Original Paper

Human Heredity

Hum Hered 2001;51:27–34

Received: June 2, 1999
Revision received: August 31, 1999
Accepted: September 3, 1999

Consanguinity in South America: Demographic Aspects

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Key Words
Parental consanguinity · South America · Social class

Abstract
A sample of 53,552 nonmalformed liveborn infants was ascertained by the Latin-American Collaborative Study of Congenital Malformations between 1967 and 1996. The mean consanguinity rate was 0.96%, with significantly higher values in Brazil and Venezuela, and lower in Argentina. Low paternal education and occupation levels were positively associated with consanguinity. First-cousin matings represented almost half of all consanguineous couples. The consanguinity was mainly of more closely related types in Brazil, while in Venezuela more remote types predominated. This could reflect differences in migration patterns and rates between these two countries.

Introduction

Population frequency and structure of parental consanguinity vary greatly among countries and cultures, their estimation being strongly dependent on ascertainment methods and analytic approaches. Thus, while the study of couples through civil or religious records is limited to certain sociocultural strata, studies based on offspring are limited to fertile matings, and the study of last names or distances is limited to indirect indicators. Therefore, while no approach is the ideal one, none is dispensable, either, and the best knowledge about the breeding structure will result from a broad vision of that given population.

Several studies on consanguinity in populations in South America have been published, based on civil [1] or church marriage records [2–5], consecutive birth series [6–8], or isonymy [9–11]. The only comparative study on population samples from different South American countries was that by Orioli et al. [7] in 1982.

The aim of this study was to further investigate the frequency and the distribution of different types of parental consanguinity, and their associated demographic factors, based on a series of consecutive normal births in South American urban populations.

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Table 1. Association between consanguinity and demographic variables

Variables		OR	CI95%	p
Time	Secular trend	0.99	0.98–1.00	0.13
Country	Argentina	0.27	0.17–0.40	<0.01
	Brazil	1.87	1.34–2.60	<0.01
	Venezuela	1.96	1.37–2.80	<0.01
Hospital	Public health system	1.16	0.91–1.47	0.21
	University affiliation	1.04	0.81–1.32	0.76
	Altitude of 2,000 m above sea-level	1.17	0.72- 1.87	0.52
Race	Latin-European	0.84	0.56–1.26	0.40
	Black	1.10	0.83–1.46	0.50
	Mestizo	1.15	0.89–1.47	0.29
Social	Low maternal educational level	0.98	0.77–1.23	0.86
	Low paternal educational level	1.72	1.35–2.17	<0.01
	Low paternal occupational level	1.29	1.05–1.57	<0.02

OR = Adjusted odds ratio; CI95% = 95% confidence intervals.

Materials and Methods

The material used in this work was obtained from 56,547 non-malformed infants, randomly ascertained by the ECLAMC (Estudio Colaborativo Latinoamericano de Malformaciones Congénitas) [12], from 1,895,243 liveborn between 1967 and 1996. Births from 110 hospitals in 53 cities of the following 9 countries were included: Argentina, Bolivia, Brazil, Colombia, Chile, Ecuador, Peru, Uruguay and Venezuela.

Following the instructions of the ECLAMC, the mothers of the infants were questioned about 50 genetic and environmental items, including consanguineous relationship with the father of the child, regardless of their marital status. In case of consanguinity, the complete pedigree was drawn, specifying the type of relationship.

A possible association between consanguinity and demographic variables was analysed by comparing consanguineous with non-consanguineous matings. Comparison was done by a logistic regression analysis for the following variables: secular trend (1-year intervals); country of residence (Argentina, Brazil, Venezuela, and the other 6, pooled due to sample size limitations); type of health system (public or private); university affiliation of the hospital; geographical altitude above sea level of the hospital (below or above 2, 000 m); ethnicity (Latin-European, Black – comprising any racial combination which includes Black –, Mestizo – comprising Native and Latin European admixture –, and others); maternal and paternal educational level: low (no education to incomplete elementary level), high (complete elementary to complete university level); paternal occupational level: low (unemployed or unskilled worker), high (skilled laborer or higher).

For this work, the cases with parental consanguinity were classified into 7 groups, according to their inbreeding coefficient (F), and to the type of relationship: F = 1/4: parent/child and brother/sister; F = 1/8: aunt/nephew, uncle/niece, half sibs, and double first cousins; F = 1/16: first cousins and half uncle/niece; F = 1/32: half cousins, and first cousins once removed; F = 1/64: second cousins; F < 1/64: more remotely related and complex-multiple matings, and NFS: consanguineous matings with unspecified relationship.

In order to determine a possible excess of male or female intermediate ancestors (those between the index couple and the common ancestors), their number was obtained in each pedigree, and compared with an expected proportion of 0.50. Observed/expected differences were tested with the χ^2 test for goodness of fit. The first-cousin couples were classified into one of the following 4 types: I (the relationship is through two fathers); II (through two mothers); III (through the husband's father and the wife's mother); and IV (through the husband's mother and the wife's father). In order to determine whether the sex of the individuals through whom the couple was related could possibly influence consanguinity, the observed ratios of the 4 types of matings were analysed as to their departures from equality (random expected proportions: 0.25). Observed/expected differences were tested with the χ^2 test for goodness of fit.

The geographical distribution of consanguineous matings was analysed by comparing observed with expected values for each country. Only those countries with a total number of matings greater than 1,000 were analysed. Differences were tested with the Z test. The consanguinity rates, types of consanguineous matings, as well as the autosomal (α) and X-linked (α') mean inbreeding coefficients were estimated by country.

For Brazil and Venezuela, the consanguineous couples were distributed according to 3 indicators of mating patterns: *intra-marital* (both members born in the same state, regardless of their place of residence); *isonymy* (the same paternal surname in both members), and *migration* (one or both members born outside their state of residence).

A critical value of 5% was employed throughout this work, as the limit of statistical significance.

Results

Demographic Variables

From the 56,547 ascertained liveborn, this study included 53,552 (94.7%), in whom presence or absence of parental consanguinity was specified.

Table 2. Consanguineous matings by inbreeding coefficient (F) and by type of relationship

F	Type of mating	Partial		Cumulative		IA	
		n	%	n	%	males	females
1/4	Father/daughter	2	0.39				
	Brother/sister	1	0.20				
	Subtotal 1/4			3	0.59		
1/8	Aunt/nephew	1	0.20				
	Uncle/niece	6	1.17				
	Half sibs	1	0.20				
	Double first cousins	2	0.39				
	Subtotal 1/8			10	1.95	12	10
1/16	First cousins I	42	8.20				
	First cousins II	82	16.82				
	First cousins III	40	7.81				
	First cousins IV	51	9.96				
	First cousins, not specified	12	2.34				
	Half uncle/niece	1	0.20				
	Subtotal 1/16			228	44.53	391	471
1/32	Half first cousins	10	1.95				
	First cousins once removed	67	13.09				
	Subtotal 1/32			77	15.04	151	179
1/64	Second cousins	91	17.77				
	Other	3	0.58				
	Subtotal 1/64			94	18.36	239	259
<1/64	Remote specified	26	5.08				
	Remote, not specified	9	1.76				
	Multiple-complex	2	0.39				
	Subtotal <1/64			37	7.23	64	83
NFS	Not further specified	63	12.30	63	12.30		
Total		512	100.00	512	100.00	857	1,002

IA = Intermediate ancestors.

There were 512 cases with recognised parental consanguinity (consanguinity rate: 0.96%). Taking into account that only nonmalformed infants were included in this work, the adjusted rate would be negligibly higher.

The logistic regression analysis showed that only country, paternal education, and paternal occupation were associated with consanguinity. Low paternal education and occupation were positively associated with consanguinity. The consanguinity rate was higher in Brazil and Venezuela and lower in Argentina. The remaining variables showed no association (table 1).

Types of Matings and Sex of Intermediate Ancestors

Table 2 shows the distribution of the different types of relationship, as well as the intermediate ancestors found in this sample, arranged by a decreasing value of F. F = 1/16 (first cousin matings) represented the largest group (about 45%). There were 3 cases with F = 1/4 (2 father/daughter and 1 brother/sister matings). F = 1/32 represented 15.04% and F = 1/64 18.36% of the sample; the rate of more remotely related matings was 7.23%. Two hundred and forty-one couples were grouped as closely related (F = 1/4, F = 1/8 or F = 1/16) (47.07%), and 208 as remotely related (F = 1/32, F = 1/64 or F < 1/64) (40.63%). In 63 cases, the type of relationship was not specified (12.30%).

Table 3. Consanguinity rates, types of consanguineous matings (CM), inbreeding coefficients (F), types of first-cousin matings (FCM), and sex of intermediate ancestors (IA), classified by country and by decreasing consanguinity rate

Country	Births	CM		F								FCM				IA, n	
		n	%	>1/16	1/16	1/32	1/64	<1/64	NFS	α	α′	I	II	III	IV	male	female
Venezuela	8,592	158	1.84	2	52	30	40	21	13	64	127	8	24	8	10	297	341
Brazil	11,558	185	1.60	4	93	28	26	9	25	68	108	18	26	16	26	295	338
Colombia	609	8	1.31	0	4	0	3	0	1	49	87	0	1	2	1	14	20
Ecuador	2,311	29	1.25	1	11	5	8	2	2	48	105	2	5	2	1	44	60
Uruguay	1,407	15	1.07	1	7	1	1	0	5	52	102	1	3	0	3	18	21
Chile	6,959	52	0.75	1	34	7	7	0	3	39	62	5	14	11	4	92	121
Peru	332	2	0.60	0	2	0	0	0	0	38	113	0	2	0	0	2	6
Bolivia	958	3	0.31	0	2	0	0	0	1	13	13	1	0	0	1	5	3
Argentina	20,826	60	0.29	4	23	6	9	5	13	11	16	7	7	1	5	90	92
Total	53,552	512	0.96	13	228	77	94	37	63	39	67	42	82	40	51	857	1,002

NFS = Consanguineous of not specified type of relationship. Autosomal (α), and X-linked (α′) mean inbreeding coefficients expressed as $\times 10^{-5}$ values.

Table 4. Consanguineous (CON) matings of close (CC), remote (RC), and unknown (UT) relationship types, in the Brazilian and Venezuelan samples

Country/state		Total	CON		CC	RC	UT
			n	%			
Brazil							
Pernambuco		264	6	2.27	5	0	1
Paraíba		634	1	0.16	1	0	0
São Paulo		6,175	126	2.04	68	47	11
Rio de Janeiro		128	4	3.13	2	2	0
Minas Gerais		102	0	0.00	0	0	0
Santa Catarina		1,151	11	0.96	4	4	3
Paraná		168	3	1.79	1	1	1
Rio Grande do Sul		2,936	34	1.16	16	9	9
Total		11,558	185	1.60	97	63	25
Homogeneity	χ^2		26.26		2.82		
	d.f.		7		6		
	p		<0.01		0.832		
Venezuela							
Zulia		2,482	47	1.89	15	27	5
Falcón		844	47	5.57	17	27	3
Distrito Federal		1,145	20	1.75	8	10	2
Bolívar		4,121	44	1.07	14	27	3
Total		8,592	158	1.84	54	91	13
Homogeneity	χ^2		73.76		0.65		
	d.f.		3		3		
	p		<0.01		0.886		

Close consanguinity (F: 1/16 or higher); remote consanguinity (F: 1/32 or lower); total = effective total, excluding missing data cases.

The sex of all the intermediate ancestors was specified in 399 of the 512 couples (77.9%); 857 were males and 1,002 females (table 2). These values differed significantly from the expected 50% for the whole sample ($\chi^2 = 11.31$; df = 1; $p < 0.01$). The type of consanguineous mating (I, II, III or IV) could be determined in 215 first-cousin matings. There were 42 cases of type I (19%), 82 of type II (38%), 40 of type III (19%), and 51 of type IV (24%). These values differed significantly from the expected 0.25 proportions with an excess of type II matings ($\chi^2 = 21.08$; df = 3; $p < 0.01$).

Distribution by Country

Table 3 shows the consanguinity rates, the number of consanguineous matings by F, the types of first-cousin matings, the autosomal and X-linked mean inbreeding coefficients, and the distribution of intermediate ancestors by country. The number of observed cases in Argentina was significantly lower ($Z = -9.90$, $p < 0.05$), that in Brazil ($Z = 7.12$, $p < 0.05$) and in Venezuela ($Z = 8.33$, $p < 0.05$) significantly higher than that of the whole sample.

First-cousin matings represented the largest group in all countries, 44.3% (227/512) of all consanguineous matings. However, when all consanguineous couples were grouped as close ($F \geqslant 1/16$) or remote ($F < 1/16$), the significantly high rate was predominantly due to the latter type in Venezuela (91/145 = 62.8%), and to the former in Brazil (97/160 = 60.6%) ($\chi^2 = 15.72$; df = 1; $p < 0.01$).

Data from Brazil and Venezuela were further analysed as to their sampled states (table 4), showing that their total consanguinity rates were heterogeneous among the 8 sampled states of Brazil ($\chi^2 = 26.26$; df = 7; $p < 0.01$) and the 4 of Venezuela ($\chi^2 = 73.76$; df = 3; $p < 0.01$), while the fraction due to close relationships was homogeneous in both countries (Brazil: $\chi^2 = 2.82$; df = 6; $p = 0.832$; Venezuela: $\chi^2 = 0.65$; df = 3; $p = 0.886$).

Brazil and Venezuela did not differ in the proportion of couples with both members born in the same state (intra-marital about 85%), nor in the frequency of isonymic matings (isonymy about 20%). Furthermore, the frequencies of these two variables were homogeneously distributed among the states in both countries. Nevertheless, when migration was measured by the proportion of consanguineous couples with one or both members born outside the state they lived in, Brazil presented a higher rate of migrant consanguineous couples (*migration:* 37.2%) than Venezuela (*migration:* 16.9%) ($\chi^2 = 16.09$; df = 1; $p < 0.01$). In both countries this variable was heterogeneously distributed among their respective states. Specifically, migrant consanguineous couples were more frequent in São Paulo (61/121 = 50.4%) than in the other 7 sampled Brazilian states (6/59 = 10.2%) ($\chi^2 = 27.79$; df = 1; $p < 0.01$), and in the Federal District of Caracas (16/18 = 88.9%) than in the other 3 sampled Venezuelan states (10/136 = 7.4%) ($\chi^2 = 69.61$; df = 1; $p < 0.01$).

An excess of females among the intermediate ancestors could be seen in all countries with large enough sample size, and predominance of type II first-cousin matings in most of them, though without statistical significance.

Discussion

Consanguinity in Different Populations

The frequency of consanguineous matings varies considerably among different human populations [13], and it has often been mentioned that marriage is an agreement between families rather than a union between two people [14]. In Northern Africa and the West and Southeast of Asia they represent 22–55% of the total marriages [15], or even more [16]. On the other end of the spectrum, Europe and North America have rates as low as 0.5% [14], while in Japan the frequency lies somewhere between these two extremes [17].

In spite of incomplete sampling, and ascertainment based on mating products instead of actual matings, the overall consanguinity rate of 1% observed here is similar to the results reported by other authors for South America [2, 3, 18], and other Western populations [14].

Demographic Variables

When consanguinity was analysed by country, Argentina showed the lowest rate (0.29%), and Brazil and Venezuela the highest values (1.60 and 1.84%, respectively). This observation was already reported by Orioli et al. [7] in 1982, based on a partially overlapping data set with the one presented here. These authors considered that three main demographic characteristics could be responsible for the low consanguinity rate in Argentina, namely, a low birth rate, a high proportion of urban population and a high geographic and social mobility [7].

Our study observed a positive association between consanguinity and low paternal educational and occupational levels, both reflecting a low socioeconomic status. Similar observations have been mentioned in the literature for other populations such as Japan [19], Jordan [20], and Norway [21]. Variations of these two variables could to some degree be responsible for the variable inbreeding rates observed among different South American areas. It is noteworthy that the data presented here did not show

any association between maternal educational level and rate of consanguineous mating, reflecting intriguing relationships between sociocultural factors and mating patterns. In a Brazilian sample, Morton [22] (1964) observed a complex interaction among education, race, and inbreeding, where literacy increased with inbreeding in Caucasians, and decreased in Blacks.

Types of Relationship

The predominance of first-cousin matings among consanguineous couples has already been recognized in different societies [20, 23], and age correlation between mates seems to be an important factor favoring this type of mating.

The ascertainment through mating products used here could explain why our observed 0.11% rate for first-cousin unions in Argentina was lower than the 0.37% found by Castilla et al. [1] (1991), based on civil marriage certificates. Furthermore, the 13 cases with F higher than 1/16 could also be an underestimation of the real number, as some of these matings are legally or socially unacceptable and, thus, unlikely to be declared spontaneously. On the other hand, more remotely related couples could be underascertained, due to family memory limitations, as suggested below, when considering the excess of female intermediate ancestors.

Brazil and Venezuela

As already mentioned, Brazil and Venezuela were the countries with significantly higher consanguinity rates, when compared with the rest of the sample. However, different factors seem to be involved, as these two countries differed in the type of consanguinity, being mainly of the closer type (first cousins or closer) in Brazil, and of the more remote type in Venezuela. This difference would also explain the lower mean inbreeding coefficient (α), with a relatively higher consanguinity rate in Venezuela than in Brazil.

One possible factor which could give rise to such differences in the expected inbreeding distribution is migration [24]. Although it is known that migration reduces the probability of all types of consanguineous matings, it mainly affects the more distant relationships. On the other hand, as migrants often consist of groups of related individuals, migration could, in fact, increase the level of inbreeding, and this could be an explanation for the high consanguinity rate of the closer type in this Brazilian sample. A demographic factor which affects the expected distribution of inbreeding in an opposite sense is population growth. The natural increase (not through immigration) of a population reduces the proportion of close consanguineous matings, increasing that of more distant relationships. This could be the case of some relatively isolated Latin American towns, far off any migration wave, which, however, is not the case of the cities sampled in the present study.

The cities of São Paulo and Caracas are both industrialized attraction poles of internal migration for their respective countries. Therefore, even though it is not surprising that they displayed high rates of immigration, the high values of consanguineous matings based on close relationships, found in São Paulo, seemed unexpected.

The higher values of close consanguinity sampled in the state of São Paulo and the inter-state migration both suggest immigration of nuclear families instead of isolated individuals. In the late sixties, the 'hospedaria' study produced most of our present knowledge concerning the genetic structure of the Brazilian population migrating from the Northeast into the recently industrialized Southeastern states of São Paulo and Paraná [22]. Their mean inbreeding coefficient, estimated at 0.0059 from pedigree analysis, was found to be 44% lower than the adjusted value of 0.0133 by a bioassay method including 16 polymorphic systems [25]. This difference of almost one half of the actual value was partially attributed to the role of remote inbreeding. Furthermore, the author concluded that close consanguinity is a major fraction of the total inbreeding, when intra-marital distances are short.

Conversely, the Venezuelan consanguinity data, based on remote relationships, and concentrated in states with lower immigration rates, suggest a true local mating pattern, based on relative isolation, or on undetected low and middle-range migration. Rodriguez-Larralde et al. [10] (1993) studied the genetic structure of several states in Venezuela, through the distribution of surnames, in large samples of the general population. Different researchers have shown that the coefficient of inbreeding obtained by isonymy is greater than when obtained through pedigrees [9–11]. However, two of the four states sampled in our material are included among those with highest inbreeding coefficients in Venezuela, namely, Falcón (F: 0.0055) and Zulia (F: 0.0045), while the other two, Bolívar, and Federal District (Caracas), are reported to have the lowest values (F: 0.0027 and F: 0.0020, respectively). Furthermore, the state of Falcón, contributing about 30% to the Venezuelan consanguineous matings in this study, was found by Rodriguez-Larralde [unpubl. obs.], to have the lowest mean number of last names (260.49), as compared with 627.00 from Zulia, 720.24 from Bolívar, and 2,979.33 from the Federal District. The scarcity of last

names indicates a high rate of very remote inbreeding, inaccessible to the family memory, on which pedigrees are based.

Female Predominance

This study observed a significant excess of female intermediate ancestors, as well as a significant excess of type II first-cousin matings. Many authors have described a similar predominance of type II cousin matings, mainly in European countries, revealing an excess of mothers through whom relationships occur, with few mentions in the literature on predominance of other types [2,13].

Different factors have been mentioned which could explain the sex predominance in intermediate ancestors, as well as the excess of a certain type of consanguineous mating (migration differences between men and women, age differences, maternal influence, Oedipus complex) [2]. The material for our study was obtained between 1967 and 1996, which means that the members of the included couples were born between 1940 and 1970, considering an average generation time between 25 and 30 years. In this period, Latin America underwent an increasing process of industrialization, which gave rise to an active internal migration of people, moving mainly from rural areas into the cities. These migrants often consist of whole families coming from the same region, and perhaps families, maintaining their ties through sisters, would more easily move and settle together in the big cities. This would also lead to a higher probability of marriage between the offspring of two sisters, as a possible explanation for the excess of women among the intermediate ancestors, as well as for the excess of type II first-cousin matings in these pedigrees. An excess of female intermediate ancestors in close consanguineous matings, and the predominantly female role in the maintenance of family bonds has already been mentioned by Barrai et al. [26] (1962) in a study on Italian marriage records, and by Khlat et al. [27] (1986), with a different cultural background, among Muslims.

Finally, the greater certainty about maternal than paternal identity, especially in areas with a high rate of illegitimacy, could be favoring the better identification of females within a genealogy, thus contributing to the excess of female intermediate ancestors, including type II first-cousin matings, in our Latin American population. This biasing factor may be of particular strength in this study, since the informer was the mother of the newborn infant.

The observations made through this study are strengthened by the fact that it was based on a large, unselected sample of newborn infants, taken as index cases. This allowed the inclusion of all liveborn products of biologically fertile unions, irrespective of their legal or religious status.

On the other hand, these observations are limited to a few South American urban population samples, strongly dependent upon the memory of the mother, who was the informer, and thus, introducing a bias favoring the maternal family branch, and the higher sociocultural levels, among other probably unsuspected side effects.

Acknowledgments

This study was partially supported by the Agencia Nacional de Promoción Científica y Tecnológica (BID 802/OC-AR), the Consejo Nacional de Investigaciones Cientificas y Tecnicas (CONICET) of Argentina, the Programa de Apoio à Pesquisa Estratégica em Saúde (PAPES) da Fundação Oswaldo Cruz, and the Conselho Nacional de Desenvolvimento Científico e Tecnológico (CNPq) of Brazil.

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