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Population genetics of eight new Y-chromosomal STR haplotypes in three Colombian populations: Antioquia, Chocó and Cartagena

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Abstract. In this study, eight new Y-chromosomal (GATA-C4, DYS438, DYS437, DYS461, GATA-H4, DYS439, GATA-A10 and DYS460) short tandem repeat (STR) polymorphic systems were typed by two PCR quadruplex reactions in three Colombian populations: Antioquia, Chocó and Cartagena. GATA-C4 showed the highest gene diversity in the three populations. In the Antioquia population sample, a total of 123 different haplotypes were observed among which 85 were unique and 38 were found at least two times; in the Chocó population sample: 55 different haplotypes, 47 unique and 8 at least two times; in the Cartagena population sample: 82 different haplotypes, 77 unique and 5 at least two times. The highest average gene diversity was found in Cartagena and the lowest was in Chocó. The haplotype diversity for the eight Y-chromosomal STR loci in the Antioquia, Chocó and Cartagena population samples were calculated to be 0.9949, 0.9959 and 0.9940, respectively.

Keywords: Antioquia; Chocó; Cartagena; Y-chromosome; STR; Population; Colombia

1. Introduction

Analysis of short tandem repeat (STR) markers located on the nonrecombining portion of the human Y-chromosome is a powerful tool in male identification, paternity testing and evolutionary studies [1–3].

In this study, eight recently described [4,5] microsatellites (GATA-C4, DYS438, DYS437, DYS461, GATA-H4, DYS439, GATA-A10 and DYS460) have been analysed in three Colombian populations: Antioquia, a population constituted mainly by Caucasian [6]; Chocó, whose inhabitants are mainly of African origin [7]; and Cartagena, which is an admixture of African people and Caucasian named mulattoes [8]. In this work, we analysed some of the parameters that are important for their use in population and forensic studies.

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2. Materials and methods

Blood samples were obtained from 179 unrelated Caucasian males from Antioquia, 63 unrelated males of African descent from the Chocó region and 92 unrelated mulatto males from Cartagena. Genomic DNA was extracted by a salting out extraction procedure [9]. The primer sequences of loci and cycling conditions were as recommended [4,5,10].

The eight loci were amplified in two multiplexes. Multiplex I includes four STRs: GATA-C4, DYS438, DYS437 and DYS461. Multiplex II includes: GATA-H4, DYS439, GATA-A10 and DYS460. PCR conditions were as follows: 5 ng DNA in 15 μl total reaction volume containing 2 mM $MgCl_2$, 1 × PCR Taq DNA Polimerasa buffer, 0.5 U Taq DNA Polimerasa

Table 1
Gene frequencies and diversities of the eight systems in Antioquia ($n = 179$), Chocó ($n = 63$) and Cartagena ($n = 92$) populations

Locus	Alelles	GF			GD		
		Antioquia	Chocó	Cartagena	Antioquia	Chocó	Cartagena
Y-GATA C4	17			0.011	0.771	0.741	0.735
	19	0.028		0.011			
	20	0.039	0.048	0.054			
	21	0.201	0.302	0.196			
	22	0.156	0.254	0.174			
	23	0.458	0.333	0.435			
	24	0.101	0.032	0.109			
	25	0.017	0.016	0.011			
	26		0.016				
DYS438	7			0.011	0.693	0.665	0.720
	9	0.173	0.016	0.120			
	10	0.190	0.159	0.261			
	11	0.134	0.444	0.185			
	12	0.475	0.349	0.413			
	13	0.028	0.032	0.011			
DYS437	13	0.011	0.032		0.586	0.549	0.596
	14	0.430	0.603	0.413			
	15	0.475	0.302	0.478			
	16	0.084	0.063	0.109			
DYS461	10	0.022		0.011	0.561	0.636	0.617
	11	0.196	0.111	0.207			
	12	0.615	0.476	0.533			
	13	0.162	0.365	0.250			
	14	0.006	0.048				
Y-GATA H4	26	0.034	0.048	0.011	0.564	0.574	0.568
	27	0.385	0.556	0.402			
	28	0.536	0.349	0.522			
	29	0.045	0.048	0.065			
DYS439	9	0.028	0.016	0.022	0.645	0.599	0.655
	10	0.028	0.048	0.065			
	11	0.279	0.317	0.293			
	12	0.508	0.540	0.500			
	13	0.145	0.079	0.109			
	14	0.011		0.011			
Y-GATA A10	11		0.016		0.613	0.671	0.659
	13	0.056	0.190	0.076			
	14	0.251	0.397	0.250			
	15	0.553	0.381	0.500			
	16	0.134	0.016	0.174			
	17	0.006					
DYS460	9	0.022		0.043	0.564	0.462	0.614
	10	0.436	0.317	0.380			
	11	0.497	0.667	0.489			
	12	0.045	0.016	0.087			

GF: Gene frequencies. GD: Gene diversity.

(Promega), 200 μM of each dNTP, 0.53 μM GATA-C4 primers, 1.6 μM DYS438 primers, 0.8 μM DYS437 primers, 1.6 μM DYS461 primers, 0.8 μM GATA-H4 primers, 0.67 μM DYS439 primers, 0.53 μM GATA-A10 primers and 1.06 μM DYS460 primers. The PCR products were analyzed using denaturing 4% acrylamide-bis-acrilamide gel electrophoresis and detected by silver staining (Promega). Alleles were identified based on the number of repeats, and their attribution was made by comparison with an in-house constructed allelic ladder and following the published nomenclature [10] and ISFG guidelines for STR analysis [11]. The gene frequencies and gene or haplotype diversity values were calculated using the software ARLEQUIN version 2000 [12] and Nei formulation [13].

3. Results and discussion

Gene frequencies and diversity values for each STR obtained for the three populations are shown in Table 1. GATA C4 was the most informative loci with gene diversity of 0.771, 0.747 and 0.735 for Antioquia, Chocó and Cartagena, respectively. Cartagena population showed the highest average gene diversity (0.6455); on the other hand, Chocó population showed the lower average gene diversity (0.6121).

The haplotype diversity for the eight Y-chromosomal STR loci in the Antioquia, Chocó and Cartagena population samples were 0.9949, 0.9959 and 0.9940, respectively. This implies that this set of Y-specific STR loci will be valuable additions to our current test panel and will improve the exclusion probabilities in forensic and kinship cases when individuals from Antioquia, Chocó or Cartagena are involved.

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