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Analysis of 27 Y-chromosomal STR loci of the Mestizo Peruvian population

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Abstract

Background Y-chromosome-specific short tandem repeat markers reside on the non-recombinant portion of the Y chromosome and are paternally inherited. These properties make Y-STRs a useful tool in investigations of sexual assault and other violent crimes, paternity cases, genealogical testing, and evolutionary studies. Native populations reside in the Peruvian coast, mountains, and jungle; however, the Mestizos are the largest population group in Peru.

Results Of the 290 samples, 283 distinct haplotypes were observed. Gene diversities for each Y-STR marker ranged from 0.4271 to 0.8597. The haplotype diversity for the total sample was 0.9964.

Conclusions Genetic affinity was identified between the Peruvian Mestizo population and other Peruvian populations (except Jivaro, Awajun, and Shipibo-Conibo populations) and Bolivian Mestizo populations using multidimensional scaling and phylogenetic tree analysis

Keywords Y-STRs, Haplotypes, Mestizo Peruvian population

Background

Y-STR markers are inherited via the paternal lineage, present haploid, and except for the pseudoautosomal regions do not experience recombination. These properties make Y-STRs a useful tool in sexual assault investigations, paternity testing, genealogical testing, and evolutionary studies (Roewer 2009).

The populations of Peru have undergone different migratory processes, contributing to Peruvian population variation and native populations in the coast, mountains, and jungle as well as populations with

substantial genetic admixture. Because of this diversity, studies have been carried out on autosomal STRs markers from different Peruvian populations (Delgado & Neyra 2018; Neyra et al. 2019, 2020; Neyra-Rivera et al. 2020; Neyra-Rivera et al. 2021a, b; Bermejo et al. 2022; Neyra-Rivera et al., 2022) and also on Y-STRs from native populations (Tineo et al. 2015; Neyra-Rivera et al., 2022; Neyra-Rivera et al. 2023) analysis in the case of Y-STRs the genetic diversity (GD, probability that two randomly chosen individuals can be differentiated by a particular genetic marker(s) (Nei 1987)), haplotype diversity (HD, probability that two randomly chosen individuals can be differentiated by a particular set of markers comprising a haplotype(s) (Nei 1987)), probability of a random match (MP, probability that two randomly selected individuals in a population share identical genotypes (Jones 1972)), discrimination capacity (DC, probability that two randomly selected individuals will be genetically differentiated at a particular marker(s) (Smalldon and Moffat 1973)), haploid match probability (HMP, probability that two randomly selected individuals will share the same haplotype), and

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pairwise genetic distance (RST, the correlation of allele sizes (rather than allelic states) between genetic markers sampled within populations (Excoffier 2001)). However, there have not been studies with large population samples (i.e., greater than 200 individuals) and with at most 23 Y-STR markers. Only one study with 83 individuals and 23 Y-STR markers (accession number YA005599) is represented in the YHRD database (Yhrd.org). The present investigation seeks to expand on available genetic data by studying a larger sample set of Mestizos for 27 Y-STR markers and compare these genetic data to assess relationships with populations associated either geographically or historically.

Methods

Samples and experiment

DNA from 290 unrelated individuals were collected in Nucleid Cards (Copan). The individuals reside in different departments of Peru (Amazonas $n=2$, Ancash $n=15$, Apurimac $n=4$, Arequipa $n=15$, Ayacucho

$n=15$, Cajamarca $n=2$, Cusco $n=16$, Huancavelica $n=4$, Huanuco $n=22$, Ica $n=4$, Junín $n=9$, La Libertad $n=2$, Lambayeque $n=1$, Lima $n=123$, Loreto $n=4$, Madre de Dios $n=7$, Moquegua $n=1$, Piura $n=5$, Puno $n=31$, Tacna $n=2$, Tumbes $n=2$, and Ucayali $n=4$) (Fig. 1). All participants freely and voluntarily signed informed consent to participate in this study.

A 1.2 mm punch was taken from each card and amplified by direct PCR using the Yfiler™ Plus PCR Amplification Kit (Life Technologies) following the manufacturer's instructions. Amplified products were detected in the Applied Biosystem™ 3500XL Genetic Analyzer (Life Technologies) following the manufacturer's recommended protocol. After capillary electrophoresis, the data were imported into the GeneMapper® ID-X v1.5 software (Thermo Fisher Scientific 2015) to generate the genetic profiles which were analyzed according to the manufacturer's recommendations. The population data were deposited in the YHRD database (accession number: YA006020).



Fig. 1 Departments of Peru from which the 290 samples were collected. (a) Amazonas $n=2$, (b) Ancash $n=15$, (c) Apurimac $n=4$, (d) Arequipa $n=15$, (e) Ayacucho $n=15$, (f) Cajamarca $n=2$, (g) Cusco $n=16$, (h) Huancavelica $n=4$, (i) Huanuco $n=22$, (j) Ica $n=4$, (k) Junín $n=9$, (l) La Libertad $n=2$, (m) Lambayeque $n=1$, (n) Lima $n=123$, (o) Loreto $n=4$, (p) Madre de Dios $n=7$, (q) Moquegua $n=1$, (r) Piura $n=5$, (s) Puno $n=31$, (t) Tacna $n=2$, (u) Tumbes $n=2$, and (v) Ucayali $n=4$

Statistical analysis

For the calculation of the allelic frequencies for 23 of the 27 STRs, the Power Stats V1.2 software was used (Tereba 1919); for the DYS385a/b and DYF387S1 markers, the "type" frequencies were calculated by the counting method. GD and HD were calculated with the following formulas $GD = (n/n - 1) (1 - \sum P_i^2)$ and $HD = (n/n - 1) (1 - \sum X_i^2)$, where P_i is the frequency of the i th allele, n is the number of samples analyzed, and X_i represents the calculated frequency of the i th haplotype (Sandoval et al., 2013). The MP was calculated as $MP = \sum X_i^2$ and DC was determined by dividing the number of different observed haplotypes with the sum total number of identified haplotypes. HMP was calculated with the formula $HMP = 1 - HD$. AMOVA was used to RST values between Y-STR datasets. The RST values were then used for MDS analysis. RST and p values between different populations were estimated by AMOVA and visualized on a MDS plot (YHRD) (Willuweit & Roewer 2007).

Results

Allelic frequencies and forensic statistical parameter analyses

This study analyzed 27 Y STRs in 290 males collected across different departments in Peru. Results were obtained for all 27 Y-STR loci in all samples, and no null alleles were observed. Allele (or type) frequencies for each locus of the combined data set are shown in Table 1. The DYS437 marker displayed the fewest number of alleles (3 alleles), while the DYS385a/b marker had the highest number of types (45 types). GD values ranged between 0.4271 (DYS437) and 0.9415 (DYS385a/b), and only two Y-STR loci had values less than 0.5 (DYS437

and DYS391). The two highest diversity markers, based on GD, were DYS385a/b (0.9415) and DYF387S1 (0.9363) while the lowest diversity was observed at the DYS437 locus (0.4271).

Haplotype diversity

Since there is no recombination between the Y STRs, haplotype data better display the diversity within the population. There were 283 distinct haplotypes in the total data set ($n=290$) with HD and DC values of 0.9964 and 0.9759, respectively (Table 2). The largest number of the same haplotype was two and seven pairs of two haplotypes were observed (H78 –Puno and Cajamarca-, H105 –Huancavelica and Cuso-, H124 –Lima-, H201 –Lima-, H210 –Lima-, H253 –Madre de Dios and Lima- and H255 –Lima and Cusco-). Ten microvariants were identified and were observed in the markers DYS627 (alleles 18.2, 20.2, and 21.2), DYS448 (alleles 16.2, 17.2, and 18.2), and DYS448 (allele 18.2).

Population structure for forensic analyzes

Using an online tool NevGen Y DNA Predictor (Cetkovic Gentula & Nevski, 2015) provisional haplogroups (most probable and major clade only) for the distinct haplotypes of the Mestizos from Peru were determined. In the Mestizo population, the majority (56.18%) belonged to haplogroup Q, with the rest distributed among haplogroups R (22.61%), J (7.07%), E (6.36%), G (2.83%), T (2.47%), I (2.12%), and L (0.35%) (Table 2).

Genetic distances

The RST values for genetic distances and the MDS plot are shown in Table 3 and Fig. 2 for the

Table 1 Allele frequencies and gene diversities of 27 Y-chromosome STR loci for the Mestizo population from Peru ($n=290$)

	DYS58	DYS389I	DYS65	DYS389II	DYS627	DYS460	DYS458	DYS19	YGAT4B	DYS440	DYS391	DYS456	DYS390	DYS392	DYS518	DYS578	DYS437	DYS449	DYS393	DYS439	DYS481	DYS553	Allele	No.	DYS385a/b	Allele	No.	DYS385b
7	0.069										0.0403				0.0724	0.0504							10, 13	1	0.0014	33, 36	1	0.0014
9		0.070									0.0370				0.0609		0.0069		0.0072	10, 15	1	0.0014	33, 39	2	0.0069			
10		0.0724									0.0276	0.7000			0.1552		0.0310		0.0310	11, 11	1	0.0018	33, 40	2	0.0069			
11		0.1621									0.0341	0.2310			0.1620		0.0312		0.0312	11, 12	4	0.0138	34, 34	1	0.0034			
12	0.0034	0.0034			0.0207	0.0409	0.0374			0.0207		0.0214		0.0100		0.0369	0.0366	0.0369	0.0369	0.0369	0.0369	0.0369	11, 13, 2	1	0.0014	34, 38	5	0.0172
13	0.5828										0.0034	0.2759	0.0724		0.0034	0.2414	0.6532	0.1966	0.0552	11, 13, 2	1	0.0014	34, 36	3	0.0103			
14	0.0034	0.3345				0.0034	0.0372	0.2931	0.0034		0.0135	0.0966		0.0993	0.0552	0.0241	0.7172	0.2138	0.0552	11, 14	45	0.0152	34, 37	3	0.0103			
15	0.0035	0.0034									0.0135	0.0966			0.0103		0.0172	0.7172	0.2138	0.0552	11, 15	5	0.0172	34, 38	10	0.0181		
16	0.1000										0.0069				0.0103		0.0069		0.0069	11, 16	2	0.0069	34, 39	10	0.0181			
16,2											0.0069				0.0069		0.0069		0.0069	12, 12	2	0.0069	34, 41	1	0.0034			
17	0.1799					0.0138		0.2911	0.0103		0.0097			0.0034	0.4317		0.0034		0.0034	12, 13	1	0.0014	34, 42	1	0.0034			
17,12						0.0100					0.0034				0.0034		0.0034		0.0034	12, 14	1	0.0014	34, 35	2	0.0041			
18	0.3207					0.0055	0.1517			0.0345	0.0276			0.1828		0.0034		0.0034	12, 15	2	0.0069	35, 36	38	0.0110				
18,2						0.0054		0.0609		0.0014				0.0034		0.0034		0.0034	12, 16	3	0.0069	35, 37	11	0.0179				
19	0.2452					0.0043	0.0043	0.0043		0.0179			0.1000		0.0038		0.0038	12, 17	1	0.0014	34, 38	3	0.0042					
20	0.0728				0.0276	0.2897	0.0103	0.4793			0.0069			0.0069		0.0276		0.0276	12, 18	2	0.0069	35, 39	43	0.1483				
30,2						0.0034					0.01207		0.0021		0.0054		0.0054		0.0054	13, 13	1	0.0014	35, 40	10	0.0145			
31	0.0110					0.1448	0.0779				0.01207		0.0021		0.0054		0.0054		0.0054	13, 14	1	0.0014	35, 41	2	0.0158			
31,2						0.0034					0.01207		0.0021		0.0054		0.0054		0.0054	13, 15	1	0.0014	35, 42	2	0.0069			
22	0.0034					0.5069	0.1414			0.0172		0.0041		0.0103		0.0172		0.0172	13, 16	9	0.0310	36, 36	19	0.0379				
23						0.2655	0.0855				0.0069		0.0014		0.0103		0.0179		0.0179	13, 17	2	0.0069	36, 37	19	0.0355			
24						0.0044	0.0241				0.0157		0.0034		0.0089		0.0224		0.0224	13, 18	3	0.0014	36, 39	9	0.0110			
25			0.0038			0.0038					0.00793		0.0010		0.0172		0.0172		0.0172	13, 19	2	0.0069	36, 39	9	0.0110			
26											0.0034		0.0010		0.0172		0.0172		0.0172	13, 20	1	0.0014	36, 40	3	0.0041			
27						0.0052					0.0034		0.0010		0.0172		0.0172		0.0172	14, 14	12	0.0014	36, 41	12	0.0069			
28						0.2310					0.0052		0.0010		0.01448		0.01448		0.01448	14, 15	10	0.0045	37, 37	4	0.0138			
29						0.0055					0.0034		0.0010		0.01448		0.01448		0.01448	14, 16	8	0.0026	37, 38	11	0.0179			
30						0.0055					0.0034		0.0010		0.01448		0.01448		0.01448	14, 17	21	0.0224	37, 38	10	0.0185			
31						0.0282					0.0034		0.0010		0.01448		0.01448		0.01448	14, 18	5	0.0117	38, 38	13	0.0165			
32						0.0793					0.0034		0.0010		0.01448		0.01448		0.01448	14, 19	5	0.0117	38, 38	13	0.0165			
33						0.0069					0.0069		0.0010		0.01448		0.01448		0.01448	14, 20	3	0.0110	38, 39	7	0.0244			
34											0.0069		0.0010		0.01448		0.01448		0.01448	14, 21	4	0.0114	38, 40	4	0.0177			
35											0.0069		0.0010		0.01448		0.01448		0.01448	14, 22	12	0.0041	39, 39	3	0.0100			
36											0.0069		0.0010		0.01448		0.01448		0.01448	14, 23	13	0.0041	39, 39	3	0.0100			
37											0.0069		0.0010		0.01448		0.01448		0.01448	14, 24	22	0.0041	39, 40	2	0.0167			
38											0.0069		0.0010		0.01448		0.01448		0.01448	14, 25	14	0.0048	39, 41	1	0.0033			
39											0.0069		0.0010		0.01448		0.01448		0.01448	14, 26	7	0.0110	39, 41	1	0.0033			
40											0.0069		0.0010		0.01448		0.01448		0.01448	14, 27	16	4	0.0114	39, 41	4	0.0177		
41											0.0069		0.0010		0.01448		0.01448		0.01448	14, 28	20	9	0.0045	39, 41	36	0.0163		
42											0.0069		0.0010		0.01448		0.01448		0.01448	14, 29	17	5	0.0117	39, 41	17, 18	0.0163		
43											0.0069		0.0010		0.01448		0.01448		0.01448	14, 30	17	1	0.0033	39, 41	17, 19	2	0.0067	
																			18, 18	1	0.003							
No	11	5	6	6	14	5	11	6	5	7	4	6	3	5	5	10	11	3	13	4	5	11	5					
GD	0.7864	0.9446	0.6512	0.7409	0.8201	0.9412	0.7721	0.5747	0.5632	0.6422	0.4554	0.5915	0.6673	0.6434	0.7462	0.7919	0.7213	0.4271	0.8597	0.5160	0.7083	0.8291	0.5614					
																								49	0.9415	36	0.9363	

Table 2 Haplotypes, haplotype frequencies, and relevant haplotype population data for 27 Y-STR loci in the Mestizo population ($n=290$)*

Note that DYS385a/b and DYF387S1 are presented without assigning to a particular region of these duplication loci Samples size, 290; total No. Obs. haplotype, 283; total No. unique haplotype, 283; haplotype diversity (HD), 0.9964; discriminating capacity (DC), 0.9759; match probability (MP), 0.0036; haplotype match probability (HMP), 0.0036

* Microvariants are bolded

* Haplogroups were provisionally determined using the online tool at <https://site.nevgen.org/nevgen-genealogy-tools-v1-1/>. The most probable haplogroup and only the major clade are reported

Table 3 Pairwise Rst value estimates (below the diagonal) and corresponding p value (above the diagonal) for the Mestizo population from Peru and 14 reference population datasets

Population	Peru [Mestizo] (n = 283)	Bolivia [Boliviano] (n = 218) ^a	Bogotá, Colombia [Mestizo] (n = 184) ^b	Oxapampa, Peru [Ashaninka] (n = 57) ^c	Amazonas, Peru [Awajun] (n = 18) ^d	Puno, Peru [Aymara] (n = 90) ^e	Amazonas, Peru [Aymará] (n = 57) ^f	Cuzco, Peru [Chumbivilica] (n = 10) ^g	Arequipa, Peru [Chuquibamba] (n = 6) ^h	Junín, Peru [Huancayo] (n = 13) ⁱ	Loreto, Peru [Iquito] (n = 85) ^j	Amazonas, Peru [Jívaro] (n = 21) ^k	Lima, Peru [Quechua] (n = 11) ^l	Ucayali, Peru [Shipibo-Conibo] (n = 21) ^m	Spain [Spanish] (n = 2406) ⁿ
Peru [Mestizo] (n = 283)	-	0.1771	0.0000	0.0017	0.0046	0.0000	0.0285	0.0000	0.0005	0.0014	0.0046	0.0551	0.2577	0.0013	0.0000
Bolivia [Boliviano] (n = 218) ^a	0.0017	-	0.0000	0.0017	0.0016	0.0000	0.0101	0.0009	0.0006	0.0017	0.0010	0.0257	0.1251	0.0023	0.0000
Bogotá, Colombia [Mestizo] (n = 184) ^b	0.0889	0.0891	-	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0355	0.0000	0.0000	0.0000
Oxapampa, Peru [Ashaninka] (n = 57) ^c	0.0359	0.0374	0.2072	-	0.0004	0.0005	0.0000	0.0002	0.0010	0.0003	0.0020	0.0068	0.0013	0.0000	0.0000
Amazonas, Peru [Awajun] (n = 18) ^d	0.0776	0.1004	0.2535	0.1055	-	0.0007	0.0000	0.0020	0.0026	0.0078	0.0036	0.0971	0.0039	0.0002	0.0000
Puno, Peru [Aymara] (n = 90) ^e	0.1006	0.0982	0.2932	0.0423	0.0981	-	0.0000	0.0747	0.3081	0.1221	0.0000	0.0008	0.0004	0.0000	0.0000
Amazonas, Peru [Chachapoya] (n = 106) ^f	0.0998	0.0163	0.0587	0.1015	0.1542	0.1606	-	0.0000	0.0000	0.0000	0.0000	0.0014	0.3505	0.0000	0.0000
Cuzco, Peru [Chumbivilica] (n = 10) ^g	0.1976	0.1952	0.3963	0.1917	0.2377	0.0451	0.2871	-	0.8148	0.2576	0.0001	0.0004	0.0020	0.0000	0.0000
Arequipa, Peru [Chuquibamba] (n = 16) ^h	0.1425	0.1424	0.3464	0.1114	0.1644	0.0050	0.2189	-0.0415	-	0.7902	0.0001	0.0007	0.0019	0.0000	0.0000
Junín, Peru [Huancal] (n = 13) ⁱ	0.1273	0.1342	0.3330	0.1282	0.1404	0.0257	0.1975	0.0225	-0.0343	-	0.0005	0.0016	0.0043	0.0000	0.0000
Loreto, Peru [Iquito] (n = 85) ^j	0.0190	0.0306	0.1769	0.0294	0.0683	0.0727	0.0562	0.2143	0.1325	0.1297	-	0.0852	0.0099	0.0000	0.0000
Amazonas, Peru [Jívaro] (n = 21) ^k	0.0274	0.0416	0.1795	0.0537	0.0550	0.0895	0.0863	0.2601	0.1652	0.1725	0.0212	-	0.0323	0.0010	0.0000

Table 3 (continued)

Population	Peru [Mestizo] (n=283)	Bolivia [Boliviano] (n=218)*	Bogotá, Colombia [Mestizo] (n=184) ^b	Oxapampa, Peru [Ashaninka] (n=57) ^c	Amazonas, Peru [Awajun] (n=18) ^d	Puno, Peru [Aymara] (n=90) ^e	Amazonas, Peru [Huancayo] (n=10) ^f	Cuzco, Peru [Chachapoya] (n=10) ^g	Arequipa, Peru [Chiquibambilla] (n=16) ^h	Junin, Peru [Huancan] (n=13) ⁱ	Loreto, Peru [Iquitos] (n=8) ^j	Amazonas, Peru [Ucayali] (n=21) ^k	Amazonas, Peru [Quechua] (n=11) ^l	Lima, Peru [Shibipo-Conibo] (n=21) ^m	Ucayali, Peru [Spanish] (n=2406) ⁿ
Maria Brijon, Antonio Salas, Francesca Brisighelli, Jorge Mario Cárdenas, Alejandro Blanco Verea, Sandra Beleza, Manuel Fondevila, Juan Carlos Jaime, María Victoria Lareu, Jose Lovo-Gomez, Angel Carracedo contributed to 223 Haplotypes on October 4, 2012 Filed under accession number YA003761	0.0104	0.0316	0.0680	0.1548	0.1870	0.1942	0.0033	0.3275	0.2450	0.2043	0.0841	0.0905	-	0.0001	0.0000
Yunis JI., Acevedo LE., Campo DS. and Yunis EI. (2005), "Population data of Y-STR minimal haplotypes in a sample of Caucasian-Mestizo and African descent individuals of Colombia," <i>Forensic Sci Int</i> 151(2-3):307-13	0.0830	0.0837	0.1873	0.1621	0.1900	0.2080	0.1347	0.3928	0.3082	0.3215	0.1011	0.1260	0.2086	-	0.0000
Tineo DH, Loiola S, Paredes PV, Amaya YC, Simão F, Carvalho EF, Gusmão L. Genetic characterization of 27 Y-STR loci in the native population of Ashaninka from Peru. <i>Forensic Sci. Int. Genet. Suppl. Ser.</i> 2015; 5:e220-e222. DOI: doi.org/10.1016/j.fsigs.2015.09.088	0.2377	0.2300	0.0486	0.3685	0.4239	0.4559	0.2005	0.5589	0.5178	0.5058	0.3423	0.3547	0.2351	0.3350	-
Neyra-Rivera CD, Robles CS., Delgado E., Velasquez MRE., Budowle B. Analysis of 27 STR markers of the chromosome Y in the population of the jungle from Peru. <i>Revista Española de Medicina Legal.</i> 2023. https://doi.org/10.1016/j.reml.2022.10.001	Evelyn Guevara, Antti Sajantila contributed 24 Haplotypes on June 19, 2015 Filed under accession number YA004109	Roeper, Lutz et al. Continent-wide decoupling of Y-chromosomal genetic variation from language and geography in native South Americans. <i>PLoS genetics</i> vol. 9(4) (2013):e1003460. https://doi.org/10.1371/journal.pgen.1003460	Neira-Rivera CD, Arenas T., Ramos D., Reinoso V., Rey C. and Budowle B. (2021). 'Population data of 27 Y-chromosome STRs in Aymara population from Peru' Australian Journal of Forensic Sciences 54(5):596-610	María Claudia Fiorella Bartella Cárrillo contributed 16 Haplotypes on March 13, 2013 (for reference see Roeper L., Nothnagel M., Gusmão L., Gomes V., González M., Corach D., Salà A., Alechine E., Palha T., Santos N., Ribeiro-Dos-Santos A., Géppert M., Willuweit S., Baeta M., Núñez C., Martínez-Jarreta B., González-Andrade F., De F., Silva D., Bulles JJ., Turbón D., Parra L., Arroyo-Pardo E., Toscanini U., Borjas L., Barletta C., Stewart E., Santos S. and Krawczak M. (2013), "Continent-wide decoupling of Y-chromosomal genetic variation from language and geography in native South Americans," <i>PLoS Genet</i> 9(4):e1003460	Cláudia Fiorella Bartella Cárrillo contributed 16 Haplotypes on March 13, 2013 (for reference see Roeper L., Nothnagel M., Gusmão L., Gomes V., González M., Corach D., Salà A., Alechine E., Palha T., Santos N., Ribeiro-Dos-Santos A., Géppert M., Willuweit S., Baeta M., Núñez C., Martínez-Jarreta B., González-Andrade F., De F., Silva D., Bulles JJ., Turbón D., Parra L., Arroyo-Pardo E., Toscanini U., Borjas L., Barletta C., Stewart E., Santos S. and Krawczak M. (2013), "Continent-wide decoupling of Y-chromosomal genetic variation from language and geography in native South Americans," <i>PLoS Genet</i> 9(4):e1003460	Rakesh Shresta, Chuan-Chao Wang contributed 11 Haplotypes on May 22, 2014 Filed under accession number YA003949	Martinez-Cadenas C., Blanco-Verea A., Hernando B., George Bj., Brion M., Carracedo A., Salas A. and Capelli C. (2016). "The relationship between surname frequency and Y chromosome variation in Spain," <i>European Journal of Human Genetics</i> 24(1):20-8	Martinez-Cruz B., Mendizabal I., Harmant C., Pablo D., Ioana M., Angelicheva D., Kouvatzi A., Makutuk H., Neetea MG., Pamjav H., Zalán A., Tournef I., Marushiková E., Popov V., Bertranpetti J., Kalaydjieva L., Quintana-Murci Comas D. and Genographic Consortium (2016), "Origins, admixture and founder lineages in European Roma," <i>Eur J Hum Genet</i> 24(6):937-43							

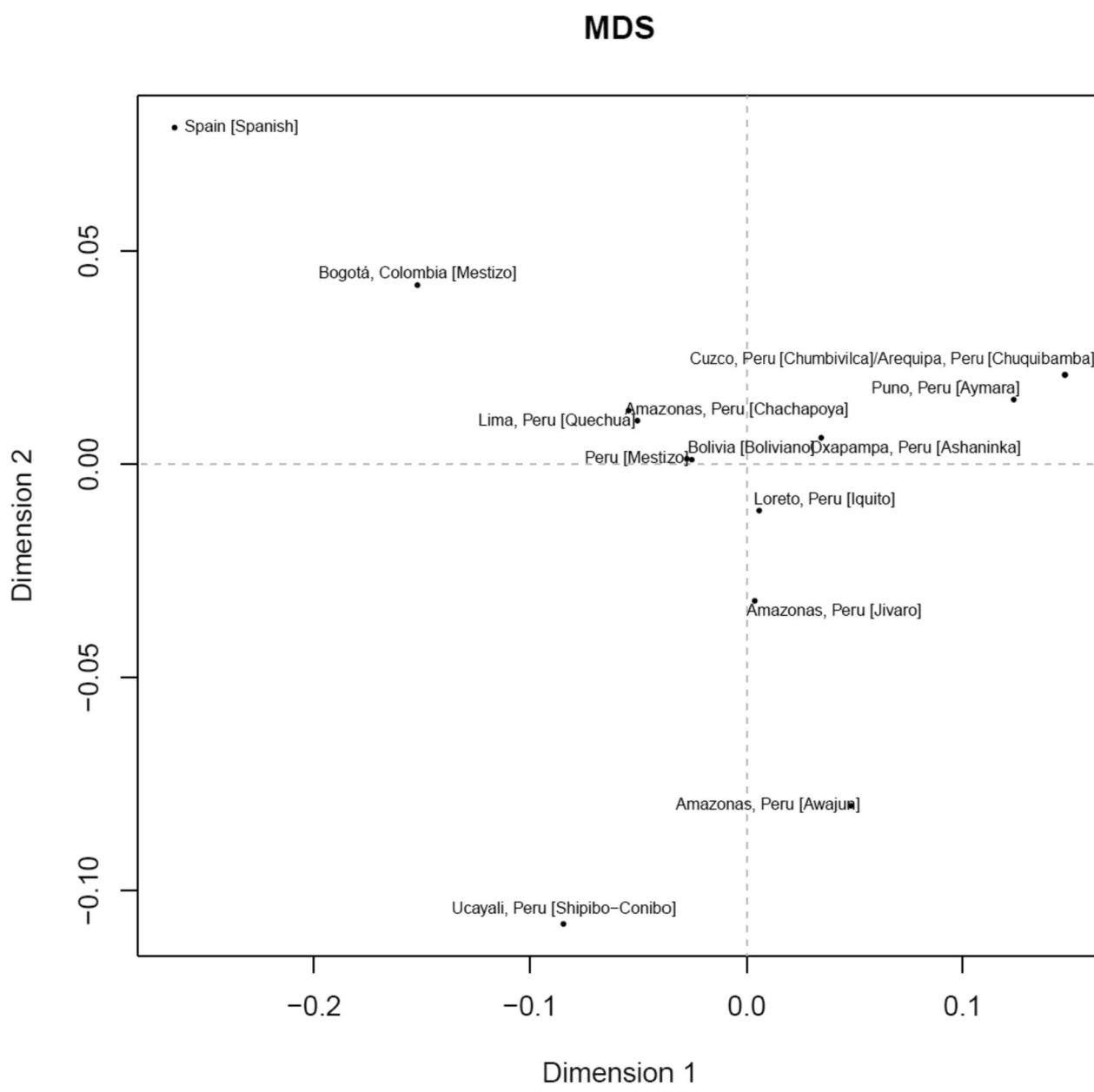


Fig. 2 Multidimensional scaling (MDS) analysis for Mestizo population ($n=290$) and 14 other sample populations. Bolivia [Boliviano] ($n=218$)^a; Bogotá, Colombia [Mestizo] ($n=184$)^b; Oxapampa, Peru [Ashaninka] ($n=57$)^c; Amazonas, Peru [Awajun] ($n=18$)^d; Puno, Peru [Aymara] ($n=90$)^e; Amazonas, Peru [Chachapoya] ($n=106$)^f; Cuzco, Peru [Chumbivilca] ($n=10$)^g; Arequipa, Peru [Chuquibamba] ($n=16$)^h; Junín, Peru [Huanca] ($n=13$)ⁱ; Loreto, Peru [Iquito] ($n=85$)^j; Amazonas, Peru [Jivaro] ($n=21$)^k; Lima, Peru [Quechua] ($n=11$)^l; Ucayali, Peru [Shipibo-Conibo] ($n=21$)^m; and Spain [Spanish] ($n=2406$)ⁿ deposited in the YHRD. Filed under accession numbers: a: YA005867, b: YP000047, c: YA005583, d: YA005891, e: YP000861, f: YA004108, g: YA003820, h: YA003821, i: YA003822, j: YP000860, k: YA005058, l: YA003949, m: YA003825, n: YP001017

Mestizo population and other datasets from Boliviano (Bolivia), Mestizo (Bogotá, Colombia), Aymara (Puno, Peru), Ashaninka (Oxapampa, Peru), Awajun (Amazonas, Peru), Chachapoya (Amazonas, Peru), Chumbivilca (Cuzco, Peru), Chuquibamba (Arequipa, Peru),

Huanca (Junín, Peru), Iquito (Loreto, Peru), Jivaro (Amazonas, Peru), Quechua (Lima, Peru), Shipibo-Conibo (Ucayali, Peru), and Spanish (Spain). There were 17 Y-STRs in common among the data sets (DYS456, DYS389I, DYS390, DYS389II, DYS458,

DYS19, DYS385, DYS393, DYS391, DYS439, DYS635, DYS392, YGATAH4, DYS437, DYS438, DYS448).

Discussion

Molecular biology technologies offer enhanced capabilities for human identification. Forensic DNA typing has an important role in some criminal investigations, which in turn benefits society, by providing critical data for developing investigative leads to identify donors of biological evidence who may be perpetrators of crime as well as to exonerate those that may be wrongly associated with biological evidence. In order to contribute to assessing the significance of an association by DNA between a reference sample and an evidence sample, a population study on a sample population of Peruvians was carried out by analyzing 27 Y-STRs contained within a widely used commercial kit (i.e., Yfiler™ Plus PCR Amplification Kit).

The per locus GD of the studied population ranged between 0.4271 (DYS437) and 0.9415 (DYS385a/b). In contrast, the GD values for the Peruvian Aymara population were between 0.2424 (DYS437) and 0.9298 (DYS385a/b), Peruvian Ashaninka populations were between 0.1337 (DYS437) and 0.6582 (DYS438) and the population of the Peruvian Jungle were between 0.000 (DYS635, DYS448 and DYS437) and 0.7094 (DYS449) (Tineo et al. 2015; Neyra-Rivera et al., 2022; Neyra-Rivera et al. 2023). The values have a similar distribution with those of the Mestizo population of Spain, DYS393 (0.4625) had the lowest GD (Martin et al., 2004).

Because there is no recombination on the Y chromosome where the Y-STRs reside, haplotype diversity is more meaningful than GD to compare diversity among populations. The Y-STR HD was 0.9964 with 283 distinct haplotypes out of 290 individuals analyzed in the Peruvian population studied herein. This level of diversity is slightly lower than that of the Peruvian Aymara (0.9998, $n=90$) and Peruvian Asháninca (1.000) populations and higher than that of the Peruvian Jungle population (0.9910, $n=141$) (Tineo et al. 2015; Neyra-Rivera et al., 2022; Neyra-Rivera et al. 2023). In addition, the diversity value was lower than that of the Mestizo populations of Spain (0.9995, $n=144$), Colombia (0.9971, $n=137$), and Bolivia (1.000, $n=100$) (Martin et al., 2004; Vullo et al. 2015; Yunis et al. 2005). The difference compared with the Mestizo populations may be due in part to fewer samples being typed, a fewer number of markers tested (17 vs 27 markers), and/or stochastic variation.

The specific haplogroups carried by the males analyzed in the Peruvian Mestizo population provide insight into the male lineages of the population. The most prevalent was haplogroup Q (56.18%), which also was the most common haplogroup in the Ashaninka (between 99 and 100%), Aymara (86.52%), and La Selva (91.56%)

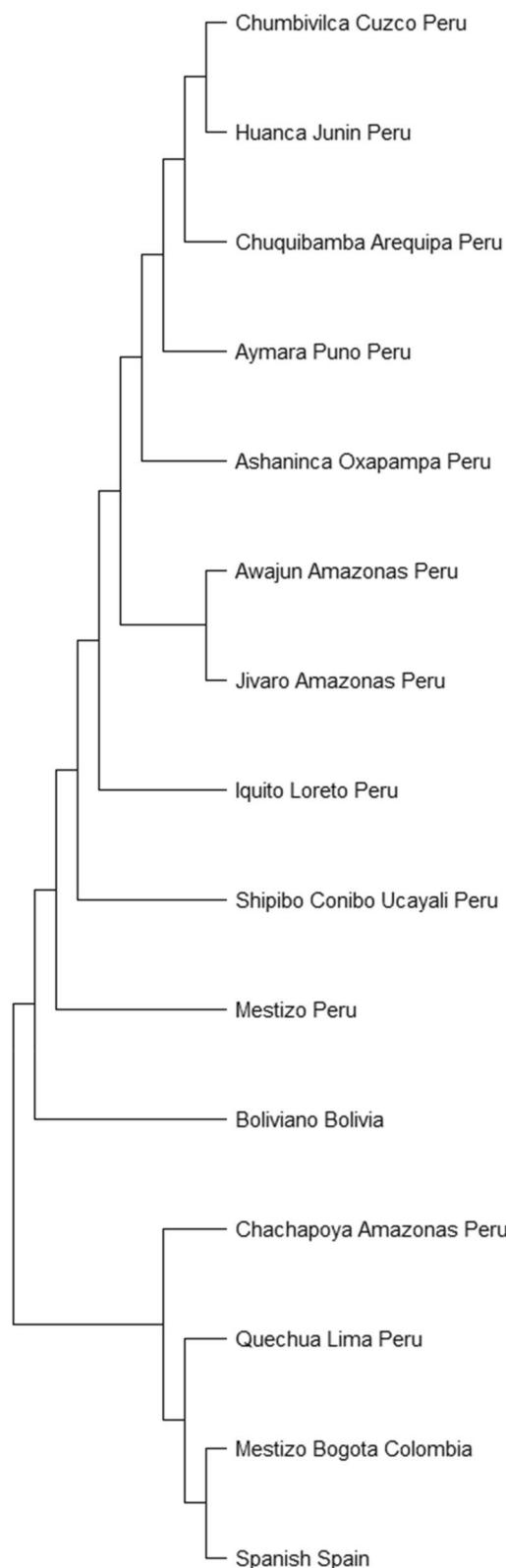


Fig. 3 UPGMA tree based on 17 Y-STR markers in common among 15 populations and Rst estimates

populations. The lower proportion of haplogroup Q in the Mestizo population may be due to admixture that has occurred and continues to occur in Peru; for various reasons, native populations have migrated from their communities to the cities in Peru (Tineo et al. 2015; Neyra-Rivera et al., 2022; Neyra-Rivera et al. 2023).

The RST values were used to generate a UPGMA tree using the Molecular Evolutionary Genetics Analysis (MEGA) 6.06 software (Tamura et al. 2013). The Mestizo population was closest to Bolivian and other Peruvian populations (except Chachapoya and Quechua populations) (Fig. 3). The Mestizo population was closest to the Bolivian population (Bolivia, Boliviano) and farthest from the Jivaro and Awajun (Amazonas, Peru) and Shipibo-Conibo (Ucayali) (Fig. 2). These results suggest that there is a high genetic affinity between the Mestizo population and the other Peruvian populations (except Jivaro, Awajun and Shipibo-Conibo populations). Additionally, the Jivaro, Awajun, and Shipibo-Conibo populations were distant from all groups analyzed which may be due to genetic differences and/or small population sample sizes ($n \leq 21$) (Fig. 3).

Conclusions

In conclusion, 27 Y-STR loci in 290 unrelated Mestizo males were typed, providing a larger dataset for this population group. The data obtained have been deposited in the YHRD database (accession number: YA006020). These data are larger than any other available data for the Mestizo population from Peru. For example, YHRD (Yhrd.org) contains 394 haplotypes (394 with 12 Y-STR haplotypes, 229 with 17 Y-STR haplotypes, 83 with 23 Y-STR haplotypes, and until the study herein 0 with 27 Y-STR haplotypes) (filed under accession numbers YA003399 and YA005599). A specific limitation of this study is the small sample sizes for some groups which can lead to errors in the allele frequency estimates and subsequent population genetic calculations. Regardless of this limitation, the analyses carried out herein show that the Y-STR haplotypes are highly polymorphic and thus can be useful in human identification forensic cases, with proper statistical methods that account for uncertainty, as well as may have value for studies on the history of the human populations in Latin America.

Abbreviations

AMOVA	Analysis of molecular variance
MDS	Multidimensional scaling plot
PCR	Polymerase chain reaction
YHRD	Y Haplotype Reference Database
Y-STR	Y-chromosome-specific short tandem repeat

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Authors' contributions

Carlos Neyra, Edgardo Delgado, and Ericson Gutierrez participated in the design of the study. Carlos Neyra and Edgardo Delgado processed the samples and obtained the genetic profiles. Carlos Neyra, Edgardo Delgado, Ericson Gutierrez, and Bruce Budowle carried out the statistical analysis. All participated in the writing and approval of the final version of the article.

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Availability of data and materials

The datasets analyzed during the current study are available from the corresponding author.

Declarations

Ethics approval and consent to participate

The study was approved by the ethics committee of Instituto de Medicina Tropical "Daniel Alcides Carrion" from San Marcos National University (Certificate of Approval CIEI-2018-015).

Consent for publication

Consent has been given by all authors for publication of this study.

Competing interests

The authors declare that they have no competing interests.

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