

Fabricio González-Andrade,<sup>1</sup> M.D., Ph.D.; Dora Sánchez,<sup>1</sup> M.Sc.; Begoña Martínez-Jarreta,<sup>2</sup> M.D., Ph.D.; and Bruce Budowle,<sup>3</sup> Ph.D.

## Y-Chromosome STR Haplotypes in Three Different Population Groups From Ecuador (South America)

**POPULATIONS:** Over 102 unrelated Mestizos, 102 Native Amerindian (Kichwas), and 102 African Americans who represent the three largest communities of the country Ecuador (South America).

**KEYWORDS:** forensic science, DNA profiling, STRs, Y-Chromosome, population genetics, Mestizos, Amerindians, Kichwas, African Americans, Hispanics, Ecuador

Peripheral blood samples were collected in EDTA vacutainers tubes by venipuncture from healthy unrelated Mestizo, Kichwa, and African American populations, born and living in Ecuador ( $n = 102$  individuals for each group). Genomic DNA was extracted using Wizard system<sup>®</sup> (Promega Corp., Madison, WI), and the quantity was estimated by ultraviolet (UV) absorbance. PCR amplification and typing were carried out by using the kit PowerPlex Y (Promega Corp.) and following the manufacturer's recommendations. The ABI Prism 310 Genetic Analyzer (Applied Biosystem, Foster City, CA) was used for detection of the amplified products. The recommendations of the DNA Commission of the International Society for Forensic Genetics were followed for typing and interpretation (1). Quality control on allele designations was assured by internal protocols of our laboratory (2) and by successful participation in the quality control test of the Grupo Español y Portugués, International Society for Forensic Genetics (GEP-ISFG). Gene diversity at each locus, the number of haplotypes, and haplotype diversity were calculated using a program developed by Chakraborty and Lee (<http://Cgi.uc.edu/download/haplo>). Linkage disequilibria tests, with 5000 permutations, were performed using the same program. The DYS385 locus was treated as a single locus for allele frequency estimates (3). The diversity estimates for each of the DYS385 loci were calculated by arbitrarily assigning the smaller size allele to the DYS385a locus.

Table 1 summarizes the allele frequency distribution and gene diversity for each locus in the three populations studied. Most frequently observed correct haplotypes are shown in Table 2. Complete data are available upon request through electronic mail from the corresponding author at: [fabriciogonzaleza@yahoo.es](mailto:fabriciogonzaleza@yahoo.es).

Observed haplotype diversities were in the in range between 0.997 and 0.999 (see Table 3).

### References

1. Gill P, Brenner C, Brinkmann B, Budowle B, Carracedo A, Jobling MA, et al. DNA Commission of the ISFG: recommendations on forensic analysis using Y-chromosome STRs. *Int J Legal Med* 2001;114:305–9.
2. González-Andrade F, Sánchez D, Martínez-Jarreta B. Evaluation of 1495 cases of disputed paternity in Ecuador (South America) resolved with STR-PCR polymorphisms. *Proceedings of International Association of Forensic Sciences*. Bologna (Italy): Monduzzi Ed: 2002;261–4.
3. Budowle B, Adamowicz M, Aranda X, Barna C, Chakraborty R, Eisenberg AJ, et al. Twelve short tandem repeat loci Y chromosome haplotypes: genetic analysis on populations residing in North America. *Forensic Sci Int* 2005;50:1–15.

Additional information and reprint requests:

Fabricio González-Andrade, M.D., Ph.D.  
Hospital Metropolitano, Molecular Genetics Laboratory  
Av. Mariana de Jesus Oe8  
Quito, EC170104  
E-mail: [fabriciogonzaleza@yahoo.es](mailto:fabriciogonzaleza@yahoo.es)

<sup>1</sup>Molecular Genetics Laboratory, Metropolitan Hospital, Quito, EC170104 Ecuador.

<sup>2</sup>Department of Legal Medicine, University of Zaragoza, Faculty of Medicine, C/Domingo Miral s/n, 50.009 Zaragoza, Spain.

<sup>3</sup>Laboratory Division, FBI, Quantico, VA

TABLE 1—Allele frequencies and gene diversities of the 12 Y-chromosome STR loci in three Ecuadorian populations.

Genetic marker	Mestizos (n = 102)	Kichwas Amerindian (n = 102)	Blacks Afroamericans (n = 102)
<i>DYS19</i>			
12	0.0196	0.0294	0.1765
13	0.3235	0.6078	0.2059
14	0.4510	0.2157	0.3137
15	0.1667	0.0980	0.1471
16	0.0392	0.0392	0.1569
18	—	0.0098	—
Gene diversity	0.6688	0.7756	0.7896
<i>DYS385a</i>			
9	—	0.0098	—
10	0.0049	—	0.0049
11	0.1814	0.0294	0.0735
12	0.0931	0.0784	0.0341
13	0.1030	0.0490	0.0441
14	0.2598	0.2745	0.1324
15	0.0882	0.2010	0.1569
16	0.0686	0.0882	0.1814
17	0.1078	0.0980	0.1863
18	0.0441	0.0588	0.1177
19	0.0343	0.0784	0.0441
20	0.0098	0.0245	0.0196
21	0.0049	0.0098	0.0049
Gene diversity	0.7672	0.8606	0.8396
<i>DYS385b</i>			
9	—	0.0098	—
10	0.0049	—	0.0049
11	0.1814	0.0294	0.0735
12	0.0931	0.0784	0.0341
13	0.1030	0.0490	0.0441
14	0.2598	0.2745	0.1324
15	0.0882	0.2010	0.1569
16	0.0686	0.0882	0.1814
17	0.1078	0.0980	0.1863
18	0.0441	0.0588	0.1177
19	0.0343	0.0784	0.0441
20	0.0098	0.0245	0.0196
21	0.0049	0.0098	0.0049
Gene diversity	0.7672	0.8606	0.8396
<i>DYS389I</i>			
12	0.1569	0.1863	0.1275
13	0.6078	0.6078	0.6373
14	0.2255	0.2059	0.2353
15	0.0098	—	—
Gene diversity	0.5604	0.5589	0.5274
<i>DYS389II</i>			
27	0.0196	—	—
28	0.7843	0.0490	0.0784
29	0.3824	0.2451	0.1667
30	0.3624	0.3529	0.4118
31	0.1078	0.2843	0.3039
32	0.0490	0.0683	0.0392
Gene diversity	0.7086	0.7346	0.7096
<i>DYS390</i>			
20	—	—	0.0098
21	0.0196	0.0098	0.4118
22	0.0588	0.0392	0.0980
23	0.2157	0.2059	0.1274
24	0.6078	0.5784	0.2451
25	0.0984	0.1667	0.0980
26	—	—	0.0098
Gene diversity	0.5762	0.5995	0.7420
<i>DYS391</i>			
9	0.0294	0.1078	0.0392
10	0.5392	0.6177	0.6569
11	0.3726	0.2647	0.2843
12	0.0490	0.0098	—
13	0.0098	—	0.0196
Gene diversity	0.5727	0.5420	0.4906

TABLE 1—Continued.

Genetic marker	Mestizos (n = 102)	Kichwas Amerindian (n = 102)	Blacks Afroamericans (n = 102)
<i>DYS392</i>			
10	0.0196	—	—
11	0.2451	0.0686	0.6471
12	0.0196	0.0294	0.0490
13	0.4706	0.1863	0.1765
14	0.2059	0.5294	0.1078
15	0.0392	0.1373	0.0196
16	—	0.0196	—
17	—	0.0980	—
18	—	0.0196	—
Gene diversity	0.6805	0.6663	0.5411
<i>DYS393</i>			
10	—	—	0.0098
11	0.0098	0.0098	0.0588
12	0.1177	0.0098	0.5196
13	0.7157	0.7255	0.2549
14	0.1373	0.2157	0.1569
15	0.0197	0.0392	—
Gene diversity	0.4591	0.4296	0.6432
<i>DYS437</i>			
13	0.0686	0.0098	0.0490
14	0.5784	0.8235	0.7059
15	0.3137	0.1275	0.1863
16	0.0392	0.0392	0.0392
17	—	—	0.0196
Gene diversity	0.5663	0.3069	0.4673
<i>DYS438</i>			
8	—	0.0980	0.0294
9	0.1275	0.0490	0.1961
10	0.2157	0.0980	0.1471
11	0.1961	0.7059	0.5588
12	0.4510	0.1373	0.2451
18	0.0098	—	—
Gene diversity	0.7022	0.4754	0.6108
<i>DYS439</i>			
9	0.0098	0.0294	—
10	0.0196	—	0.0392
11	0.2941	0.3137	0.3235
12	0.4902	0.5196	0.5098
13	0.1667	0.1275	0.1275
14	0.0098	0.0098	—
15	0.0098	—	—
Gene diversity	0.6511	0.6205	0.6238

TABLE 2—Haplotypes found in more than two individuals.

H	n	Frequency	DYS19	DYS385a	DYS385b	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393	DYS437	DYS438	DYS439
<i>Mestizos</i>														
h1	2	0.0196	16	12	15	13	29	23	10	11	12	9	11	15
h2	2	0.0196	12	15	17	13	30	23	10	14	13	10	12	13
h3	2	0.0196	14	11	14	13	29	24	11	14	13	12	12	14
<i>Kichwas (native Amerindians)</i>														
h1	2	0.0196	14	15	17	13	31	25	10	14	13	14	11	12
h2	2	0.0196	15	14	14	12	31	24	10	18	13	16	11	11
h3	2	0.0196	15	14	15	13	29	23	10	14	13	14	9	11
h4	2	0.0196	16	19	19	13	30	25	10	13	14	15	10	11
h5	2	0.0196	13	15	16	13	29	24	10	14	13	14	11	12
h6	2	0.0196	14	14	20	13	29	25	10	15	14	14	11	12
h7	3	0.0294	13	14	14	13	30	24	10	13	13	14	11	12
h8	2	0.0196	14	12	14	12	29	24	11	13	13	15	12	11
h9	2	0.0196	15	9	21	14	30	24	11	12	13	14	11	9
h10	2	0.0196	13	19	19	14	31	24	11	14	13	14	11	12
<i>African Americans</i>														
h1	2	0.0196	13	13	14	13	30	24	9	11	13	10	10	14
h2	2	0.0196	15	16	15	12	28	23	10	12	15	10	11	14
h3	2	0.0196	15	16	17	13	30	21	10	11	15	11	12	13
h4	2	0.0196	16	15	18	13	30	21	10	11	14	11	12	14
h5	2	0.0196	15	15	17	13	31	21	10	11	13	11	12	14
h6	2	0.0196	15	17	18	13	31	21	10	11	15	11	12	14
h7	3	0.0294	14	11	18	13	29	23	10	13	13	12	13	15
h8	2	0.0196	17	16	18	13	30	21	10	11	15	12	13	14
h9	2	0.0196	15	15	16	14	31	21	10	11	13	11	11	14
h10	2	0.0196	14	11	14	13	29	25	11	13	13	12	12	15
h11	3	0.0294	15	16	17	13	31	21	11	11	13	11	12	14
h12	2	0.0196	17	17	18	14	30	21	11	11	14	11	11	14
h13	2	0.0196	14	11	14	13	28	24	13	13	13	12	12	15

Haplotypes occurring more than once, highest haplotype frequency (observed one time) = 3 (0.0294).

TABLE 3—Haplotype information on 12 Y STR loci in three Ecuadorian populations.

	Mestizos	Kichwas Amerindians	Blacks Afroamericans
Sample size	102	102	102
Number of unique haplotypes	99	91	89
Haplotype diversity	0.9994	0.9977	0.9975
Random match probability	0.0006	0.0023	0.0025