African populations during the transatlantic slave trade [6]. The relative contributions of these ancestral sources differ among islands according to colonial administrative history, settlement intensity, and the scale of plantation economies, so that the Caribbean populations of today are not genetically interchangeable for forensic or demographic purposes. Published Y-STR datasets from the region include studies from Puerto Rico, Jamaica, and Haiti [7, 8], but population-specific databases remain incomplete for most islands.

The Dominican Republic occupies the eastern two-thirds of Hispaniola and has a population of approximately 11 million. Prior Y-chromosome work in the country is limited to a preliminary study based on 12 Y-STR loci [9]; no PPY23-based reference dataset and no systematic Y-SNP haplogroup characterization had previously been published for the Dominican population. The present study was undertaken to fill this gap. It had two objectives: (1) to characterize Y-STR haplotype diversity in a sample of 412 unrelated Dominican males typed with the PPY23 system; and (2) to situate the Dominican paternal pool within the Caribbean context through comparison with available Yfiler-compatible datasets from Puerto Rico, Jamaica, and Haiti.

2 Materials and Methods

2.1 Study population

This study included 412 unrelated Dominican males whose samples were processed between 2020 and 2025 in the Human DNA Identification (HID) Department of REFERENCIA Laboratorio Clínico, Santo Domingo, Dominican Republic. Participants self-reported Dominican ancestry with no known paternal relationship within three generations. All participants provided written informed consent, and the study was approved by the National Bioethics Committee (CONABIOS), Dominican Republic (Ref. 070-2024). Donors were not selected by region of origin; the sampling strategy and its implications for within-country structure are discussed in Section 3.5.

2.2 DNA extraction

Genomic DNA from buccal swabs (n = 341) was extracted using SwabSolution™ (Promega Corporation, Madison, WI, USA) following the manufacturer's protocol. Blood samples collected on Whatman FTA cards (n = 71) were processed directly by adding a 1.2-mm punch to the PCR reaction mixture without prior extraction, as validated for the PPY23 system by the manufacturer's technical documentation.

2.3 Y-STR typing

The PowerPlex® Y23 System (PPY23) was used to co-amplify 23 Y-STR loci: DYS19, DYS385a/b, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS437, DYS438, DYS439, DYS448, DYS456, DYS458, DYS481, DYS533, DYS549, DYS570, DYS576, DYS635, DYS643, and YGATAH4. Amplification followed the manufacturer's recommended conditions on a GeneAmp PCR 9700 thermocycler (Thermo Fisher Scientific). Amplicons were separated by capillary electrophoresis on an ABI 3500 Genetic Analyzer and analyzed with GeneMapper® ID-X v1.5 software.

2.4 Treatment of DYS385a/b

The duplicated locus DYS385a/b was treated as a single genetic marker because PPY23 co-amplifies both copies, preventing their distinction based on fragment length alone. Accordingly, alleles were recorded as ascending ordered pairs (e.g., 11–14), including identical pairs (e.g., 12–12). Genetic diversity was calculated based on allele-pair states rather than individual alleles. A total of 51 distinct allele-pair combinations were identified, and the PPY23 panel was considered to comprise 22 independent units of analysis, consistent with previously published approaches.

2.5 Intrapopulation statistics

Statistical calculations were performed using the STRAF platform v2.2.2, with allele and haplotype frequencies estimated by direct counting. For each of the 22 units of analysis, we report the number of observed alleles or allele-pair states (Nall) and gene diversity (GD) as descriptive measures of variability. The 22 units analyzed here lie within the male-specific, non-recombining region of the Y chromosome and are in complete linkage disequilibrium, transmitted as a single inherited block. Multiplying locus-specific probabilities would substantially overstate discriminating power. Forensic parameters are therefore presented exclusively at the haplotype level, the only unit of inheritance for this marker system.

2.6 Interpopulation analysis

Pairwise genetic distances (RST) and an analysis of molecular variance (AMOVA) were computed in Arlequin v3.5 using the 15 single-copy loci shared across all comparison datasets. The original datasets comprised 17 markers for Jamaica (n = 140) and Haiti (n = 122), 19 markers for Puerto Rico (n = 116), and 22 markers for the Dominican Republic (n = 412). To ensure comparability among populations, analyses were restricted to the 15 loci shared by all datasets (DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS437, DYS438, DYS439, DYS448, DYS456, DYS458, DYS635, and YGATAH4). DYS385a/b was excluded from interpopulation analyses because its two copies cannot be distinguished based on fragment length alone (Section 2.4), consistent with its exclusion by Purps et al. [5] from their global AMOVA and RST analyses. AMOVA and pairwise RST were based on the sum of squared differences in repeat number, accounting for the stepwise mutation model of microsatellites. DYS389II was entered as DYS389II – DYS389I to avoid double-counting the DYS389I repeat block. Significance of variance components and pairwise RST values was assessed using 10,100 permutations, and the 95% confidence interval of the global RST was estimated using 20,000 bootstrap replicates over loci. Relationships among individuals from the four populations were also explored through principal component analysis (PCA) in STRAF v2.2.2.

2.7 Data availability

The deposition of the dataset in the Y Chromosome Haplotype Reference Database (YHRD), normally required for the publication of Y-STR population data [4], was not feasible at the time of submission due to the current restrictions on database functionality. Since the implementation of release 67 (7 February 2022), access to haplotype matching and the associated ancestry inference function has been restricted

as part of precautionary measures concerning the use of lineage markers for biogeographic ancestry inference [10]. No YHRD accession number could therefore be obtained. The complete set of 412 haplotypes (22-unit definition, DYS385a/b combined) is provided as Supplementary Table S1 and is available from the corresponding author on request; the data will be submitted to YHRD as soon as the database resumes accepting new datasets.

3 Results

3.1 Haplotype diversity

A total of 412 Y-chromosome haplotypes from unrelated Dominican males were analyzed. Using the 22-unit definition of the haplotype (DYS385a/b combined; Section 2.4), 392 distinct haplotypes were identified: 373 occurred once (singletons, 90.53% of the sample), 18 were shared by two individuals, and one was shared by three individuals. The discrimination capacity (DC) was 0.9515. Haplotype diversity (HD) was 0.99975 and the haplotype match probability (HMP) was 0.00268 (Table 1). Allele frequencies are provided in Supplementary Table S2.

Table 1
Summary haplotype diversity statistics for 412 Dominican males (PPY23, 22-unit definition)

Parameter	Value
Individuals analyzed (n)	412
Distinct haplotypes	392
Singletons	373 (90.53%)
Shared by 2	18
Shared by 3	1
Discrimination capacity (DC)	0.9515
Haplotype diversity (HD)[a]	0.99975
Haplotype match probability (HMP)[a]	0.00268

[a] *HD and HMP were calculated by direct counting from the raw 412-haplotype dataset (Supplementary Table S1) with DYS385a/b treated as a combined marker.*

3.2 Variability by locus

Table 2 presents Nall and GD for each of the 22 units of analysis, with DYS385a/b reported as a single combined marker (Fig. 2). Under this treatment, DYS385a/b is the most variable unit in the panel (GD = 0.9019, 51 distinct allele-pair states among 412 individuals), followed by DYS481 (GD = 0.8137), DYS570

(GD = 0.8117), and DYS576 (GD = 0.8052). The least variable loci were DYS393 (GD = 0.5339) and DYS389I (GD = 0.4643).

Table 2
Number of alleles or allele-pair states (Nall) and gene diversity (GD) for 22 units of analysis (n = 412)

Locus	Nall / states	GD
DYS385a/b*	51	0.9019
DYS481	13	0.8137
DYS570	12	0.8117
DYS576	9	0.8052
DYS458	12	0.7610
DYS643	8	0.7314
DYS448	6	0.7314
DYS390	7	0.7285
DYS635	8	0.7238
DYS549	8	0.7131
DYS389II	8	0.6973
DYS438	6	0.6956
DYS19	6	0.6852
DYS456	7	0.6703
DYS439	6	0.6114
DYS392	6	0.5872
YGATAH4	4	0.5783
DYS533	5	0.5752
DYS437	4	0.5730
DYS391	6	0.5444
DYS393	4	0.5339
DYS389I	4	0.4643

** DYS385a/b reported as an ordered allele pair (*Section 2.4*); Nall = number of distinct allele-pair states.*

3.3 Interpopulation RST and AMOVA

AMOVA across the four Caribbean samples (n = 790; 15 loci, DYS385a/b excluded) placed the majority of paternal variation within populations: 92.34% of the total variance was found within samples and 7.66% among them (RST = 0.0766; 95% bootstrap interval 0.0321–0.1173; P < 0.001).

Pairwise RST values (Table 3) separate the samples into two pairs. The Dominican Republic and Puerto Rico did not differ significantly (RST = 0.0004; P = 0.332), whereas both differed significantly from Jamaica (Dominican Republic–Jamaica: RST = 0.0820; Puerto Rico–Jamaica: RST = 0.1086; both P < 0.001) and from Haiti (Dominican Republic–Haiti: RST = 0.1244; Puerto Rico–Haiti: RST = 0.1486; both P < 0.001). Jamaica and Haiti did not differ from each other (RST = 0.0038; P = 0.151). Notably, Haiti (which shares Hispaniola with the Dominican Republic) was the population most distant from the Dominican sample in terms of RST (Fig. 3).

At the locus level, the among-population variance component was largest for DYS19 (RST = 0.207), DYS390 (0.173), DYS448 (0.105), DYS389II − I (0.084), and DYS392 (0.084) (all P < 0.001). DYS456, DYS389I, DYS458, DYS439, and DYS438 showed no significant differentiation among populations.

Table 3
Pairwise RST values among four Caribbean populations (15 loci, DYS385a/b excluded, n = 790)

Dominican Republic	**Dominican Republic**	**Puerto Rico**	**Jamaica**	**Haiti**
	—	**0.0004 (ns)**	**0.0820*** **	**0.1244*** **
Puerto Rico		—	0.1086***	0.1486***
Jamaica			—	0.0038 (ns)
Haiti				—

*ns: not significant (P > 0.05); ***: P < 0.001; 10,100 permutations.*

3.4 Principal component analysis

The PCA showed broad overlap among individuals from the Dominican Republic, Puerto Rico, Haiti, and Jamaica (Fig. 1). The PC1–PC2 plane explained 11.99% of the total variation (PC1 = 8.40%, PC2 = 3.59%); the PC1–PC3, PC2–PC3, and PC1–PC4 planes explained 10.80%, 5.99%, and 10.52%, respectively. None of these planes separated the four populations into discrete clusters. The first four principal components together accounted for only 16.51% of the total variation, indicating that most individual variability is distributed across many components, as expected for highly polymorphic Y-STR haplotypes under the prevalent within-population diversity pattern seen in the AMOVA.

3.5 Within-country structure

The regional paternal origin of donors was not recorded systematically, so no AMOVA among Dominican regions was performed and the 412 haplotypes are reported as a single national sample. This is

consistent with the ISFG guidelines for the publication of genetic population data [11, 12], which require unrelated individuals, a documented description of the population and sampling method, and data quality control, but do not mandate regional quotas or proportional geographic representation. Independence was ensured by including a single individual per case file, with kinship established by prior autosomal STR typing, and by excluding donors from different case files who shared both surnames and a province of origin suggestive of common paternal ancestry. The absence of regional data limits inference about substructure within the country; this limitation is addressed in Section 4.

4 Discussion

The DYS385a/b locus was treated as a single ordered-pair marker throughout this study, consistent with the structure of the PPY23 assay and with published recommendations for multicopy Y-STR loci [5, 13]. Under this convention, the panel resolves into 22 independent units of analysis, and locus-specific statistics for the two copies are not computed separately. The resulting dataset shows DYS385a/b as the single most variable unit in the Dominican sample, a result expected given its high mutation rate and duplicated architecture, followed by DYS481, DYS570, and DYS576.

The discrimination capacity of the panel (DC = 0.9515) and the proportion of singleton haplotypes (90.53%) are related but distinct quantities. DC expresses the proportion of the sample resolved into distinct haplotypes overall (392/412), whereas the singleton proportion specifically describes haplotypes observed exactly once (373/412) and is the figure most directly comparable to the "percentage of unique haplotypes" reported by Purps et al. [5] for the global PPY23 study (~ 92.9%). On that corrected basis, the Dominican sample shows a marginally lower singleton proportion than the global PPY23 reference population. This difference is consistent with a population that retains detectable founder effects and moderate lineage sharing, rather than indicating a panel performance problem: at DC = 0.9515 and HD = 0.99975, the system provides forensic discrimination equivalent to that reported for other Caribbean and Latin American PPY23 datasets [5].

The interpopulation analysis places the Dominican paternal pool next to Puerto Rico and apart from Haiti and Jamaica. The near-zero RST between the Dominican Republic and Puerto Rico (RST = 0.0004; P = 0.332) is consistent with a shared colonial history of Spanish settlement and comparable demographic processes, although small comparison sample sizes (n = 116 for Puerto Rico) mean that values close to zero should be interpreted as an absence of detectable differentiation rather than proof of genetic identity. Conversely, Haiti (the geographically closest population, sharing the island of Hispaniola) showed the highest RST with the Dominican Republic (RST = 0.1244; P < 0.001), a result that records the demographic divergence produced by separate colonial administrations and by the very different scale of enslaved African labor mobilized under French rule in what is now Haiti. The loci carrying the largest among-population variance component (DYS19, DYS390, DYS392, DYS448) have modal alleles that differ between the main Y-haplogroups characteristic of western European and sub-Saharan African ancestry, supporting the interpretation that the detected RST structure reflects different relative proportions of

paternal European and African lineages across populations. This reading cannot be confirmed with STR data alone (haplogroup assignment requires Y-SNP typing) and is discussed further under limitations.

For forensic interpretation, these results demonstrate that haplotype frequencies from the nearest Caribbean neighbors cannot serve as a valid substitute for a Dominican-specific database: even the closest population (Puerto Rico) is genetically distinct at some loci, and the RST with Haiti and Jamaica is large enough to introduce systematic error in likelihood ratio calculations. The present dataset provides, for the first time, a PPY23-based population reference for the Dominican Republic that meets the ISFG minimum reporting standards [11, 12] and can be used for haplotype frequency estimation in casework.

4.1 Limitations

Three limitations should be noted. First, Y-SNP haplogroup data were not generated for this sample. Haplogroup assignment would allow the RST structure to be linked directly to continental ancestry proportions and would enable validation of in silico haplogroup prediction tools (including NevGen and Athey's Haplogroup Predictor) in admixed Caribbean populations. Future work should include Y-SNP typing of the major haplogroups expected in this population (R1b, E1b1a, E1b1b, J, I, G, and Q). Second, the regional origin of donors was not recorded systematically, so within-country substructure cannot be evaluated by AMOVA. The sampling met the ISFG independence criteria, and a single national database is the appropriate starting point, but regional differences cannot be excluded given the documented population history of the Dominican Republic. Third, the comparison datasets for Puerto Rico, Jamaica, and Haiti are based on Yfiler-compatible panels and are smaller (116–140 individuals) than the Dominican sample. Larger Caribbean samples genotyped with PPY23 would allow a more statistically robust comparison and might reveal additional population substructure.

5 Conclusions

This study provides the first PPY23 Y-STR reference dataset for the Dominican Republic. With DYS385a/b treated as a single combined marker, 412 haplotypes resolved into 392 distinct haplotypes (DC = 0.9515; 90.53% singletons; HD = 0.99975), and DYS385a/b was the most variable unit of the panel. These values support the use of the PPY23 system in Dominican forensic casework, with match probabilities computed at the haplotype level as recommended for Y-STR markers.

In the Caribbean comparison, the Dominican paternal pool did not differ significantly from that of Puerto Rico and differed significantly from those of Jamaica and Haiti, with 7.66% of paternal variance attributed to among-population differences (RST = 0.0766). Haiti, despite sharing Hispaniola with the Dominican Republic, was the most genetically distant population, reflecting divergent colonial and demographic histories.

As a continuation of this work, we propose Y-SNP typing of the major haplogroups in this sample or a representative subset. That typing would directly characterize the paternal ancestry of the Dominican

population, test whether the RST structure observed here reflects different proportions of European and African Y-lineages, and allow in silico haplogroup predictors to be validated in admixed Caribbean populations. Systematic recording of the regional paternal origin of future donors would additionally allow within-country substructure to be evaluated by AMOVA.

Declarations

Ethics approval and consent to participate

This study was approved by the National Bioethics Committee (CONABIOS), Dominican Republic (Ref. 070-2024). All participants provided written informed consent prior to inclusion.

Consent for publication

Not applicable.

Competing interests

The authors declare no competing interests.

Data availability

The datasets generated and analyzed during the current study are available within the published article and its supplementary materials. The individual Y-STR haplotype electropherograms are provided as supplementary files. Additional information is available from the corresponding author upon reasonable request.

Funding

This research received no specific grant from any funding agency in the public, commercial, or not-for-profit sectors. Sample collection and laboratory analyses were conducted within the routine operations of REFERENCIA Laboratorio Clínico.

Authors' contributions

All authors contributed to the study conception and design. Sample collection, DNA extraction, and PCR amplification were performed by T.R. and R.R. Statistical analysis was performed by T.R. and E.R. The first draft of the manuscript was written by T.R. and E.R., and all authors commented on previous versions of the manuscript. All authors read and approved the final manuscript.

Acknowledgements

The authors thank the staff of REFERENCIA Laboratorio Clínico for their support and all participants for their voluntary contribution to this study.

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Figures

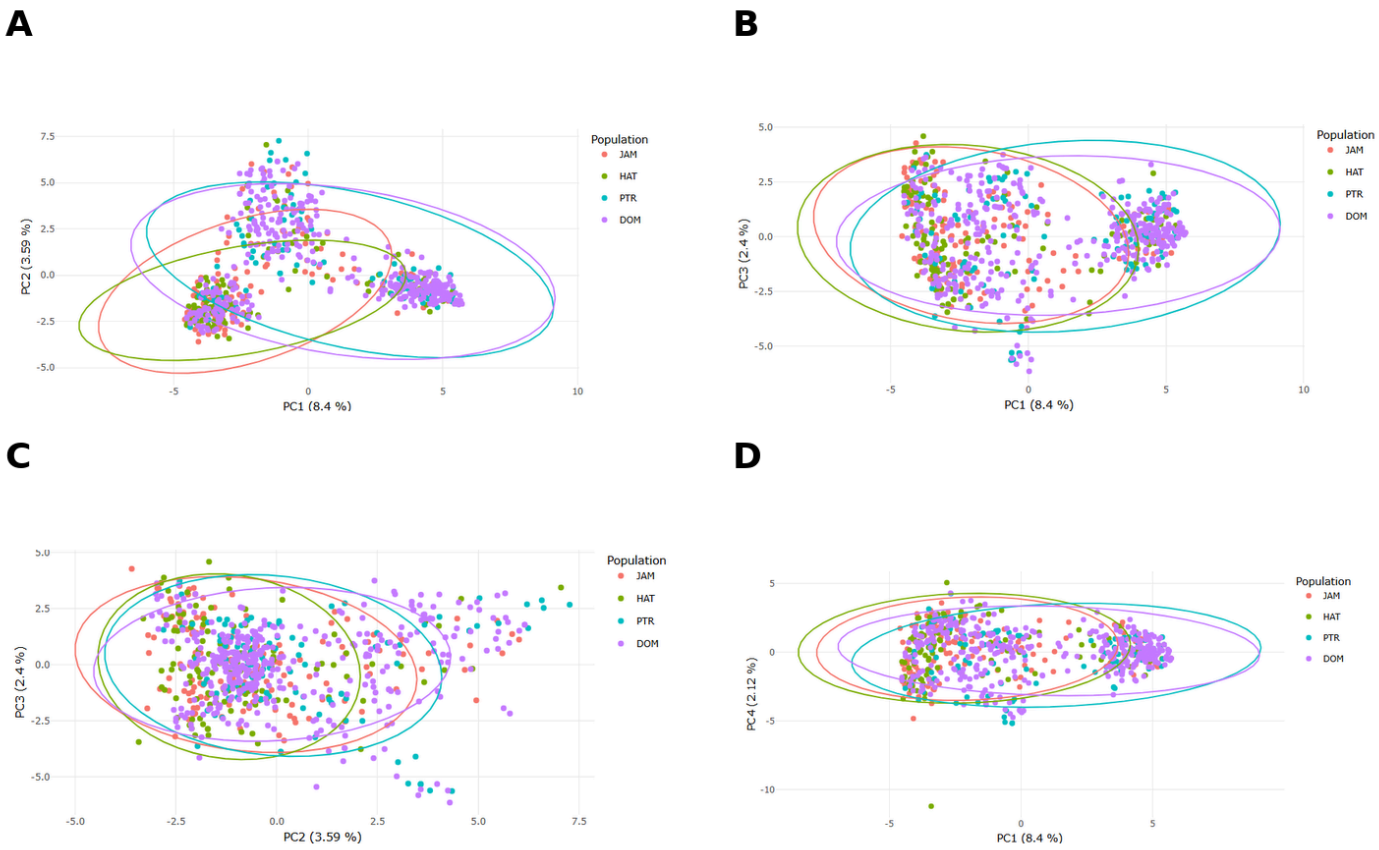

Figure 1

Principal component analysis (PCA) of 790 Y-STR haplotypes from four Caribbean populations: Dominican Republic (DOM, n = 412), Puerto Rico (PTR, n = 116), Jamaica (JAM, n = 140), and Haiti (HAT, n = 122), based on 15 shared single-copy Y-STR loci. Each point represents an individual, colored according to population, and the ellipses indicate the population distributions. (A) PC1 versus PC2; (B) PC1 versus PC3; (C) PC2 versus PC3; and (D) PC1 versus PC4. The percentages in parentheses indicate the proportion of total genetic variation explained by each principal component. The broad overlap among populations reflects the limited genetic differentiation observed in the PCA and is consistent with the AMOVA results, which attributed more than 92% of the total genetic variation to within-population differences.

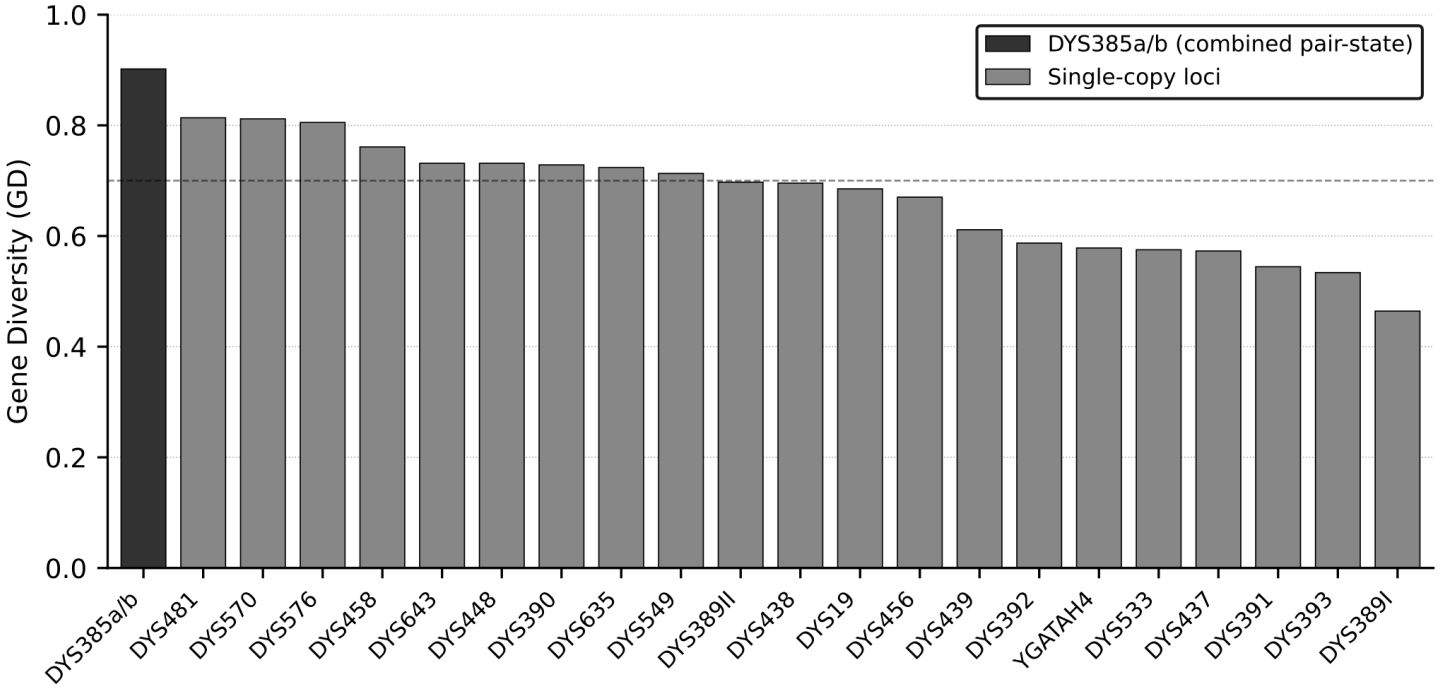

Figure 2

Gene diversity (GD) across the 22 independent units of analysis of the PowerPlex® Y23 panel in the Dominican population (n = 412). The combined DYS385a/b allele-pair states are represented as a single marker (dark bar), while the remaining single-copy Y-STR loci are shown individually (gray bars). Gene diversity was estimated from the observed allele or allele-pair-state frequencies. DYS385a/b exhibited the highest diversity (GD = 0.9019), followed by DYS481 (GD = 0.8137), DYS570 (GD = 0.8117), and DYS576 (GD = 0.8052). The dashed horizontal line indicates GD = 0.70.

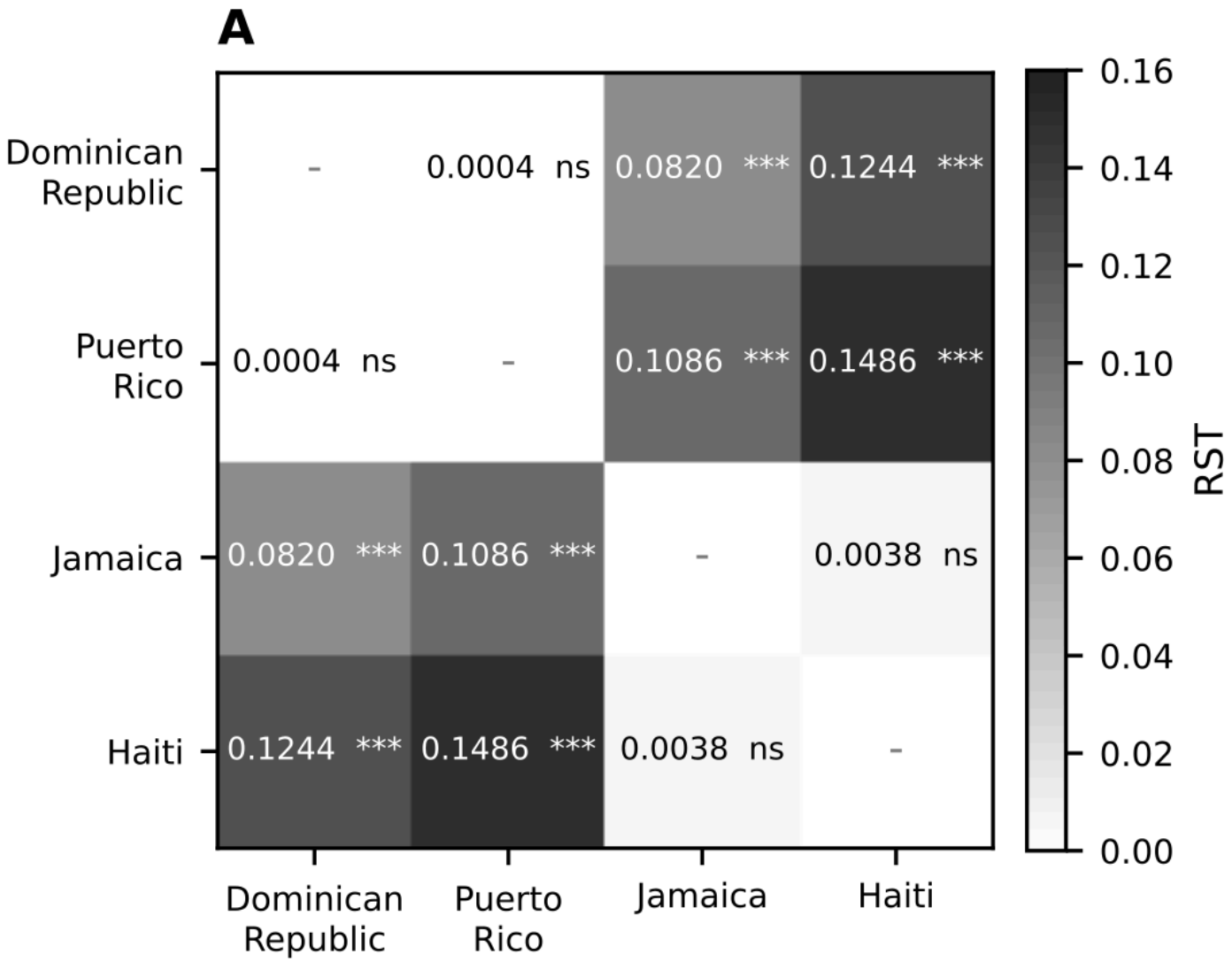

Figure 3

Genetic differentiation and relationships among four Caribbean populations based on Y-STR haplotypes. Heatmap of pairwise RST values among the Dominican Republic (n = 412), Puerto Rico (n = 116), Jamaica (n = 140), and Haiti (n = 122), calculated using 15 shared single-copy Y-STR loci. Darker shading indicates greater genetic differentiation. Pairwise RST values and their statistical significance are shown within the matrix (***P < 0.001; ns, not significant).

Supplementary Files

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- ESM1SupplementaryTableS1.xlsx
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