



## Y-chromosome DNA haplotypes in human samples from Bahia, Brazil

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### Abstract

*Background:* The Y-chromosome polymorphisms have a great value to population genetic studies and evolutionary aspects since these markers show high levels of heterogeneity within and between populations, permitting the geographical distribution and ancestry of patrilineages to be evaluated. They complement the autosomal STRs and mtDNA matrilineage information. Y-linked STRs remain stable in given paternal lineages over many generations so they are useful for identification in forensic cases, namely in criminal stain with male/female contributions. *Methods:* In this study, we report Y-chromosome haplotypes in a population sample with 68 unrelated males blood donors from Bahia state (Northeastern Brazil). The Y-STRs (DYS19, DYS389 I/II, DYS390 and DYS393) were determined after PCR pentaplexing with an automatic ABI sequencer. *Results:* In the Bahia population, we observed 47 different Y-chromosome haplotypes, of which 37 Y-haplotypes were unique. Only two Y-types were present in more than two persons. The most significant values of gene diversity were found in the markers DYS390, DYS389 II and DYS19 in decreasing order. The haplotype diversity value of the population studied was of 96.62. *Conclusion:* Comparing this Brazilian population with other Brazilian, Portuguese and African populations, we observed that the Bahia population has a similar haplotype diversity value to other Brazilian and Portuguese populations.

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*Keywords:* Y-STRs; Brazil population; Gene diversity; Y-chromosome haplotype diversity

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Table 1  
Haplotypes and haplotypic frequency of the population studied

Haplotypes	DYS19	DYS389 I	DYS389 II	DYS390	DYS393	Observed number	Haplotype frequency
H1	13	10	17	13	23	1	0.01
H2	13	10	18	13	23	1	0.01
H3	13	10	18	14	24	1	0.01
H4	13	10	19	12	24	1	0.01
H5	13	11	16	13	24	1	0.01
H6	13	11	19	12	24	1	0.01
H7	13	9	17	13	24	1	0.01
H8	14	10	16	13	23	5	0.07
H9	14	10	16	12	24	2	0.03
H10	14	10	16	13	24	10	0.15
H11	14	10	16	13	25	1	0.01
H12	14	10	17	12	21	1	0.01
H13	14	10	17	12	22	1	0.01
H14	14	10	17	12	23	1	0.01
H15	14	10	17	13	24	2	0.03
H16	14	11	15	12	24	1	0.01
H17	14	11	16	13	24	1	0.01
H18	14	11	17	15	21	1	0.01
H19	14	9	16	13	23	1	0.01
H20	14	9	16	13	24	2	0.03
H21	14	9	16	13	25	1	0.01
H22	14	9	17	12	22	1	0.01
H23	14	9	17	13	24	1	0.01
H24	15	10	15	12	24	1	0.01
H25	15	10	16	12	22	1	0.01
H26	15	10	16	13	22	1	0.01
H27	15	10	16	12	23	2	0.03
H28	15	10	16	13	23	2	0.03
H29	15	10	16	13	24	2	0.03
H30	15	10	17	14	21	1	0.01
H31	15	10	17	14	22	1	0.01
H32	15	10	17	13	23	1	0.01
H33	15	10	18	13	21	1	0.01
H34	15	10	18	13	22	1	0.01
H35	15	10	19	13	21	1	0.01
H36	15	11	15	13	23	1	0.01
H37	15	11	17	13	23	1	0.01
H38	15	9	14	13	24	2	0.03
H39	15	9	16	12	24	2	0.03
H40	15	9	16	13	24	1	0.01
H41	15	9	17	14	22	1	0.01
H42	15	9	17	12	23	1	0.01
H43	15	9	18	13	22	1	0.01
H44	16	10	16	14	23	1	0.01
H45	16	10	17	13	22	1	0.01
H46	16	11	17	12	24	1	0.01
H47	16	11	18	13	21	1	0.01

Table 2  
Number of alleles, gene frequency and diversity

	DYS19				DYS389I			DYS389II				DYS390				DYS393						
Alleles	13	14	15	16	9	10	11	14	15	16	17	18	19	21	22	23	24	25	12	13	14	15
Individuals number	7	32	25	4	15	43	10	2	3	36	18	6	3	6	9	17	33	3	17	45	5	1
Gene frequency	0.103	0.471	0.368	0.059	0.221	0.632	0.147	0.029	0.044	0.530	0.265	0.088	0.044	0.088	0.132	0.250	0.485	0.044	0.250	0.662	0.074	0.015
Different alleles number	4				3			6				5				4						
Gene diversity	63.90				53.80			64.66				68.50				50.01						

## 1. Introduction

The Brazilian population is composed of individuals from different ethnic origins varying according to the geographic region. In Bahia state (Northeastern Brazil), the contribution of three groups is evident: Amerindian, European (namely Portuguese) and African, especially from the Northwest region. In this work, we show the study of the Y-chromosome haplotypes in a sample population from Bahia and the comparative results with other populations.

## 2. Material and methods

Bloodstains obtained from 68 unrelated voluntary donors were submitted at Chelex 100 [1,2]. The samples, after purification, were quantified using Human DNA Quantitation kit QuantiBlot™ [3].

The loci DYS19, DYS389 I, DYS389 II, DYS390, DYS393 were amplified in a PCR pentaplex format using a Perkin Elmer 9600 thermal cyclor according to Gusmão et al. [4].

The separation was carried out by capillary electrophoresis on an ABI PRISM 310 Genetic Analyser instrument performed in Performance Optimised Polymer sieving medium (POP4™; 1-ml syringe).

The fragment size was collected using ABI PRISM Collection Software application, with the module GS POP4 and GeneScan™ 500 (Rox) internal lane size standard.

Table 3  
Comparison of the gene and haplotype diversity between populations

Y-STRs	Gene diversity					Individuals number	Haplotypes number	Haplotype diversity
	DYS19	DYS389I	DYS389II	DYS390	DYS393			
Bahia/Brazil	63.90	53.80	64.66	68.50	50.10	68	47	96.62
São Paulo/Brazil [9]	58.39	56.74	55.95	55.01	46.62	53	33	97.17
Amazonas/Brazil [9]	65.75	47.54	63.07	68.96	29.11	42	32	98.37
Portuguese Population [10]	59.61	50.58	47.34	66.56	51.82	119	63	96.43
Northwest African Population [11]	44.80	49.60	51.45	63.70	28.70	185	56	87.61

Table 4  
Matrix of genetic distances

	Bahia/ Brazil	Amazonas/ Brazil	S. Paulo/ Brazil	Northwest Africa	Portugal
Bahia/Brazil	–	0.0380	0.0269	0.2750	0.2837
Amazonas/Brazil	0.0380	–	0.0571	0.1893	0.3068
S. Paulo/Brazil	0.0269	0.0571	–	0.2629	0.2516
Northwest Africa	0.2750	0.1893	0.2629	–	0.4315
Portugal	0.2837	0.3068	0.2516	0.4315	–

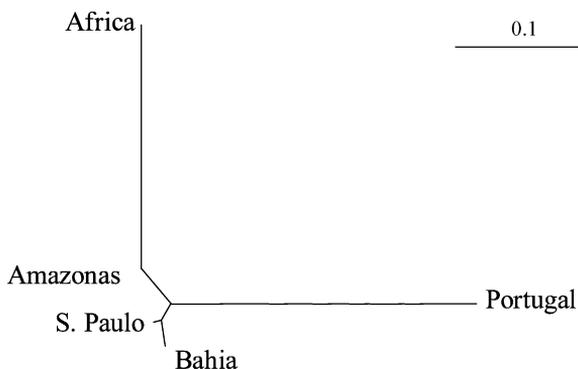


Fig. 1. Phylogenetic tree of the populations from Brazil, Portugal and Northwest Africa.

The repeat nomenclature follows that of Kayser et al. [5], Rolf et al. [6] and Gill et al. [7].

Allele and haplotype frequencies were estimated by gene counting. Gene and haplotype diversity were computed for each locus according to Nei [8].

### 3. Results

In the 68 males from Bahia, we observed 47 different Y-chromosome haplotypes, of which 37 Y-haplotypes were unique. Only two Y-types were present in more than two persons (Table 1). The most significant values of gene diversity were found in the markers DYS390, DYS389 II and DYS19 in decreasing order (Table 2). The haplotype diversity value of the population studied was of 96.62 (Table 3). The comparative studies are represented in Tables 3 and 4 and Fig. 1.

### 4. Discussion

Comparing this Brazilian population with other Brazilian, Portuguese and African populations, we observed that the Bahia population has a similar haplotype diversity value to other Brazilian and Portuguese populations, but a higher value when comparing with the Northwest African populations.

As expected, the genetic distances were lower between Brazilian populations. These results confirm the importance of the Y-STRs, being highly polymorphic markers and informative of the relationships between populations.

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