

FOR THE RECORD

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A Genetic Population Study of Six Y STRS (Y-Plextm6) in an Uruguayan Sample and Its Comparison with Other Populations

POPULATION: 91 unrelated males from Uruguay (South America)

KEYWORDS: forensic science, DNA typing, population genetics, Y chromosome, Uruguay, DYS393, DYS19, DYS389II, DYS390, DYS391

Genomic DNA was extracted from blood samples of 91 unrelated Uruguayan males that were initially spotted on FTA^R paper (Whatman, Clifton, NJ). The six loci (DYS393, DYS19, DYS389II, DYS390, DYS391, and DYS385) were amplified using the Y-PLEXTM6 kit (Reliagene Technologies, Inc., New Orleans, LA) according to the manufacturer's recommendations and to published experiences (1). The amplified products were analysed by capillary electrophoresis using the ABI Prism 310 DNA Sequencer (PE Biosystems, Foster City, CA). Allele assignment was performed by comparison with self-established allelic ladder, positive and negative controls included in the kit.

Several external populations were used for comparison from previously published papers; these are samples from Southern Spain and the Spanish Canary Islands (2), Northern Argentinian Amerindians (3), Italians (4), Mozambique (5), Caucasian Argentinians (6), Brazilians (5), and from Portugal (7). Since duplicated loci such as DYS385 cannot be dealt with in AMOVA, the study had to be confined to the systems DYS393, DYS19, DYS389II, DYS390, DYS391. STR allele frequencies, diverse Y-chromosome diversity indices and population differentiation were tested using software ARLEQUIN version 2.000 (8).

The allele frequencies and gene diversity of the STR's DYS393, DYS19, DYS389II, DYS390, DYS391 y DYS385 from Uruguay are shown in Table 1. We counted 76 different haplotypes in a population sample of 91 Uruguayan males giving a haplotype diversity

of 0.9946. This combination is sufficient to distinguish the involved male stain donors. For AMOVA, populations were pooled to form six groups (taking in consideration previous results of sample differentiation): 1. Uruguay, Argentina and Brazil, 2. Peninsular Spain and Spanish Canary Islands, 3. Northern Argentina Amerindians, 4. Mozambique, 5. Portugal, and 6. Italy.

TABLE 1—Allele frequencies obtained for DYS393, DYS19, DYS389II, DYS390, DYS391 and DYS385 in Uruguay.

Allele number	DYS393	DYS19	DYS389II	DYS390	DYS391	DYS385I
9					0,07692	
10					0,45055	0,02198
11					0,43956	0,24725
12	0,16484				0,03297	0,0989
13	0,71429	0,08791				0,12088
14	0,12088	0,7033				0,30769
15		0,12088				0,07692
16		0,07692				0,06593
>16		0,01099				0,02198
17						0,03297
18						0,00549
19						
20						
21						
22				0,06593		
23				0,26374		
24				0,53846		
25				0,12088		
>25				0,01099		
26						
27						
28			0,14286			
29			0,42857			
30			0,32967			
31			0,0989			

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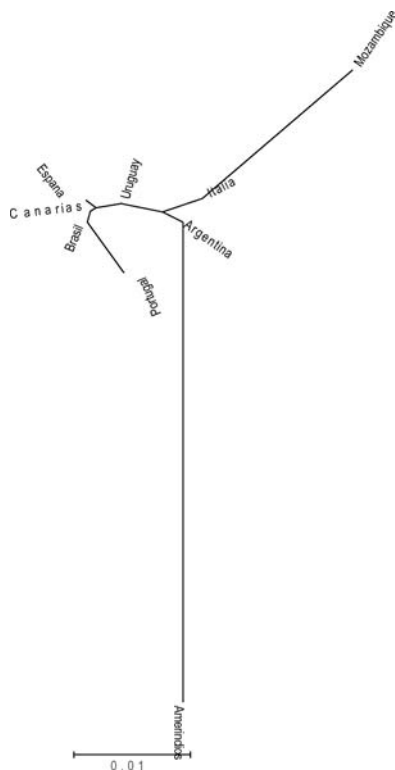
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TABLE 2—Exact test of sample differentiation between populations (below the diagonal) and their significance, positive or negative (above the diagonal).

	Uruguay	Portugal	Mozambique	Italia	Spain (Peninsula)	Spain CanaryIslands	Argentina	Amerindians	Brasil
Uruguay		+	+	−	+	−	−	+	−
Portugal	0,00000		+	+	+	+	+	+	+
Mozambique	0,00000	0,00000		+	+	+	+	+	+
Italia	0,06285	0,00000	0,00000		+	+	+	+	+
Spain (peninsula)	0,03370	0,00000	0,00000	0,00000		−	−	+	+
Spain (Canary Is.)	0,08420	0,00000	0,00000	0,00000	0,20755		−	+	−
Argentina	0,16030	0,00000	0,00000	0,01555	0,13430	0,35490		+	−
Argentina (Amerindians)	0,00000	0,00000	0,00000	0,00000	0,00000	0,00000	0,00000		+
Brasil	0,19315	0,00000	0,00000	0,01095	0,00000	0,26360	0,31535	0,00000	

FIG. 1—NJ tree based on F_{ST} distances connecting the 9 population samples.

Using AMOVA it was estimated the relative contribution to the total observed genetic variance of: (i) the genetic variance between groups, (ii) the genetic variance between populations within groups, and (iii) the genetic variance between individuals within populations (Table 3). Two percent of genetic variation was found to be between populations, while the remaining 98% was found within populations.

Phylogenetic analysis: the unrooted NJ tree, based on pairwise F_{st} values (see Fig. 1), shows the Amerindians to one edge while Mozambique is placed at the opposite end. Uruguay has an intermediate position between the rest of the population groups.

The complete dataset is available to any interested researcher upon request to the contacting author.

TABLE 3—AMOVA in 9 populations based on five Y-SYR loci.

Source of Molecular Variation	ϕ_{ST} Statistic	
	Variance (%)	P
Among groups	2.03	<0.00001
Among populations within groups	0.13	<0.00001
Within populations	97.85	<0.00001

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