

Blood protein genetic markers in a Northeastern Uruguayan population

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ABSTRACT

A total of 20 blood genetic systems (23 loci) was studied in a sample of 127 individuals from the town of Tacuarembó, Uruguay. Generally the frequencies observed are compatible with a mainly Caucasian population, but the presence of some markers (*FY*null*, *DI*a*, *G6PD*A*, *PGM2*2*), as well as the frequencies of some alleles (*RH*cde*, *GLO1*2*, *HP*1*, *HLA*A2*) suggest the presence of African and Amerindian derived genes. Differences in relation to allele frequencies previously obtained in Montevideo were also noted.

INTRODUCTION

Latin American populations have been fairly well-studied in relation to genetic markers (general reviews in Salzano, 1971; Lisker, 1981; Roychoudhury and Nei, 1988). But this has not been true for Uruguay. Until recently few studies had been made there, using a very limited number of loci. We therefore decided to start a long-term investigation aiming at the description and understanding of the factors responsible for the genetic variability of Uruguayan groups. Using blood polymorphisms we have recently described the characteristics of the population of Montevideo and surrounding regions (Sans *et al.*, 1993, 1994; Alvarez *et al.*, 1993).

The present investigation extends these studies to a northeastern community, Tacuarembó.

MATERIAL AND METHODS

The Department of Tacuarembó is situated at the northeast of the Republic. The 1985 census listed a population of 82,809 persons living there, and about half of them inhabit the Departmental capital which has the same name. Historically the region had strategic value due to its proximity with the Brazilian-Portuguese Empire. Its first village, named San Fructuoso de Tacuarembó, was founded in 1832, the Department being delimited in 1837 (Michoelsson, 1981).

The area was occupied at the time of the European conquest by Charrua Indians; between the 17th and 19th century Guarani Indians also arrived there, migrating from the Jesuitic Missions. The European and African components of the population came not only from other regions of the Republic, but also from Brazil. Therefore, both Spanish and Portuguese-

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derived persons peopled the area, carrying with them their captives of African descent (Pi and Vidart, 1969; Acosta y Lara, 1985; Gonzalez Risotto and Rodriguez Varese, 1989; general review in Sans, 1994).

Historical, social and demographic data assembled by Sans (1994) indicate that Tacuarembó differs markedly from Montevideo in a series of characteristics. Mobility is quite low (average individual migration, 1880-1990: 62 km; marital distance, same period, 48 km), and inbreeding reached its highest value ($F = 26 \times 10^{-4}$) in 1900-1919.

The samples were collected at Tacuarembó's Regional Hospital from puerperal women and refrigerated shortly after collection. They were then transported to Montevideo, where blood group and HLA typing was performed as indicated in Sans *et al.* (1993) and Alvarez *et al.* (1993). HLA reagents were those furnished by the XI International Workshop, held in Japan, 1991. Aliquots of the samples were afterwards brought to Porto Alegre, where the serum protein and red cell enzyme determinations were performed according to techniques listed in Bortolini *et al.* (1992).

Blood group, serum protein and red cell enzyme allele frequencies were estimated by the MAXLIK program (Reed and Schull, 1968), while for HLA we used a computer program kindly made available to us by Prof. R. Chakraborty (University of Texas, Houston).

RESULTS AND DISCUSSION

The results concerning 20 systems (23 loci) are presented in Tables I and II. Generally the frequencies are compatible with a mainly Caucasian population, but there are some exceptions (for instance, the presence of *FY*null*, *DI*a*, *G6PD*A* and *PGM2*2*). The blood group prevalences are similar to those found in Montevideo (Sans *et al.*, 1993) except for the MNSs system (*L*Ms*, Tacuarembó: 42%; Montevideo: 23%; *L*Ns*: 41% and 51%, respectively). The frequency of *RH*cde* is also lower in Tacuarembó (27% vs 36% in Montevideo), suggesting non-Caucasian influences in this community (for pertinent interethnic frequency comparisons see Bortolini *et al.*, 1992).

No data for the electrophoretic systems are available for Montevideo; we therefore compared them with the Caucasian portion of another large South American city (Porto Alegre; Franco *et al.*, 1981; Silva *et al.*, 1981; Weimer *et al.*, 1981; unpublished) situated not too far away from Tacuarembó. Again, differences were not marked, but the prevalences of *GLO1*2* and *HP*1* are increased (Tacuarembó, *GLO1*2*: 67%; *HP*1*: 51%. Porto Alegre, 58% and 41%, respectively), sug-

Table I - Phenotype and allele distributions for 20 genetic systems observed in Tacuarembó, Uruguay.

Systems	Phenotypes	No. of indiv.	Alleles or haplotypes	Frequencies	
ABO	O	70	<i>I*O</i>	0.738	
	A	35	<i>I*A</i>	0.172	
	B	17	<i>I*B</i>	0.090	
	AB	5			
MNSs	MS	3	<i>L*MS</i>	0.126	
	MSs	4	<i>L*Ms</i>	0.424	
	Ms	9	<i>L*NS</i>	0.044	
	MNS	5	<i>L*Ns</i>	0.406	
	MNSs	17			
	MNs	66			
	Ns	5			
Rh	CDEe	1	<i>RH*CDE</i>	0.005	
	CDe	30	<i>RH*CDe</i>	0.431	
	CcDEe	18	<i>RH*cDE</i>	0.202	
	CcDe	30	<i>RH*cdE</i>	0.011	
	cDE	6	<i>RH*cde</i>	0.269	
	cDEe	23			
	cDe	7			
	cdEe	1			
	cde	10			
	Kell	K	1	<i>KELL*k</i>	0.987
Kk		1			
k		117			
Duffy	a+b-	23	<i>Fy*a</i>	0.365	
	a+a+	44	<i>Fy*b</i>	0.517	
	a-b+	42	<i>Fy*null</i>	0.118	
	a-b-	2			
Kidd	a+b-	34			
	a+b+	38	<i>JK*b</i>	0.523	
	a-b+	39			
Diego	a+	1	<i>DI*b</i>	0.995	
	a-	104			
Hemoglobin	A	76	<i>HB*A</i>	1.000	
	Glucose-6-phosphate dehydrogenase	A	1 ^a		
		AB	3 ^a	<i>G6PD*B</i>	0.974
	B	92 ^a			
Phosphogluconate dehydrogenase	A	97	<i>PGD*A</i>	0.980	
	AC	5			
Phosphoglucomutase 1	1-1	59	<i>PGM1*1</i>	0.743	
	2-1	35			
	2-2	9			
Phosphoglucomutase 2	1-1	102	<i>PGM2*1</i>	0.995	
	2-1	1			

Continued

Table I - Continued.

Systems	Phenotypes	No. of indiv.	Alleles or haplotypes	Frequencies
Adenylate kinase	1-1	101	<i>AK*1</i>	0.990
	2-1	2		
Acid phosphatase	A	5	<i>ACP*A</i>	0.257
	AB	41		
	AC	2		
	B	49		
	BC	6		
Esterase D	1-1	71	<i>ESD*1</i>	0.825
	2-1	28		
	2-2	4		
Glyoxalase 1	1-1	17	<i>GLO1*2</i>	0.671
	2-1	35		
	2-2	53		
Haptoglobin	1-1	29	<i>HP*1</i>	0.512
	2-1	72		
	2-2	26		
Transferrin	C	127	<i>TF*C</i>	1.000
Albumin	A	127	<i>ALB*A</i>	1.000
Ceruloplasmin	B	127	<i>CP*B</i>	1.000

^aFemales only.

gesting admixture with people of either African or Amerindian extraction (for estimated South American parental gene frequencies consult Bortolini *et al.*, 1992).

As for the HLA prevalences (Table II), there is an overall similarity with the Montevideo frequencies (Alvarez *et al.*, 1993). The exception is the low prevalence of *HLA*A2* (Tacuarembó: 13.4%; Montevideo: 24.1%), which could indicate African-derived admixture in the former (reference tables in Imanishi *et al.*, 1992).

Summing up, while Tucuairembó shows a predominantly Caucasian genetic pattern, it also presents indications of non-Caucasian admixture. As expected from the historical and socio-demographic information, there are differences in relation to the Montevideo results. In an effort to further characterize this north-eastern Uruguayan population we collected a small subsample (49 individuals unevenly studied for the several markers) of this population (both at the hospital and in home visits) selected for alleged non-Caucasian (mainly Indian) admixture. Information concerning this subsample can be found in Sans (1994), but the differences observed between it and the random sample

Table II - Phenotype and allele frequencies for the HLA system observed in Tacuarembó, Uruguay.

Locus and no. studied	Antigen (splits in parentheses)	No. observed	Allele	Frequency (%)
HLA-A (N=56)	1	3	<i>HLA*A1</i>	2.7
	2	14	<i>HLA*A2</i>	13.4
	3	12	<i>HLA*A3</i>	11.3
	9 ^a	3	-	-
	11 ^a	2	<i>HLA*A11</i>	6.4
	11 (11.1)	5	-	-
	9 (23)	2	<i>HLA*A23</i>	1.8
	9 (24)	8	<i>HLA*A24</i>	7.4
	10 (25)	1	<i>HLA*A25</i>	0.9
	10 (26)	2	<i>HLA*A26</i>	1.8
	28 ^a	7	-	-
	28 (68)	3	<i>HLA*A68</i>	2.7
	28 (69)	3	<i>HLA*A69</i>	2.7
	19 (29)	2	<i>HLA*A29</i>	1.8
	19 (30)	3	<i>HLA*A30</i>	2.7
	19 (31)	9	<i>HLA*A31</i>	8.4
	19 (32)	4	<i>HLA*A32</i>	3.6
	19 (33)	4	<i>HLA*A33</i>	3.6
	10 (34)	1	<i>HLA*A34</i>	0.9
36	2	<i>HLA*A36</i>	1.8	
43	2	<i>HLA*A43</i>	1.8	
Blank	20	-	-	
HLA-B (N=52)	7	2	<i>HLA*B7</i>	1.9
	8	3	<i>HLA*B8</i>	2.9
	13	2	<i>HLA*B13</i>	1.9
	14	4	<i>HLA*B14</i>	3.9
	15 ^a	4	-	-
	15 (62)	4	<i>HLA*B15</i>	3.9
	15 (63)	2	-	-
	17 ^a	1	-	-
	18	2	<i>HLA*B18</i>	1.9
	22 ^a	2	-	-
	27	2	<i>HLA*B27</i>	1.9
	35	7	<i>HLA*B35</i>	7.0
	16 (38)	2	<i>HLA*B38</i>	1.9
	16 (39)	1	<i>HLA*B39</i>	1.0
	40 ^a	1	<i>HLA*B40</i>	2.9
	40 (60)	2	-	-
	41	1	<i>HLA*B41</i>	1.0
	42	4	<i>HLA*B42</i>	3.9
	12 (44)	8	<i>HLA*B44</i>	8.0
	12 (45)	2	<i>HLA*B45</i>	1.9
	46	1	<i>HLA*B46</i>	1.0
47	1	<i>HLA*B47</i>	1.0	
48	1	<i>HLA*B48</i>	1.0	
21 (49)	1	<i>HLA*B49</i>	1.0	
21 (50)	4	<i>HLA*B50</i>	3.9	
5 (51)	2	<i>HLA*B5101</i>	1.9	
5102	5	<i>HLA*B5102</i>	4.9	
22 (54)	1	<i>HLA*B54</i>	1.0	
22 (55)	1	<i>HLA*B55</i>	1.0	
59	1	-	-	
40 (61)	1	-	-	

Continued

Table II - Continued

Locus and no. studied	Antigen (splits in parentheses)	No. observed	Allele	Frequency (%)
	70	1	-	-
	Blank	28	-	-
HLA-C (N=39)	1	1	HLA*CW1	1.3
	2	6	HLA*CW2	8.0
	3 (9)	6	HLA*CW3	14.6
	3 (10)	5	-	-
	4	10	HLA*CW4	13.7
	5	6	HLA*CW5	8.0
	6	6	HLA*CW6	8.0
	7	14	HLA*CW7	19.9
	Blank	24	-	-

^aDoes not include those with identified splits.

described here were not large enough to warrant listing in this communication. Quantitative estimates of the relative contributions of Caucasians, Africans and Amerindians to this gene pool can be made, but they depend on the methods used and on the set of frequencies assumed to represent the parental populations.

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RESUMO

Um total de 20 sistemas genéticos que se expressam no sangue (23 loci) foi estudado em uma amostra de 127 indivíduos da cidade de Tacuarembó, Uruguai. Geralmente as frequências observadas são compatíveis com uma população predominantemente

caucásica, mas a presença de alguns marcadores (*FY*nu*, *Di*a*, *G6PD*A*, *PGM2*2*), assim como as frequências de alguns alelos (*RH*cde*, *GLO1*2*, *HP*1*, *HLA*A2*) sugerem a presença de genes de origem africana e ameríndia. Foram também notadas diferenças com relação a frequências alélicas previamente obtidas em Montevideo.

REFERENCES

- Acosta y Lara, E.** (1985). Salsipuedes 1831: los lugares. *Rev. Fac. Hum. Cienc., ser. Cienc. Antropol.* 1: 65-88.
- Alvarez, I., Sans, M., Toledo, R., Sosa, M., Bengochea, M. and Salzano, F.M.** (1993). HLA gene and haplotype frequencies in Uruguay. *Intern. J. Anthropol.* 8: 163-168.
- Bortolini, M.C., Weimer, T.A., Franco, M.H.L.P., Salzano, F.M., Layrisse, Z., Schneider, H., Schneider, M.P.C. and Harada, M.L.** (1992). Genetic studies in three South American Black populations. *Gene Geography* 6: 1-16.
- Franco, M.H.L.P., Salzano, F.M. and Maia de Lima, F.A.** (1981). Blood groups and serum protein types in two Brazilian populations. *Rev. Brasil. Genet.* 4: 689-704.
- Gonzalez Risotto, R. and Rodriguez Varese, S.** (1989). La importancia de las Misiones Jesuíticas en la formación de la sociedad uruguaya. *Estudios Ibero-Americanos (Pontificia Universidade Católica do Rio Grande do Sul)* 15: 191-214.
- Imanishi, T., Akaza, T., Kimura, A., Tokunaga, K. and Gojobori, T.** (1992). Allele and haplotype frequencies for HLA and complement loci in various ethnic groups. In: *HLA 1991* (Tsuji, K., Aizawa, M. and Sasazuki, T., eds.), vol. 1. Oxford University Press, Oxford, pp. 1065-1220.
- Lisker, R.** (1981). *Estructura Genética de la Población Mexicana. Aspectos Médicos y Antropológicos*. Salvat, México, pp. 158.
- Michoelsson, O.** (1981). *Tacuarembó. Un Antecedente Artiguista*. Imprenta del Ejército, Montevideo, pp. 193.
- Pi, R. and Vidart, D.** (1969). El legado de los inmigrantes. *Nuestra Tierra* 29: 1-60.
- Reed, T.E. and Schull, W.J.** (1968). A general maximum likelihood method estimation program. *Am. J. Hum. Genet.* 20: 579-580.
- Roychoudhury, A.K. and Nei, M.** (1988). *Human Polymorphic Genes. World Distribution*. Oxford University Press, Oxford, pp. 393.
- Salzano, F.M.** (1971). *The Ongoing Evolution of Latin American Populations*. Charles C. Thomas, Springfield, pp. 717.
- Sans, M.** (1994). Estudio genético e histórico de la población del Departamento de Tacuarembó, Uruguay. Doctoral Thesis, Universidad de la República, Montevideo.
- Sans, M., Sosa, M., Alvarez, I., Toledo, R., Bengochea, M. and Salzano, F.M.** (1993). Blood group frequencies and the question of race admixture in Uruguay. *Interiencia* 18: 29-32.
- Sans, M., Callegari-Jacques, S.M. and Salzano, F.M.** (1994). Genetic similarity and mate selection in Uruguay. *J. Biosoc. Sci.* 26: 285-289.
- Silva, R.S., Weimer, T.A. and Salzano, F.M.** (1981). Rare and common types of phosphoglucomutase in two Brazilian populations. *Hum. Biol.* 53: 227-238.
- Weimer, T.A., Salzano, F.M. and Hutz, M.H.** (1981). Erythrocyte isozymes and hemoglobin types in a southern Brazilian population. *J. Hum. Evol.* 10: 319-328.