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# Unequal Contributions of Male and Female Gene Pools From Parental Populations in the African Descendants of the City of Melo, Uruguay

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ABSTRACT In admixed populations, genetic contributions from males and females of specific parental populations can be of different proportions due to past directional mating during the process of genetic admixture. In this research paper, we provide evidence of such male- and female-specific differential admixture components of African, European, and American Indian origin in an admixed population from the city of Melo, in the northeastern region of Uruguay. From data on 11 autosomal markers from a sample of 41 individuals of mixed African descent, we estimated 47% African, 38% European, and 15% Amerindian contributions. In contrast, 6 mtDNA site-specific polymorphic markers showed that the mtDNA genome of these individuals was 52% African, 19% European, and 29% Amerindian, while from 3 Y-specific polymorphic sites, we estimated 30% African, 64% European, and 6% Amerindian contributions. We argue that this heterogeneity of admixture estimates results from disproportionate unions of European males with African and American Indian females from which this mixed African population was formed. Also, we argue that the asymmetry of the admixture estimates from the three sets of markers (autosomal, mtDNA, and Y-linked) is a result of the changes in the direction of mating during the history of the population. Implications of such evidence of directional mating are discussed, indicating the need of further demographic data for a quantitative assessment of the impact of directional mating on genetic structure of admixed populations. Am J Phys Anthropol 118:33–44, 2002.
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The utility of genetic markers in evaluating genetic compositions of admixed populations is well-documented (reviewed in Chakraborty, 1986). Genetic admixture is a process governed by various factors, which are influenced by physical proximity of parental populations, mate choice, and linguistic, social, and cultural attributes. In humans in particular, historical data suggest that frequently admixed populations do not receive equal contributions of male and female gene pools from all parental populations (Panunzio, 1941; Nelson, 1942; Kennedy, 1944, 1952; Spuhler and Clark, 1961; Jones, 1979; Lecompte, 1979; Swadesh, 1979). However, until recently, the genetic impact of differential contributions of male and female gene pools from parental populations on the composition of an admixed population was difficult to assess, because male- and female-specific polymorphic markers were not widely studied in admixed populations.

The advent of mitochondrial DNA (mtDNA) and Y-chromosome polymorphisms alleviated this problem to some extent, since mtDNA is maternally transmitted, and Y-linked genes are contributed

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from male lineages of individuals. Thus, differences of estimates of components of genetic admixture revealed from nuclear genes, mtDNA, and Y-chromosome markers can serve as indicators of the effects of gender-influenced gene flow (also called directional mating; see Merriwether et al., 1997) on the genetic composition of an admixed population. Feasibility of such studies, through the examination of heterogeneity of genetic contributions of specific parental gene pools in the autosomal, Y-linked, and mtDNA genes of admixed populations, has been established. For example, Ritte et al. (1993) compared genetic diversity at Y-linked and mtDNA markers in several Jewish communities and thus documented evidence of the influence of past directional mating (outside contributions are more from males than from females in all Jewish communities examined, except in the Ethiopians). In the United States, Merriwether et al. (1997) showed heterogeneity of genetic contributions of the American Indian gene pools in the nuclear vs. mtDNA genomes in the Mexican American and Anglo populations of Colorado. However, by estimating Caucasian admixture in African-Americans, Hsieh and Sutton (1992) could not detect the evidence of the influence of directional mating.

Directional mating in other African-derived populations has been studied in North America and the Caribbean region (Parra et al., 1998, 2001), as well as in South America (Bravi et al., 1997; Bortolini et al., 1999). In contrast to the findings of Hsieh and Sutton (1992), the analyses show that, in general terms, the maternal African and Amerindian contributions are larger than their respective paternal contributions in admixed populations. The European contribution is larger when Y-chromosome markers are studied. Indeed, a total absence of male African contribution was observed in Curiepe, Venezuela. However, there are some exceptions: e.g., in Brazil, locations such as Ribeirão Preto, Trombetas, and Paredão have a slightly lower African contribution when mtDNA is analyzed than when the Y-chromosome is analyzed (Bortolini et al., 1999). Intriguingly, autosomal African contributions in Ribeirão Preto, Brazil, are lower than those obtained from both mtDNA and Y-chromosome markers.

The purpose of this research is to add to the knowledge on this subject by studying the genetic composition of an African-derived population from the city of Melo, located in the northeastern region of Uruguay. Earlier studies described the ethnic composition of Uruguayan populations by using morphological characteristics (Kolski and Scazzocchio, 1961; Oyhenart-Perera, 1976; Sans et al., 1986, 1991, 1993a; Bertoni et al., 1994) and blood groups and protein markers (Alvarez et al., 1981, 1993; Miller et al., 1986, 1987; Sans et al., 1993b, 1995, 1997). These later studies, in particular, where estimates of genetic admixture components from various parental populations were given, demonstrated considerable heterogeneity of admixture components among different regions within Uruguay. For example, Sans et al. (1993b, 1997) showed that non-European gene pools make substantial contributions to the populations of Montevideo and Tacuarembó, but larger in Tacuarembó than in Montevideo. More recently, Bravi et al. (1997) presented data on mtDNA and Y-chromosome haplotype diversity in individuals of African descent from the city of Melo. Focusing on the population specificity (European, African, or American Indian) of haplotypes, they determined the population-origin of 93% of mtDNA haplotypes, while for Y-haplotypes, only in 41% of males could the population-origin be unequivocally established. Nevertheless, the data strongly suggest that the contributions to admixture from the maternal and paternal lineages differ (49% African, 12% European, and 32% American Indian in the mtDNA genome, vs. 18% African, 23% European, and no American Indian contribution in the Y-chromosome). These admixture components do not add up to 100% because, for 7% of the mtDNA haplotypes, the population specificity was not unequivocal, and for the Y-chromosome 59% of the haplotypes were of undetermined origin.

Therefore, in the present work we took a somewhat different approach to estimate admixture components from the same data described in Bravi et al. (1997). The ambiguities of the estimations based on the mtDNA and Y-chromosome sites studied by Bravi et al. (1997) were avoided by considering them as separate markers, although they are linked and do not recombine. In this procedure, the admixture estimates are estimated as the average of the contributions estimated, analyzing each marker under a trihybrid model of population admixture. The heterogeneity of admixture estimates provides indications of effects of past directional mating in this population. We also show that this inference is not an artifact of our selection of less informative autosomal markers and/or using the mtDNA and Y-chromosome markers as independently segregating. Implications of the findings in the context of modeling the dynamics of impact of directional mating are also discussed; the need for relevant demographic data is emphasized for supplementing genetic surveys in describing the genetic composition of admixed populations.

Also, new data on genotype/phenotype and allele frequencies at 11 autosomal markers are presented. Admixture estimates were obtained from these data by using the least-squares approach of Chakraborty (1985). We added autosomal markers in the admixture estimation, since this allows a better understanding of the dynamics of marriages. Examining how far the autosomal contribution is from the median of the lineage-specific (i.e., mtDNA and Y-linked) contributions revealed possible changes in the direction of mating.

MATERIALS AND METHODS

Population and sample

The city of Melo is located in the northeastern region of Uruguay, along the border with Brazil, approximately 400 km from the capital city of Montevideo. Melo was founded in 1795, being one of the first villages in the northeastern part of the country, and was subsequently populated by Luso-Brazilians, especially during the Portuguese domination of Uruguay (1814–1828) (Acevedo, 1933; González Mieres, 1968). According to the 1985 census, the population of Melo consisted of 42,326 inhabitants (Sexto Censo de Población y Cuarto de Viviendas, 1985). The history of Melo, as well as of other major cities of Northern Uruguay (such as Tacuarembó), is important to understand the genetic composition of the country. Traditionally it is believed that African genes came into the country mainly through the arrival of slaves from the port city of Montevideo. However, since slavery was abolished in Uruguay earlier (in 1842) than in Brazil (in 1888), apparently a large influx of slaves from the southern part of Brazil moved to northern Uruguay in search of freedom (Carvalho Neto, 1965; Rama, 1967; Ramos, 1976). Unlike the slaves brought earlier through Montevideo, these immigrant Brazilian slaves might have already been admixed, having European as well as American Indian genes in their gene pool. Nevertheless, genetic admixture studies indicate that in general, the percentage of genes of African ancestry in northeastern Uruguay is larger than in the Montevideo population. For example, Sans et al. (1997) estimated that the African contribution to the gene pool of the Tacuarembó population is about 18%, while in Montevideo the African contribution is between 8–10%. Unpublished data on 17 autosomal genetic markers indicate that of the total Melo population, 11–13% of its genes are of African origin.

Although the Afro-Uruguayan population of the city of Melo is not entirely culturally segregated, individuals of mixed African ancestry present a partial cultural identity, allowing studies of genetic substructuring within this population. For example, some of these individuals are members of an association, called "Club Uruguay," founded more than 50 years ago specifically because individuals of African ancestry were not allowed to participate in the other associations of the city mainly dominated by the "white" middle-class. Some cultural activities, especially those related to carnivals, also include participation from a high percentage of individuals of African descent. Thus, it is possible to collect demographic as well as genetic data from their constituencies, particularly from those who believe themselves to be of African descent. Nevertheless, as with the rest of the individuals of the city population, there is individual variation with regard to the proportion of genes of African, European, and American Indian ancestry within subgroups. We collected blood samples from 41 such unrelated individuals (19 females and 22 males), which constituted the basic materials for this study.

Genetic systems

From this collection, samples from 33 individuals allowed genetic analysis for 11 autosomal markers: two blood groups, ABO (with antisera −A and −B) and RH (with antisera −C, −c, −D, −E, and −e), and nine protein enzyme systems (phosphogluconate dehydrogenase, PGD; phosphoglucomutase 1 and 2, PGM1 and PGM2; acid phosphatase, ACP; esterase D, ESD; glyoxalase 1, GLO1; adenylate kinase, AK; haptoglobin, HP; and transferrin, TF) for which the methods of laboratory analysis are described in Sans et al. (1995).

DNA samples from 41 individuals were used for studying mitochondrial genetic variation, for which six markers were used. These are the presence/absence of restriction sites *Hae*III (nt663), *Hpa*II (nt3592), *Alu*I (nt5176), *Dde*I (nt10394), and *Hinc*II (nt13259); and the presence/absence of the region V 9-bp deletion at the respective positions of the consensus mtDNA sequence (Anderson et al., 1981). These markers are related to mutations which characterize population-specific mtDNA haplotypes (Torroni et al., 1992, 1994; Chen et al., 1995). For Y-chromosomal variation, DNA samples from the 22 males were analyzed for three markers: DYS19, α-h, and YAP (DYS287) (Hammer, 1994; Spurdle et al., 1994; Santos et al., 1995, 1996). The laboratory procedures for scoring genetic variation at the mtDNA and Y-chromosome sites, as well as the frequencies of each marker in the Melo sample of African-descent people, are given in Bravi et al. (1997). As mentioned before, complete haplotype data were not obtained from the mtDNA and Y-chromosome studies; nor are such haplotype data available for the relevant parental populations contributing to the admixture event. Therefore, we used the frequency of each marker separately.

Statistical analysis

Allele frequencies at the autosomal markers (blood groups and protein enzyme loci) were estimated from their respective genotype (phenotype) frequencies in the Melo sample by using the MAXLIK program of Reed and Schull (1968). Tests for goodness-of-fit of Hardy-Weinberg proportions of observed phenotype (genotype) frequencies were conducted by the exact test as described by Guo and Thompson (1992), since the sample sizes were too small to apply the standard large-sample tests. For mtDNA and Y-chromosome markers, relative frequencies were obtained by the counting method (see Bravi et al., 1997).

To obtain estimates of the contributions of African, Caucasian, and Amerindian genes for each type of marker (nuclear, mtDNA, and Y-chromosome), we used the ADMIX3 program of Chakraborty (1985), which requires estimated frequencies of each

marker for all three parental populations. As discussed in Sans et al. (1997), the choice of parental populations for admixture studies in populations of Uruguay is somewhat difficult, since precise knowledge of parental populations is not always available. According to historic information as well as availability of data, we used the weighted average of 13 European populations or regions, 11 South America Native populations, and 7 African populations as representations of parental populations contributing to the admixture in the Melo population. Appendix A lists these allele frequencies, together with the data sources at the 11 loci used in this analysis.

Likewise, for mtDNA and Y-chromosome markers, average frequencies for each marker from the relevant population groups were computed to represent parental populations. Appendix B lists the frequencies and data sources for mtDNA markers, and Appendix C lists similar information for the three Y-chromosome markers. In these cases, the frequencies for the Melo sample are also reproduced from Bravi et al. (1997).

At this point it should be mentioned that the pooling of data from relevant geographic groups of parental populations has an advantage in view of uncertainties of the exact populations contributing to the admixture. First, the use of average marker frequencies reduces the effect of genetic drift over time (Crow and Kimura, 1970). Second, a larger sample size, as obtained by data pooling, also minimizes the sampling errors of estimated allele frequencies in the parental populations. Using each mtDNA and Y-chromosome marker separately, instead of treating them as haplotypes, allowed us to utilize the maximum information from each marker. Even for the markers for which complete haplotype information and their frequencies are known in the Melo population (see Bravi et al., 1997), corresponding haplotype frequency data in the relevant populations of the world are not available (see references in Appendices B and C). Finally, significant differences between estimates of admixture components obtained from different sets of markers (e.g., autosomal vs. mtDNA vs. Y-chromosome markers) were tested by constructing the 95% confidence limit (i.e., estimate ± 1.96 SE) of each estimate, following the suggestion of Reed (1969).

RESULTS

Table 1 presents the genotype (phenotype) frequencies, maximum likelihood estimates of allele frequencies, and the exact test empirical level of significance values (*P*, based on 2,000 replications of simulations) of agreement with Hardy-Weinberg proportions for the 11 serological markers for the Melo sample, since these data have not been reported before. Three loci (PGD, PGM2, and TF) are monomorphic in this sample, though rare alleles may not have been observed in the small sample size of this study. Nevertheless, no locus showed any significant departure from the respective Hardy-Weinberg expectations of genotype/phenotype frequencies.

The estimates of proportions of genes of African, European, and American Indian origins, for the 11 autosomal, 6 mtDNA, and 5 Y-chromosome markers, are shown in Table 2, along with the R^2 values (an indicator of the goodness-of-fit of the admixture model) for the trihybrid model of admixture in the population of African descent in Melo. The high values of R^2 for all three types of markers indicate that the trihybrid model of admixture in the Melo population is a reasonable approximation of the process of admixture that occurred in the individuals of African descent in the city of Melo.

Before comparing the admixture components for the three sets of markers, we also checked whether the choice of markers affected our estimates. Appendix A shows that of the 11 autosomal markers, 5 (AK, PGD, PGM1, PGM2, and TF) had comparatively little allele frequency differences among the parental populations. Thus, we recomputed the admixture components using only the 6 remaining informative loci (shown in Table 1). Considering the sampling errors of the estimates, obviously this did not substantially change the autosomal admixture components. In fact, the 95% confidence intervals (CI) of the three admixture components (African, European, and American Indian) based on the protein loci, with and without exclusion of the relatively uninformative 5 loci, all overlap, signifying that the exclusion of uninformative protein loci did not affect the admixture estimates.

The effect of treating the six mtDNA markers separately, instead of treating them as haplotypes, was studied by computing mtDNA admixture estimates from the haplotype data as well. As mentioned before, Bravi et at. (1997) ascribed 5 of the Melo mtDNA haplotypes as European, 20 as of African descent, and the 13 remaining as from the American Indian gene pool (of the total of 38 for which unequivocal population-specificity could be established). Thus, mtDNA haplotype data yielded 13% European, 53% African, and 34% Amerindian contributions to the Afro-Uruguayan Melo population, not substantially different from the values (19%, 52%, and 29%) obtained from the independent consideration of the six mtDNA markers. A similar check for the three linked Y-chromosome markers could not be attempted, since relevant haplotype data for the parental populations are not available; nor do these markers alone provide the population-specificity of all Y-haplotypes seen in the Melo sample.

Comparison of the admixture components shows an interesting pattern: for the mtDNA markers, the contribution of African genes in this population is 52%, while the genes of paternal lineage (Y-chromosome) in the admixed individuals are of a considerably lower proportion of African origin (30%). This difference is statistically significant (at the 5% level), since the 95% confidence limits of these esti-

TABLE 1. Phenotypic and gene frequencies for blood groups, erythrocyte enzymes, and serum protein in population of African ancestry from Melo, Uruguay

Locus	Phenotype	N (frequency)	Allele	Frequency
ABO				
	A	7 (0.219)	I*A	0.151
	B	5 (0.063)	I*B	0.115
	AB	2 (0.156)	I*0	0.734
	O	18 (0.562)		
	Total	32		P = 0.83
RH				
	CCDEe	1 (0.048)		
	CcDEe	2 (095)	RH*CDc	0.214
	CcDee	5 (0.238)	RH*cDE	0.214
	ccDEe	7 (0.333)	RH*cDe	0.242
	ccDee	4 (0.191)	RH*cde	0.330
	ccdee	2 (0.095)		
	Total	21		P = 0.62
ACP				
	A	1 (0.030)	ACP*A	0.227
	AB	12 (0.364)	ACP*B	0.758
	B	19 (0.576)	ACP*C	0.015
	AC	1 (0.030)		
	Total	33		P = 0.28
AK				
	1-1	32 (0.970)	AK*1	0.985
	2-1	1 (0.030)	AK*2	0.015
ESD				
	1-1	29 (0.879)	ESD*1	0.939
	2-1	4 (0.121)	ESD*2	0.061
	Total	33		P = 1.00
PGD				
	A	33 (1.000)	PGD*A	1.000
	Total	33		
PGM1				
	1-1	19 (0.576)	PGM1*1	0.788
	2-1	14 (0.424)	PGM1*2	0.212
	Total	33		P = 0.28
PGM2				
	1-1	33 (1.000)	PGM2*1	1.000
	Total	33		
GLO1				
	1-1	6 (0.200)	GLO1*1	0.417
	2-1	13 (0.433)	GLO1*2	0.583
	2-2	11 (0.367)		
	Total	30		P = 0.71
HP				
	1-1	10 (0.476)	HP*1	0.619
	2-1	6 (0.286)	HP*2	0.381
	2-2	5 (0.238)		
	Total	21		P = 0.08
TF				
	C	21 (1.000)	TF*C	1.000
	Total	21		

TABLE 2. Estimates of proportions of African, European, and American Indian genes among individuals of African descent in Melo, Uruguay, from protein (autosomic loci), mitochondrial, and y-chromosome markers[1]

	Protein (11 loci)	Protein (6 loci)	mtDNA	Y-chromosome
African	0.469 ± 0.044	0.521 ± 0.032	0.523 ± 0.004	0.302 ± 0.006
European	0.376 ± 0.041	0.392 ± 0.044	0.190 ± 0.005	0.641 ± 0.020
American Indian	0.155 ± 0.055	0.087 ± 0.036	0.287 ± 0.006	0.057 ± 0.015
R^2	0.9638	0.9963	0.9998	0.9999

[1] R^2, multiple correlation coefficient, indicating goodness-of-fit of trihybrid model (see Chakraborty, 1985, for computational equation).

mates (0.515–0.531 for mtDNA, and 0.290–0.314 for Y-markers) are nonoverlapping. Even greater and statistically significant ($P < 0.05$) heterogeneity of the contributions of genes of European and American Indian origin exists among the three types of markers. For example, at the Y-chromosome, 64% of the genes are of European origin (95% CI, 0.602–0.680), while mtDNA shows only 19% European genes (95% CI, 0.180–0.200). American Indians contributed significantly less (6%) than did the Europeans through the male lineage, and more (29%) through the female lineages. In summary, the observed significant heterogeneity of these admixture estimates supports the hypothesis that early immi-

grants from Africa (through the arrival of African slaves of both sexes in Uruguay) were subsequently admixed with Europeans and American Indians. The genetically meaningful contacts were with disproportionate numbers of European males and American Indian females. In effect, these admixture estimates indicate that the present genetic composition of individuals of African descent in the city of Melo is the result of past directional mating that formed this admixed population.

In contrast, when the contribution of African genes is analyzed by comparing protein markers with both mtDNA and Y-chromosome markers, there is no statistically significant difference between proteins and mtDNA (95% CI, 0. 458–0.584 for 6 proteins, 0.383–0.555 for 11 proteins, and 0.515–0.531 for mtDNA); the same is not true when European and Amerindian contributions are analyzed in the same way.

DISCUSSION AND CONCLUSIONS

Recent interest in detecting differential contributions of parental populations through male and female lineages (the effect of directional mating) in admixed populations prompted this work. We have shown evidence that the current gene pool of individuals of African descent in Melo, Uruguay, consists of genes from three parental sources (Africa, Europe, and American Indian), for each of which the contributions through male and female lineages are unequal. The conclusions are consistent with historical records (mentioned below), but some limitations of data as well as methods must be stated, in view of which our findings should be regarded as preliminary. First, the sample size of this study (number of individuals typed from Melo) is undoubtedly small, as a result of which the allele frequencies are only approximate. In principle, this may have also compromised our ability to detect deviation of genotype frequencies in this admixed population from their respective Hardy-Weinberg expectations (HWE). However, since admixture events in Melo occurred for several generations in the past, it is not unlikely that free mixing of admixed individuals within Melo may have brought this population into Hardy-Weinberg equilibrium (as one generation of random mating is enough to do so; Li, 1976). Second, the markers studied at Y-chromosomes and mtDNA were scored in a manner which precluded defining the population-specificity of haplotypes unambiguously. As a consequence, instead of considering mtDNA and Y-chromosome data as haplotypes, we treated the separate markers as independent “loci.” However, as shown in Table 2, treating linked markers as independent apparently has little effect on the estimates per se.

Use of marker allele frequencies at different loci of the Y-chromosome separately removes the uncertainties found by Bravi et al. (1997), and modifies their admixture estimations in the Melo sample based on haplotypes. Bravi et al. (1997) showed that 18% of the Y-haplotypes in Melo are of African and 23% of European origin, with an additional 14% that could be of either African or European origin. They did not detect any definitive American Indian haplotype, but 59% (or 45%) of the haplotypes in the Melo sample are of undetermined origin in their study.

The dependence of mtDNA and Y marker allele frequencies across sites (recall that these sites do not recombine) affects the standard errors of the admixture components, as shown in Table 2. At this point, we may recall that the standard errors of the least-squares-based admixture estimates (Chakraborty, 1985) were evaluated from interlocus deviations of the admixture model, and these do not reflect the evolutionary stochastic variation of allele frequencies during the evolution of the admixed population. As mentioned before, geographic as well as drift variation of allele frequencies in parental populations are to some extent controlled by the choice of allele frequencies (weighted frequencies over several relevant populations; see Appendix A–C). Nonetheless, we admit that the admixture estimates for mtDNA and Y-chromosome data are not as precise as those indicated in Table 2, and should be considered downwardly biased, the magnitude of which may be assessed from the comparison of standard errors of the estimates based on haplotypes and on the mtDNA and Y-chromosome sites separately.

As in all admixture studies, the choice of parental populations is also approximate in this analysis. However, elsewhere (Sans et al., 1997) we showed that when genetically affinal groups are considered as parental populations (from several alternatives of Europeans, American Indians, and Africans), the autosomal markers showed quite consistent admixture estimates in another admixed population of Uruguay. Thus, we argue that the observed finding of heterogeneity of admixture components from protein, mtDNA, and Y-chromosome markers is not due to the approximate choice of allele frequencies from parental populations.

The trend of admixture estimates in the Afro-Uruguayan Melo population, when compared with similar estimates in other admixed populations of the American continent (reviewed in the Introduction), shows that the direction of heterogeneity of admixture estimates is the same as in most of them. Thus, we may argue that the process of admixture introduced American Indian genes into the African-derived groups predominantly through female lineages (i.e., through mating of American Indian females and males of other origins). The Y-chromosome data support this assertion, showing the considerably lower contribution of male American Indian genes in the Melo sample, in favor of European genes (Table 2). The origin of African genes is mostly from African women.

These heterogeneous contributions of the parental gene pools through paternal and maternal lineages are supported by the historical and demographic

information available on this population. In relation to the American Indian contribution, various historical references are of interest. The French naturalist, de Saint-Hilaire (1961), who visited the country during 1820–1821, refered to the frequent number of families consisting of a Spanish man, an American Indian woman, and generally a large number of children. Further, after the episodes that essentially eradicated the Charrúa Indians as an ethnic group, the survivors were mainly women and children who were given to families in the countryside and in Montevideo city (Acosta y Lara, 1979; Cabrera, 1983). Finally, Rosenblat (1954) mentioned that Spanish women were scarce and that European immigrants were predominantly men who had unions with native women.

It is not so easy to explain why the proportion of genes of African origin from the protein and mtDNA markers are nearly identical (about 50%). Recall that the admixture components, as estimated by models such as that employed in the present work, only estimate the proportion of genes in the extant admixed population from the different ancestral gene pools. Thus, the measure of admixture is the cumulative effect of a time-dependent admixture process. As a consequence, it is unlikely that the autosomal estimates of admixture would be exactly the average of those from mtDNA and Y-linked markers. Refined data on mtDNA and Y-chromosome haplotypes, together with nuclear DNA markers with more population-specificity of alleles (Shriver et al., 1997), should eliminate such ambiguities. In addition, the documentation of the relative frequency of interpopulation marriages that is available in archives can substantiate the effects of directional mating, and this approach will be attempted in our future studies. With such data in hand, it will be possible to more firmly predict the effect of directional mating, instead of the approximations that are currently suggested (e.g., Hsieh and Sutton 1992; Bravi et al., 1997).

Our finding that mtDNA and protein markers show the same African admixture component in Melo should not be considered evidence against the effect of directional mating. Indeed, our data on differences of admixture contributions from Europeans and American Indians, and in particular the Y-chromosome marker data, strongly suggest that the composition of the present gene pool of the Afro-Uruguayans of Melo is largely the result of directional mating of the past involving these ethnic groups. We argue that the first moment of admixture was between European male and African female. The Río de la Plata region, which includes Uruguay, part of Argentina, and southern Brazil, received slaves to work as house servants in urban areas, or as “peones” on ranches and as “saladeros” in the countryside, instead of working on plantations. This fact prompted the acquisition of a great number of black women. Women were sometimes more in demand than men (Isola, 1975). Further, although unions of European men and African women were considered illegitimate, and though they were well-scrutinized in the society, Isola (1975) mentioned that unions between European colonizers with women of African descent (slaves or free) were not rare. In contrast, unions between European women and African men were almost nonexistent. The archives of the Cathedral of Melo, which were started in 1797, refer to numerous “pardos” (i.e., mixed black and white), and in all the cases these pardos are reported as the sons or daughters of a “black” mother and an “unknown” father. Because of the definition of “pardo,” these fathers should be “white.” In Montevideo, 8% of the children born in 1778 were “pardos” (Isola, 1975). In addition, the genealogical data we obtained from the people sampled showed that unions between black or mulatto women and white men were more common than the inverse. In 57 genealogies recorded that belong to phenotypically “black” or “mulatto” people from Melo, 37 (65%) were of sons or daughters of two black or mulatto parents, 15 (26%) of black or mulatto mother and white father, and only 5 (9%) of black or mulatto father and white mother. The difference between these last two categories is statisticaly significant at the level of 5% ($\chi^2 = 4.05$, $P<0.05$). In conclusion, both historical and genetic data show evidences of directional matings.

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APPENDIX A. Frequencies used to estimate contribution of parental populations for enzyme-protein and erythrocyte systems (11 loci)

Populations[1]	Allele frequencies								N	References[2]
ACP										
	ACP*A	ACP*B	ACP*C	ACP*R						
Europeans	0.289	0.668	0.043	0.000					16,205	(1–8)
Africans	0.175	0.811	0.003	0.011					2,654	(3,5,9)
South American Indians	0.187	0.813	0.000	0.000					2,988	(3,10,11)
AK										
	AK*1	AK*2								
Europeans	0.961	0.039							4,732	(2,3,5,8)
Africans	0.999	0.001							1,635	(3,5)
South American Indians	1.000	0.000							1,425	(11–15)
ESD										
	ESD*1	ESD*2								
Europeans	0.869	0.131							12,175	(1–3,7,8,16)
Africans	0.932	0.068							1,679	(3,17)
South American Indians	0.636	0.364							848	(11,13–15)
PGD										
	PGD*A	PGD*C	PGD*Natal	PGD*R						
Europeans	0.975	0.025	0.000	0.000					13,256	(1–8,16)
Africans	0.937	0.062	0.000	0.001					2,155	(3,5,17)
South American Indians	0.998	0.000	0.002	0.000					971	(3,5,11,14)
PGM1										
	PGM1*1	PGM1*2								
Europeans	0.742	0.258							12,594	(1–3,5,8,16)
Africans	0.805	0.195							2,315	(3,5,17)
South American Indians	0.804	0.196							3,135	(10,12,15)
PGM2										
	PGM2*1	PGM2*2	PGM2*6	PGM2*9	PGM2*11					
Europeans	1.000	0.000	0.000	0.000	0.000				4,356	(2,3)
Africans	0.979	0.019	0.001	0.001	0.000				1,702	(3,5,17)
South American Indians	0.969	0.001	0.000	0.000	0.030				1,051	(11–13,17)
GLO1										
	GLO1*1	GLO1*2								
Europeans	0.414	0.586							11,245	(1–3,7,8,16)
Africans	0.297	0.703							1,617	(3,17)
South American Indians	0.376	0.624							317	(3)
HP										
	HP*1	HP*2								
Europeans	0.366	0.634							11,635	(3,5,5,8)
Africans	0.653	0.347							2,426	(3,5)
South American Indians	0.492	0.508							3,598	(9,10,18,19)
TF										
	TF*C	TF*B	TF*D							
Europeans	0.993	0.006	0.001						7,081	(3,5,20)
Africans	0.971	0.000	0.029						2,203	(3,5)
South American Indians	0.999	0.000	0.001						1,745	(11–13,15)
ABO										
	I*A	I*B	I*O							
Europeans	0.278	0.085	0.637						270,052	(5,21)
Africans	0.141	0.129	0.730						5,731	(21)
South American Indians	0.008	0.004	0.988						2,509	(5,10,11,13,18,19,21–23)
RH										
(RH*)	*CDE	*Cde	*cDE	*cDe	*CdE	*Cde	*cdE	*cde		
Europeans	0.009	0.455	0.111	0.045	0.001	0.012	0.006	0.361	29,161	(3,5,9)
Africans	0.001	0.052	0.068	0.634	0.000	0.010	0.000	0.235	5,303	(3,5,21)
South American Indians	0.048	0.549	0.340	0.039	0.000	0.000	0.000	0.024	3,346	(10,11,13,21,22,24)

[1] Geographically or ethnically-defined populations: Europe: South Italy, North Italy, France, Basques (Spain and France), Portugal, Canary Islands (Spain), Galicia (Spain), Sephardic Jews, Ashkenazic Jews, Germany, Russia, and Turkey. Africa: Senegal and Gambia, Liberia and Ivory Coast, Togo, Ghana and West Nigeria, East Nigeria and Cameroon, Angola and Congo, Mozambique, Rwanda, Burundi, and Zaire. South American Indians: Cayapo, Caingang, Guarani, Xavante, and Parakana (Brazil), Choroti, Ayoreo, and Guayaqui (Paraguay), Mataco and Toba (Paraguay and Argentina), and Mapuche (Chile and Argentina).

[2] References: 1) Rickards et al. (1992); 2) Barbeiro et al. (1987); 3) Roychoudhury and Nei (1988); 4) Aguirre et al. (1989); 5) Tills et al. (1983); 6) Morilla et al. (1988); 7) Picornell et al. (1990); 8) Nevo et al. (1989); 9) Adams and Ward (1973); 10) Salzano and Callegari-Jacques (1988); 11) Salzano et al. (1978); 12) Salzano et al. (1972b); 13) Salzano et al al. (1980); 14) Neel (1978); 15) Black et al. (1980); 16) Afonso et al. (1989); 17) Nurse et al. (1985); 18) Matson et al. (1968); 19) Brown et al. (1974); 20) Moreno et al. (1991); 21) Mourant et al. (1976); 22) Salzano et al. (1972a); 23) Matson et al. (1969); 24) Salzano (1964)

APPENDIX B. Mtdna markers, parental populations, and frequencies of mutations for estimation of different contributions

Marker	Frequency	N (total)	References[1]
+*Hae*III,663 bp			
Melo	0.098	41	(1)
African	0.000	101	(2)
European	0.000	383	(3–5)
American Indian	0.227	278	(6–9)
+*Hpa*II, 3592 bp			
Melo	0.366	41	(1)
African	0.731	175	(2,10)
European	0.002	516	(3–5,11,12)
American Indian	0.000	278	(6–9)
−*Alu*I, 5,176 bp			
Melo	0.000	41	(1)
African	0.000	101	(2)
European	0.000	383	(3–5)
American Indian	0.201	278	(6–9)
Region V deletion			
Melo	0.122	41	(1)
African	0.067	851	(2,13)
European	0.005	383	(3–5)
American Indian	0.300	323	(6–8,14)
+*Dde*I 10394			
Melo	0.610	41	(1)
African	0.930	101	(2)
European	0.000	383	(3–5)
American Indian	0.385	278	(6–9)
−*Hinc*II 13259			
Melo	0.098	41	(1)
African	0.000	101	(2)
European	0.000	383	(3–5)
American Indian	0.237	278	(6–9)

[1] References: 1) Melo, Uruguay (Bravi et al., 1997); 2) Senegal (Pygmees excluded), (Chen et al., 1995); 3) Turkey, Basques (Spain), Northern Germany, Portugal, Northern Spain, and UK (Richards et al., 1996); 4) Italy (Torroni et al., 1994); 5) Europeans (Horai et al., 1993); 6) Mapuche (Argentina) (Ginther et al., 1993; Foster et al., 1996); 7) Kraho (Brazil) and Mataco (Argentina) (Torroni et al., 1993); 8) Mapuche (Argentina) (Bailliet et al., 1994); 9) Amazonian (Brazil) (Santos et al., 1996); 10) Bantu and San (Johnson et al., 1983); 11) Rome (Italy) (Brega et al., 1986); 12) Jews (Israel) (Bonné-Tamir, 1986); 13) Bantu Central, Southeastern Bantu, and Khoisan (South Africa) (Soodyall et al., 1996); 14) Mapuche (Chile) (Horai et al., 1993; Merriwether et al., 1995).

APPENDIX C. Y-chromosome markers, parental populations, and frequencies of mutations for estimation of different contributions

Populations	Allele frequencies											N	References[2]
DYS19													
	A	B	C	D	E	F							
Melo	0.091	0.364	0.455	0.045	0.000	0.045						41	(1)
Africans	0.000	0.357	0.500	0.143	0.000	0.000						14	(2)
Europeans	0.061	0.519	0.218	0.161	0.041	0.000						293	(3,4)
South American Indians	0.863	0.098	0.029	0.000	0.010	0.000						102	(5)
α-h													
	I	II	III	IV	V	VI	VII	IX	X	XII	XIII		
Melo	0.000	0.409	0.227	0.046	0.136	0.000	0.000	0.182	0.000	0.000	0.000	41	(1)
Africans	0.000	0.000	0.000	0.000	0.071	0.000	0.000	0.858	0.071	0.000	0.000	14	(2)
Europeans	0.024	0.708	0.196	0.000	0.000	0.024	0.024	0.000	0.000	0.000	0.024	293	(3,4)
South American Indians	0.049	0.862	0.049	0.000	0.010	0.000	0.000	0.010	0.000	0.010	0.000	102	(5)
YAP													
	+												
Melo	0.318											41	(1)
Africans	0.789											526	(6,7)
Europeans	0.095											89	(8,9)
South American Indians	0.052											58	(10)

[1] References: 1) Bravi et al. (1997); 2) Kenya, Mathias et al. (1994); 3) Europeans, Santos et al. (1996); 4) Germans, Mueller et al. (1994); 5) Chorote, Chulupe, Gavião, Karitiana, Mapuche, Surui, Toba, Waiwai, Wichi, Xavante, and Zoro (Santos et al., 1996); 6) East Africa, Gambia, Nigeria, and Zambia (Hammer, 1994); 7) Bantu and Khoisan (Dama) (Spurdle et al., 1994); 8) Italy and UK (Hammer, 1994); 9) France (CEPH) (Bianchi et al., 1997); 10) Mapuche and Wichi (Argentina) (Bianchi et al., 1997)

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