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Data analysis of D1S1656, D12S391 and D18S535 loci in two Colombian populations: Antioquia and Chocó

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Abstract. In the present study, we analyzed allelic and genotypic distribution of three short tandem repeat (STR) loci determined in samples from unrelated individuals from two Colombian populations: Antioquia (314) and Chocó (140). The three loci were found in Hardy-Weinberg equilibrium and they are highly polymorphic in the two populations studied with the locus D18S535 (77.2%) having the lowest observed heterozygosity (Antioquia population), and the locus D1S1656 (89.8%) displaying the highest heterozygosity. The most discriminating locus was D1S1656, Antioquia (PD = 0.976) and Chocó (0.974). The combined probability of exclusion for the three STR loci is 0.964 in Antioquia and 0.967 in Chocó populations. The combined probability of discrimination is 0.9999 in both populations, demonstrating to be a group of excellent markers to be used as complement in the paternity testing, individual identification and other forensic applications in general.

Keywords: Antioquia; Chocó; Colombia; Population; DNA; STR; D1S1656; D12S391; D18S535

1. Introduction

Short tandem repeats (STR) are polymorphic markers widely distributed throughout the human genome [1] that can be amplificated by PCR techniques. Only some of the them have been studied for their use in forensic genetic and paternity testing. The general criteria for selecting STRs include characteristics such as high heterozygosity, low stutter, few and low extra-peaks, robustness, easy multiplexing and a low mutation rate [2]. The aim of this study was to report allele frequency distributions for the D1S1656, D12S391

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Table 1
Allele frequencies and statistical parameters

Allele	D1S1656		D12S391		D18S535	
	Antioquia	Chocó	Antioquia	Chocó	Antioquia	Chocó
9					0.04369	0.02878
10	0.00647	0.01095	0.00161		0.00243	0.01079
11	0.04531	0.05839	0.00161		0.01942	0.12590
12	0.10032	0.06204	0.00161		0.22573	0.26259
13	0.06634	0.09854		0.00362	0.33981	0.23381
14	0.11974	0.20438			0.24515	0.25180
15	0.16019	0.17153	0.02412	0.09420	0.10922	0.08273
15.3	0.05178	0.01825				
16	0.16990	0.09489	0.04019	0.06522	0.01214	0.00360
16.3	0.04207	0.09124				
17	0.06634	0.02920	0.09003	0.12319	0.00243	
17.3	0.12621	0.10584				
18	0.00809		0.22990	0.22464		
18.3	0.03398	0.04745				
19			0.27492	0.17754		
19.3	0.00324	0.00730				
20			0.11415	0.18116		
21			0.07878	0.03986		
22			0.05466	0.03623		
23			0.06592	0.03261		
24			0.01286	0.01449		
25			0.00804			
26			0.00161	0.00725		
Minimun	0.0101	0.0226	0.0097	0.0307	0.0092	0.0206
N	314	140	314	140	314	140
Parameters						
Ho	0.883	0.898	0.836	0.804	0.772	0.799
He	0.889	0.883	0.836	0.856	0.761	0.792
HWE-H	0.811	0.878	0.689	0.873	0.379	0.324
Exact test	0.981	0.510	0.177	0.157	0.893	0.634
PD	0.976	0.974	0.949	0.958	0.899	0.915
PE	0.762	0.791	0.667	0.607	0.548	0.596
MP	0.024	0.026	0.051	0.042	0.101	0.085
PIC	0.880	0.870	0.820	0.840	0.720	0.760
TPI	4.290	4.890	3.050	2.560	2.190	2.480

Ho: Heterozygosity observed, He: Heterozygosity expected, HWE-H: Hardy-Weinberg test, PD: Power of Discrimination, PE: Power of Exclusion, MP: Matching Probability, PIC: Polymorphism information content, TPI: Typical Paternity Index.

and D18S535 loci in two Colombian populations: Antioquia [3,5,6] and Chocó [4,7], as well as statistical evaluations, including the possible divergence from Hardy-Weinberg equilibrium and other forensic parameters of interest.

2. Materials and methods

Samples were obtained from 140 unrelated individuals of African descent from the Chocó region and 314 unrelated Caucasian individuals from Antioquia with a similar proportion of male and females. Genomic DNA was extracted by a salting out extraction procedure [8]. The primer sequences of D1S1656, D12S391 and D18S535 loci and cycling conditions were as recommended by Lareu et al. [2]. Briefly, each PCR reaction contained 2–10 ng human genomic DNA, 1 × Taq buffer, 1.5 mM MgCl2, 200 μM each dNTP, 1U Taq polymerase (Promega, USA), 0.25 μM each primer in a total volume of 15 μl. A total of 30 cycles were carried out in a PTC-100 (MJ-Research) PCR System with denaturation for 45 s at 95 °C, annealing for 60 s at 6 °C and extension for 60 s at 72 °C.

The PCR products were analyzed using denaturing 4% acrylamide-bis-acrilamide gel electrophoresis and detected by silver staining (Promega). Typing was made by comparison with the sequenced allelic ladders, kindly provided by the Institute of Legal Medicine, University of Santiago de Compostela (Spain). The frequency of each allele for each locus tested was calculated from the number of observed genotypes in the sample using GENEPOP software ver. 3.2a [9]. The exact-test for within-locus (Hardy-Weinberg equilibrium) and between-loci testing (linkage desequilibrium) was carried out by using GENEPOP ver. 3.2a [9] and GDA ver. 1.0 softwares. Forensic parameters were performed using POWERSTATS ver. 12 (Promega) and GDA ver. 1.0 [10] softwares.

3. Results and discussion

Antioquia and Chocó population data for the three STRs are shown in Table 1. No deviations from Hardy-Weinberg equilibrium were observed for all markers in both populations. A comparative study between the two populations showed significant differences for the three loci ($P = 0.000$). On the other hand, Antioquian population showed a similar allelic distribution for the three loci with other Caucasian European populations, confirming findings of previous research showing a high Caucasian component in the Antioquia population [3,5,6]. D1S1656 was the most informative in both populations. The Combined Power of Discrimination (CPD) was of 0.9999 in both populations and the Combined Power of Exclusion (CPE) was 0.964 and 0.967 in the populations of Antioquia and Chocó, respectively.

In conclusion, two population databases have been established for the Caucasian population of Antioquia and for the African descent population of Chocó for STR loci D1S1656, D12S391 and D18S535. The results showed that all three loci are very useful for personal identification and paternity testing in the Colombian populations of Antioquia and Chocó.

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