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Discrimination index of Y-chromosomal haplotypes in an Antioquia (Colombia) population sample

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Abstract. Y-chromosome STR haplotypes (DYS19, DYS385, DYS389I, DYS389II, DYS390, DYS391, DYS392 and DYS393) in a sample of 507 healthy male blood donors from Antioquia (Colombia) were studied. 323 different haplotypes were found, 256 of them being unique. Gene diversity ranged between 0.3640 at DYS393 and 0.8900 at DYS385. The haplotype diversity value (power of discrimination (PD) and power of exclusion (PE)) calculated from all eight loci combined was 0.9946, which is informative. This study provides further information on the Y-chromosome polymorphisms commonly used in paternity testing, forensic genetics and in the population genetic studies of complex patterns of sex differential racial admixture.

Keywords: Antioquia; Colombia; DNA; Y-chromosome; STR; Population

1. Introduction

The Y-chromosome nonrecombinant portion represents a paternally inherited haploid transmission pattern [1]. Because of that Y-STRs can be used to construct highly discriminative Y haplotypes which are useful in stain analysis [2], paternity testing (lineage cases with male offspring) [1–3], and in discriminating complex differential sex patterns of admixture in population genetic studies as the one described in the population inhabiting the Antioquian State in Colombia, South America [4].

Here we report gene frequencies, gene and haplotype diversity for nine Y-STR loci in the Antioquia (Colombia) population. These will increase the database and the knowledge of polymorphisms on Colombian populations, particularly on the Antioquian communities (Paisa community).

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2. Materials and methods

Samples of 507 unrelated males were obtained from healthy individuals from Antioquia (Colombia) and from routine paternity cases (fathers). Genomic DNA was extracted by a salt precipitation extraction procedure [5]. The loci DYS389 I/II (0.1 μM) and DYS390 (0.6 μM) were amplified in a triplex PCR reaction, the loci DYS19 (0.2 μM), DYS391 (0.05 μM), DYS392 (0.4 μM) and DYS393 (0.2 μM) in quadruplex PCR reaction and the DYS385 (0.1 μM) as a singleplex.

PCR reactions were carried out in 15 μl and consisted of 20 ng of genomic DNA, 0.5 U Taq DNA Polymerase (Promega), 200 μM dNTPs, 1.5 μl 10 × Buffer, 2.0 mM $MgCl_2$. Samples were amplified with 30 cycles of 95 °C for 30 s, 53 °C for 1 min and 72 °C for 20 s. The primer sequences for DYS389 I/II, DYS390, DYS391, DYS392 and DYS393 are described in Kayser et al. [6], for DYS385 in Schneider et al. [7] and DYS19 modified from Szibor et al. [8].

Detection of the amplified products was conducted by electrophoresis on a 4% denaturing gel containing 8 M urea. Alleles were visualized by silver staining (Promega) and they 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 and ISFG recommendations on forensic analysis using Y-chromosome STRs. The gene frequencies and gene or haplotype diversity

Table 1
Allele frequencies and gene diversity value of seven Y-chromosome STR loci ($n = 507$)

Allele	DYS19	DYS389 I	DYS389 II	DYS390	DYS391	DYS392	DYS393
6					0.002		
8					0.010		
9					0.041		
10		0.002			0.467	0.004	0.002
11		0.002			0.456	0.321	0.006
12		0.122			0.024	0.075	0.105
13	0.170	0.637				0.485	0.787
14	0.594	0.227				0.095	0.083
15	0.152	0.006				0.016	0.018
16	0.061					0.004	
17	0.022	0.004					
18	0.002						
20				0.004			
21				0.083			
22				0.069			
23				0.229			
24				0.479			
25				0.108			
26			0.004	0.026			
27			0.034				
28			0.118	0.002			
29			0.357				
30			0.314				
31			0.130				
32			0.036				
33			0.008				
GD	0.593	0.530	0.744	0.697	0.574	0.649	0.364
SE	0.020	0.020	0.010	0.016	0.010	0.014	0.026

GD: gene diversity. SE: standard error.

Table 2
Allele haplotype frequencies and gene diversity value at the Y-chromosome STR locus DYS385 ($n = 507$)

Genotype	Frequency	Genotype	Frequency	Genotype	Frequency	Genotype	Frequency
9/14	0.012	12/14	0.041	14/14	0.030	51/21	0.004
9/16	0.002	12/15	0.020	14/15	0.034	16/16	0.006
10/14	0.028	12/16	0.004	14/16	0.026	16/17	0.028
11/11	0.006	12/17	0.006	14/17	0.016	16/18	0.016
11/12	0.018	12/19	0.002	14/18	0.012	16/19	0.012
11/13	0.030	13/14	0.039	14/19	0.012	17/17	0.024
11/14	0.304	13/15	0.041	15/15	0.032	17/18	0.020
11/15	0.063	13/16	0.008	15/16	0.018	17/19	0.004
11/16	0.008	13/17	0.020	15/17	0.010	18/18	0.006
11/18	0.012	13/18	0.012	15/18	0.006	18/19	0.002
12/12	0.002	13/19	0.002	15/19	0.006	18/21	0.002

Gene diversity value: 0.890. Standard error: 0.008.

values were calculated using the software ARLEQUIN version 2000 [9] and Nei formulation [10].

3. Results and discussion

Allele frequencies of the systems and gene diversity values are shown in Tables 1 and 2. The highest diversity value in this study was found at the locus DYS385 (0.8900), followed by the DYS389 II (0.744). The gene and haplotype diversity have the same value as the power of discrimination (PD) [11] and chance of exclusion (CE) [12]. The nine STRs described in this study result in informative Y-haplotypes with CE and PD values of 0.9946. We identified 323 different haplotypes and 256 were seen only once. The most frequent haplotype was present in 20 of 507 males. Development of Y-chromosome specific polymorphisms will be of great benefit in analyzing mixed DNA samples, in investigating sexual assaults as well as in paternity testing where the alleged father is not available but other patrilineal relatives are.

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