



Y-STR diversity and ethnic admixture in White and Mulatto Brazilian population samples

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Abstract

We investigated 50 Mulatto and 120 White Brazilians for the Y-chromosome short tandem repeat (Y-STR) markers (DYS19, DYS390, DYS391, DYS392 and DYS393) and found 79 different haplotypes in the White and 35 in the Mulatto sample. Admixture estimates based on allele frequencies showed that the admixture of the white sample was 89% European, 6% African and 5% Amerindian while the Mulatto sample was 93% European and 7% African. Results were consistent with historical records of the directional mating between European males and Amerindian or African females.

Key words: Brazil, Mulatto, White, Y-STR.

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The Brazilian population is a result of interethnic crosses of Europeans, Africans and Amerindians, and is one of the most heterogeneous populations in the world. When the first European colonizers arrived (1500 AD), 1-5 million Amerindians already lived in the region that now is known as Brazil (Salzano and Callegari-Jacques, 1988). Before 1820, European colonization was almost exclusively composed of Portuguese while between 1820 and 1975 the great majority of immigrants were from Portugal and Italy, followed by a small number by people from Spain, Germany, Syria and Japan (Carvalho-Silva *et al.*, 2001). Between the 16th and 19th centuries approximately 3.5 million Africans were brought as slaves to Brazil, coming mainly from West, West-Central and Southeast Africa (Curtin, 1969). The colonization of Brazil involved mostly European men, many of whom produced children with Amerindian and African females.

Although the classification of races is wrong from genetic standpoint (Templeton, 1998), Brazilians are classified for census purposes based on color. According to the last Brazilian government census of the 170 million Brazilians, 84 million were males, of which 52% were White, 39% were Brown, 6% were Black and 3% were classified in other categories (IBGE, 2000). Mulatto is the term commonly used in Brazil to designate the offspring result from

the union of White and Black people. We used five Y-chromosome short tandem repeat (Y-STR) markers, recognized as good markers for population studies, to investigate genetic polymorphism and ethnic admixture in White and Mulatto Brazilian population samples.

We investigated 170 healthy, unrelated, individuals seeking paternity investigation at the Ribeirão Preto University Hospital, in the city of Ribeirão Preto, São Paulo state, Southeastern Brazil. The race of the individuals in the sample was determined based on their biomedical records, 120 individuals being White and 50 Mulatto, from Ribeirão Preto and the surrounding towns.

We assessed the Y-STR loci DYS19, DYS390, DYS391, DYS392 and DYS393 (Kayser *et al.*, 1997) using blood samples taken from the individuals in our sample, DNA being extracted by an adaptation of the protocol of Higuchi (1989). The PCR reactions were performed according to the conditions described by Kayser *et al.* (1997) and products separated using 10% denaturing polyacrylamide gel electrophoresis and visualized by silver staining (Sanguinetti *et al.*, 1994). Allele designations were determined by comparison of the sample fragments with those of control DNA samples previously cloned and sequenced.

Allele and haplotype frequencies were estimated by the gene counting method and gene and haplotype diversities calculated using the ARLEQUIN software version 2.000 (Schneider *et al.*, 2000). Admixture proportions, based on allelic frequencies, were calculated by the gene identity method (Chakraborty, 1975) using the ADMIX3

program. Parental populations used in this study were from Portugal (González-Neira *et al.*, 2000; Carvalho *et al.*, 2003), Italy (Biondo *et al.*, 1998), Guinea-Bissau (Rosa *et al.*, 2006), Angola (Corte-Real *et al.*, 2000), Mozambique (Pereira *et al.*, 2002) and South American Indians (Rodríguez-Delfin *et al.*, 1997; Tarazona-Santos *et al.*, 2001). This study was approved by the Research Ethics Committee of the Ribeirão Preto University Hospital, Ribeirão Preto School of Medicine, University of São Paulo.

New alleles were not found, the frequencies of those found being shown in Table 1. A total of 79 (62 unique) different haplotypes were identified in the White and 35 (27 unique) in the Mulatto sample. The haplotype diversity was 0.9822 ± 0.0053 in the White and 0.9747 ± 0.0118 in the Mulatto sample. The most frequent haplotype 14-24-11-

13-13 (DYS19-DYS390-DYS391-DYS392-DYS393) was encountered in 11 (9%) White and 6 (17%) of Mulatto individuals. The next most frequent haplotype (14-24-10-13-13) differed from the most frequent haplotype by only a single DYS391 repeat and was shared by 9 (7%) White and 5 (14%) Mulatto individuals. Together, the two most frequent haplotypes accounted for 26% of the White and 31% of the Mulatto sample.

The Mulatto population samples analyzed for paternal lineage in the present study showed 0.93 ± 0.01 of European and 0.07 ± 0.01 of African component (Table 2). The European contribution was different to that verified by HLA polymorphisms in the State of Paraná, where the relative contribution was 0.42 ± 0.06 (Probst *et al.*, 2000). As observed in Mulattos, previous studies with White Brazil-

Table 1 - Allele frequencies for five Y-STR *loci* in White and Mulatto samples from the State of São Paulo, Brazil.

Alleles	DYS19		DYS390		DYS391		DYS392		DYS393	
	W	M	W	M	W	M	W	M	W	M
9					0.116	0.020				
10					0.576	0.620	0.083	0.040		
11					0.292	0.340	0.376	0.340		
12					0.016	0.020	0.050	0.020	0.158	0.140
13	0.175	0.140					0.433	0.500	0.676	0.780
14	0.548	0.520					0.033	0.080	0.141	0.080
15	0.215	0.240					0.025	0.020	0.025	
16	0.042	0.080								
17	0.020	0.020								
21			0.066	0.060						
22			0.100	0.240						
23			0.210	0.160						
24			0.517	0.420						
25			0.008	0.040						
26			0.066	0.080						
27			0.033							
n	120	50	120	50	120	50	120	50	120	50

W: White; M: Mulatto; n: number of individuals.

Table 2 - Parental contribution in Brazilian samples classified as White and Mulatto.

Identification	Populations	Genetic marker	Parental contribution			Ref
			European	African	Amerindian	
White	Brazil	Y-chromosome UEPs	0.97 ± 0.00	0.03 ± 0.00	0.00 ± 0.00	1
	São Paulo (southeastern Brazil)	Y-chromosome STRs	0.89 ± 0.01	0.06 ± 0.01	0.05 ± 0.01	Ps
	Rio Grande do Sul (southern Brazil)	Y-chromosome UEPs	1.00 ± 0.00	0.00 ± 0.00	0.00 ± 0.00	2
	Paraná (southern Brazil)	HLA polymorphisms	0.81 ± 0.07	0.12 ± 0.06	0.07 ± 0.01	3
	Brazil	mtDNA	0.39 ± 0.00	0.28 ± 0.00	0.33 ± 0.00	4
	Rio Grande do Sul (southern Brazil)	mtDNA	0.48 ± 0.00	0.16 ± 0.00	0.36 ± 0.00	2
Mulatto	São Paulo (southeastern Brazil)	Y-chromosome STRs	0.93 ± 0.01	0.07 ± 0.01	0.00 ± 0.00	Ps
	Paraná (southern Brazil)	HLA polymorphisms	0.42 ± 0.06	0.49 ± 0.06	0.09 ± 0.01	3

Ref: References; Ps: Present study; 1- Carvalho-Silva *et al.*, 2001; 2- Marrero *et al.*, 2005; 3- Probst *et al.*, 2000; 4- Alves-Silva *et al.*, 2000.

ian populations have demonstrated that the degree of admixture varied depending on the genetic marker used. While the Y-chromosome lineages in White samples were almost exclusively from European origin (Carvalho-Silva *et al.*, 2001), mtDNA showed similar European, African and Amerindian contributions (Alves-Silva *et al.*, 2000). In our study, the admixture estimate for the White sample was 0.89 ± 0.01 European, 0.06 ± 0.01 African and 0.05 ± 0.01 Amerindian. As shown in Table 2, the high European component is similar to that found in the Brazilian Y-chromosome study carried out by Carvalho-Silva *et al.* (2001) and in the study by Marrero *et al.* (2005) who investigated a population sample from the southernmost Brazilian state of Rio Grande do Sul which has a predominantly White population, but greater than that verified with HLA polymorphisms (Probst *et al.*, 2000).

Ethnic admixture based on five Y-STR *loci* in the present study showed a high predominance of the European component in the White and Mulatto samples. These results are consistent with historical records of the directional mating between European males and Amerindian or African females during the colonization of Brazil.

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