TECHNICAL NOTE

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Population Frequency for the Short Tandem Repeat Loci D18S849, D3S1744, and D12S1090 in Caucasian-Mestizo and African Descent Populations of Colombia

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ABSTRACT: Blood samples from 489 unrelated Caucasian Mestizo and 252 individuals of African descent in Colombia were amplified and typed for three short tandem repeat (STR) markers (D12S1090, D3S1744, and D18S849). All markers conformed to Hardy-Weinberg equilibrium expectations in both populations studied. In addition, heterozygosity, mean exclusion chance, polymorphism information content, discrimination power, and the assumption of independence within and between loci were determined. The mean exclusion chance for all three STR markers is 0.9750 in the Caucasian Mestizo population and 0.9731 in the African Colombian Population. The discrimination power is 0.999925 and 0.999911 in the Caucasian Mestizo and African Colombian respectively.

KEYWORDS: forensic science, DNA typing, polymerase chain reaction, short tandem repeats, population genetics, D12S1090, D3S1744, D18S849, Colombia

In South America, few STR studies have been done for native, Mestizo populations (1–3), and South American Black populations (4), although many studies of STR profiles have been carried out for the ancestral Spanish and Portuguese populations (5–9). Colombia is a multi-ethnic and multi-cultural country with near 41 million people composed of three ethnic groups (10), the Caucasian-Mestizo population representing the largest group followed by the Black population of African origin and the Amerindian populations (83 ethnic groups). The Caucasian-Mestizo population is composed of Spanish descent and in a lesser extent of other European, Arabs and Jewish populations among others and is located in the Andean region of Colombia and in a lesser extent in the Caribbean plains, Pacific and Amazonian regions. The Black population is located in the Pacific and Caribbean regions of the country (11,12). The ancestors of the Black population of Colombia were brought as slaves from the west coast of Guinea, Ivory Coast, Senegal, and Mali in Africa to present day Colombia between 1580 and 1650 to work in the fields and mines (13). By 1800, around 210,000 individuals of African descent (14) were living in the country, in the Caribbean and pacific coasts of Colombia. In 1993 a total of 502,343 individuals (1.5% of the population) were counted (10) as Blacks.

We present the population frequencies of three polymorphic STR loci analyzed in two major Colombian populations named D12S1090 (n = 486 Caucasian-Mestizo; n = 248 Blacks), D3S1744 (n = 484 Caucasian-Mestizo; n = 252 Blacks) and D18S849 (n = 489 Caucasian Mestizo; n = 250 Blacks). The population frequencies obtained from these markers, not only are important for forensic science and paternity testing studies, but to characterize genetically the Colombian population and sub-populations.

Materials and Methods

Whole blood was obtained from unrelated Caucasian-Mestizo individuals from the Andean Region of Colombia requesting paternity testing studies and from unrelated individuals of African descent collected in five different towns (around 50 individuals from each town) of the Choco department in the Pacific region of Colombia. Informed consent was obtained before drawing the blood samples. The selection of the African descent individuals was based on the fact that at least two generations did not have admixture with Caucasian or Amerindians based on interrogation at the moment of the sample collection. None of these samples are presumed to be from first-degree relatives.

Genomic DNA was isolated from whole blood by the Quick Light DNA isolation kit (Lifecodes Corporation, Stamford, CT) or by the Wizard Genomic DNA isolation kit (Promega Corporation, Madison, WI) following the manufacturer's recommendations. The D12S1090, D3S1744, and D18S849 loci were amplified by PCR in a multiplex format using the Multiplex I kit (Lifecodes Corporation, Stamford, CT) in a PTC100VG thermocycler (MJ Research, Watertown, MA) following the manufacturers protocols. The PCR products were resolved in a 4% Acryl-

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amide-Bis-Acrylamide denaturing gel at 2000 V/1 h and detected by silver nitrate staining (15) and manually interpreted by two persons. Allele designations were based on the allelic ladder provided by the manufacturer.

Statistical evaluations were performed using the HWE-Analysis software package (HWE-Analysis, Version 3.3. Christoph Puers, Institute of Legal Medicine, University of Münster). Analyses included the possible divergence from Hardy-Weinberg expectations and other parameters of forensic importance: observed and expected heterozygosity (16), mean exclusion chance (MEC) (17), mean paternity exclusion probability (MEP) (18), polymorphic information content (PIC) (19), and discrimination power (DP) (20). In addition the GDA computer program (Lewis, P.O., Zaykin, D. 1999. Genetic Data Analysis. Computer program for the analysis of allelic data. Version 1.0 (d12), free program distributed by the authors over the internet from the GDA Home Page at http//chee.unm.edu/gda/), was used to analyze the possible associations between loci.

Results and Discussion

Table 1 shows the allele frequency distribution for D12S1090, D3S1744, and D18S849 in the Caucasian-Mestizo and African descent individuals of Colombia. In addition, minimum allele frequencies for PCR based loci based on statistical and populations genetics theory were determined (21–23) (Table 1). Therefore, a greater confidence with current size databases can be attained for DNA profile frequency estimates in forensic casework. Also, the results of the different test procedures to determine the correspondence of the genotype frequencies with their Hardy-Weinberg equilibrium are shown. All markers were found to be in HW equilibrium by all three test used (X^2-test, the logarithmic likelihood ratio (G test) and exact test) (24). An interclass correlation test analysis demonstrated that there is no evidence of associations between any pair of loci in any of these two populations (data not shown), as should be expected when independent loci in different chromosomes are analyzed.

To evaluate the differences between these two populations, a R $\times$ C contingency table was used (data not shown). The allele frequency distribution for locus D3S1744 did not show any statistical significant difference between these two populations ($0.1 < p < 0.2$). Statistical significant differences were detected for locus D12S1090 and D18S849 ($p < 0.0005$). A similar comparison (data not shown) was carried out between the Colombian Caucasian Mestizo population and the USA Caucasian population, between the Colombian Caucasian Mestizo and the USA Hispanics (reported by the manufacturer) and between Black population from Colombia and Blacks from the USA (17) showing no statistically significant differences between them.

No statistical significant differences were observed when R $\times$ C tables were used to compare the data obtained from the five different towns of the Choco department sampled despite the low number of individuals analyzed (data not shown). This findings could

TABLE 1—*Observed allele frequencies and tests for Hardy-Weinberg equilibrium for D18S849, D3S1744, and D12S1090 loci, in Caucasian-Mestizos and Black individuals from Colombia.*

Allele	Caucasian Mestizo $n = 489$ D18S849	Caucasian Mestizo $n = 484$ D3S1744	Caucasian Mestizo $n = 486$ D12S1090	Blacks $n = 250$ D18S849	Blacks $n = 252$ D3S1744	Blacks $n = 248$ D12S1090
9	0.001		0.069	0.062		0.022
10			0.017			0.010
11	0.001		0.053	0.004		0.057
12	0.003		0.063	0.016		0.111
13	0.010		0.034	0.006		0.036
14	0.048	0.004	0.045	0.020	0.002	0.042
15	0.212	0.096	0.012	0.218	0.066	0.012
16	0.408	0.084	0.006	0.390	0.089	0.002
17	0.197	0.142	0.004	0.192	0.173	0.012
18	0.095	0.367	0.018	0.086	0.365	0.020
19	0.022	0.173	0.055	0.006	0.175	0.050
20	0.003	0.101	0.083		0.079	0.099
21		0.027	0.076		0.042	0.093
22		0.007	0.108		0.006	0.119
23			0.066		0.004	0.061
24			0.066			0.034
25			0.058			0.057
26			0.077			0.046
27			0.060			0.038
28			0.011			0.008
29			0.012			0.032
30			0.007			0.020
31						0.012
32			0.001			0.002
33						0.004
Frec.min	0.006	0.006	0.007	0.011	0.012	0.014
HET obs.	26	29	141	23	27	116
HET Ex. SE.	24.34 ± 4.31	27.33 ± 3.46	144.37 ± 10.91	23.11 ± 4.41	24.95 ± 3.84	114.78 ± 12.72
Homo obs.	5	6	14	5	6	9
Homo Ex. SE	4.93 ± 1.31	6.28 ± 1.06	12.07 ± 2.91	4.63 ± 1.43	5.69 ± 1.69	8.18 ± 3.35
X^2 test	0.147	0.200	0.463	0.908	0.939	0.377
G test	0.116	0.196	0.412	0.931	0.923	0.148
Exact test	0.055	0.118	0.472	0.952	0.802	0.127

TABLE 2—*Statistical parameters of forensic importance for D12S1090, D3S1744, and D18S849 loci in Caucasian Mestizo and Blacks from Colombia.*

	Caucasian Mestizo	Caucasian Mestizo	Caucasian Mestizo	Blacks	Blacks	Blacks
Locus	D18S849	D3S1744	D12S1090	D18S849	D3S1744	D12S1090
HET obs	0.714	0.775	0.930	0.732	0.774	0.944
MEC	0.519	0.604	0.869	0.539	0.599	0.862
MEP	0.491	0.579	0.870	0.515	0.576	0.865
PIC	0.701	0.763	0.932	0.717	0.760	0.928
DP	0.892	0.929	0.990	0.908	0.927	0.987

	Caucasian Mestizo	Blacks
MEC Total	0.9750	0.9731
DP total	0.999925	0.999911

be due in part to a process of genetic "homogenization" carried out by the slavers in Colombia (13). In that process, individuals speaking the same language were separated in order to avoid emancipation movements among the slaves. A similar result was obtained comparing Black populations of Brazil and Venezuela (4). This admixture process will limit the usefulness of STR markers to trace back the origin of the Colombian Black population with their specific African ancestors.

Table 2 shows several parameters of statistical importance for the loci studied in the Caucasian Mestizo and Black populations of Colombia such as observed and expected heterozygosity, mean exclusion chance (MEC), mean paternity exclusion (MEP), polymorphic information content (PIC) and discrimination power (DP). Similar results were obtained in both populations for the observed heterozygosity, MEC, and DP.

In summary, the population frequency of Caucasian-Mestizo and African Colombians has been established for STR loci D12S1090, D3S1744, and D18S849. All STR loci were found to be in HW equilibrium. The combined power of exclusion is estimated as 0.9750 in the Caucasian Mestizo and 0.9731 in the African Colombian population. The results obtained in this study can be used to derive estimates of multiple loci profiles frequencies for forensic purposes, to calculate paternity indexes and the probability of paternity in parentage testing studies and to genetically characterize the Colombian population in population studies.

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