Interethnic admixture and the evolution of Latin American populations

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Abstract

A general introduction to the origins and history of Latin American populations is followed by a systematic review of the data from molecular autosomal assessments of the ethnic/continental (European, African, Amerindian) ancestries for 24 Latin American countries or territories. The data surveyed are of varying quality but provide a general picture of the present constitution of these populations. A brief discussion about the applications of these results (admixture mapping) is also provided. Latin American populations can be viewed as natural experiments for the investigation of unique anthropological and epidemiological issues.

Keywords: interethnic admixture, Latin America, population structure.

The Latin American Microcosm

Geographically, Latin America extends over a vast area, from 32° North to 60° South and from 120° to 20° West of Greenwich. Within this territory, Middle America stretches from approximately 8° North to 32° North, while South America continues to the region’s southern limit.

Throughout this area lives a very diverse population, inhabiting equally diverse environments with distinct geological and ecological characteristics. Tropical, subtropical, and extremely cold climates all occur in the region, along with altitude variations and moisture extremes (for example, the Atacama desert in Chile is one of the driest places in the world).

Middle America has a population of approximately 165 million people, with slightly more than half of them characterized as ethnically admixed (Mestizos). Amerindian ancestry is especially prevalent in Mexico and Guatemala, while people of African descent are more common in the Caribbean area. South America contains roughly twice as many persons (approximately 322 million), and the influence of European ancestry is more marked in this region, although interethnic admixture is as common as in Middle America.

The human populations of Latin America have been investigated from an array of disciplinary perspectives. Global evaluations of the genetics and evolution of these populations have been conducted by Salzano (1971) and Salzano and Bortolini (2002). The admixture process was considered by Sans (2000), while Gibbon et al. (2011) examined the ways in which ethnic identities, genetic/genomic ancestry and health interacted in Argentina, Brazil, Colombia, and Uruguay.

Origins

General

The present Latin American populations are the consequences of a process that began in Northeastern Asia as early as 15,000-18,000 years ago (among others, Schurr and Sherry, 2004; Fagundes et al., 2008; Perego et al., 2009; Salzano, 2007, 2011). After the arrival of Europeans and Africans a little over five centuries ago, a complex process of admixture took place. This recent period has involved populations from a broad range of origins, making Latin American history unique (Sans, 2000). Presently, Latin America can be seen as a natural experiment for anthropological and epidemiological studies in which polymorphic loci and linkage disequilibrium can be used to infer the genetic basis for traits of interest (Chakraborty and Weiss, 1988).

The populations that entered into contact during the last several centuries were, as previously mentioned, very diverse. The original background of the region was composed by Native Americans; Europeans, mostly Spanish and Portuguese but also other nationalities; and Africans, who were initially brought to the region as slaves and came...
from different areas of that continent. Some Latin American countries have also received East Indian, Chinese, Japanese, Javanese, and other Asian populations. Examples include the Japanese immigrants that came to São Paulo and Pará in Brazil or the Chinese-origin populations that initially settled in the coastal valleys of Peru and were later distributed across the whole country.

Native Americans

As indicated previously, populations originating from Asia entered America approximately 15,000-18,000 years ago, but these dates, as well as the origin or origins of these populations in Asia, are still discussed. One of the most accepted views involves a coastal route that includes a stage in Beringia, a land bridge that appeared intermittently between 70,000 and 12,000 years ago. Fagundes et al. (2008) suggested a complex model that involved an early differentiation from Asian populations, a slow and lengthy evolution in Beringia, and a rapid expansion in America due primarily to a maritime route and coastal settlements along the Pacific coast. This first expansion would have been followed by a terrestrial expansion through the Mackenzie corridor in North America and continued to the south by land. Other proposals, including the entrance through the Atlantic Ocean bordering Greenland, the "Solutrean solution" proposed by Stanford and Bradley (2002), and the crossing of the North pole proposed by O’Rourke and Raff (2010), cannot be rejected, but if they occurred, they most likely involved fewer migrants.

The genetic characteristics of these initial populations have not been completely identified, but several studies indicate a wide initial diversity, as seen mainly in mitochondrial DNA (mtDNA) studies (Tamm et al., 2007; Fagundes et al., 2008; Malhi et al., 2010; Perego et al., 2010; Yang et al., 2010; Bisso-Machado et al., 2012). Part of the initial variation could have been lost, as suggested by Cui et al. (2013) in an analysis of mtDNA from skeletal remains. This diversity can also be observed in the analysis of the Y chromosome (Schurr and Sherry, 2004; Bisso-Machado et al., 2011, 2012). As for autosomes, a total of 678 microsatellite markers, genotyped in 422 individuals from 24 North, Central and South American natives, were studied by Wang et al. (2007); 364,470 single nucleotide polymorphisms were examined in 52 Native American and 17 Siberian groups by Reich et al. (2012). This second study indicated the presence of at least three different streams of Asian gene flow in the prehistoric colonization of the Americas, a view suggested nearly three decades ago by Greenberg et al. (1986).

Salzano and Bortolini (2002), based on several studies, have estimated that approximately 45 million Native Americans were living in Latin America at the beginning of the European Conquest, but projections range from 30 to 90 million. This population, however, rapidly decreased as a consequence of epidemics and the violence occurring as wars, massacres, and maltreatment (Pereña, 1992). As stated by Salzano and Callegari-Jacques (1988), native populations were in different phases of cultural and demographic development at the onset of the European Conquest. Sedentary communities in more densely populated regions suffered the structural impacts of epidemics and other factors most strongly, while many small groups became extinct. However, after this severe depopulation, Native American populations recovered, and their present numbers (approximately 63 million, unpublished estimate) are higher than in the 16th century.

Africans

Most people from Africa were forced to migrate to America due to the slave trade, but Pedro Alonso Niño, Columbus’s pilot, was the first African immigrant (Brawley, 2001). As early as 1518, the Spanish crown had issued the first license for slave traffic, while Portugal had established its own companies and colonies to capture slaves. The first captives apparently entered Brazil through Bahia in 1531 (Ramos, 1934). The trade started in the Caribbean region and soon afterwards extended to Central America and the north of Brazil; its prohibition occurred over a lengthy process from 1830 to 1870.

The origins of the slaves ranged from the Guinea coast to Mozambique. Rout (1976) defined four main regions: upper Guinea, lower Guinea, the Congo Delta and Angola, and Mozambique. The flow from each of these areas varied over the long history of the slave trade and was also dependent on the nationality of the slave traders (Salzano and Bortolini, 2002).

The number of slaves brought to Latin America is difficult to determine, and factors such as slave mortality during the journey, illegal traffic, and the lack of registration due to ship interference at sea only complicate this estimation. Reader (1998) approximated that nine million slaves were shipped across the Atlantic between 1452 and 1870.

Europeans

Different processes influenced the European migrations over the five centuries after the initial contact. The first period, characterized by the arrival of the Spanish and Portuguese, was related to the Conquest. Spanish colonization was ruled by the Crown and was consequently planned as an emigration policy under royal permissions. Later, laws became more permissive due to the scarcity of migrants. This policy aimed to stimulate the migration of married settlers, farmers and artisans (Konetkze, 1991). However, because part of the migration was related to military campaigns, more men than women reached the continent. These regulations were in effect until the 18th century, when migration became independent of the Spanish government. The state was less involved and enacted fewer regulations in the immigration of the Portuguese, French, Dutch, and others to America (Konetkze, 1991).
Estimating the number of Europeans who entered Latin America is a complex undertaking. McAdoo (1993), referring to immigrants to the United States, stated that “the waves of persons who came to these shores are a portion of American history that is too often hidden away, for it embarrasses Americans,” and the same statement can be applied to Latin America. Moreover, this migrant flux continued until relatively recently: the last wave occurred after World War II, and the origins of the migrants changed to include the entire Mediterranean region as well as other European countries, particularly those in Eastern Europe. It is reasonable to suggest that a new wave, involving mainly Africans and East Asians, has recently started to arrive to Latin America.

The numbers, origins and destinations of European migrants depended on the time of and reasons for the migration. For example, during the 16th century, 300,000 people belonging to different economic and social levels left Spain to come to America and spread to different parts of the continent; however, this figure refers only to travelers, not to people who remained in the continent (Konetkze, 1991). The quantity of people entering America increased quickly, and as a result, Europeans and their descendants numbered 850,000 in 1650, 13,470,000 in 1825, and 221,160,000 in 1950, according to Rosenblat (1954).

History of the Admixture Process

The admixture process started soon after Christopher Columbus first disembarked at La Hispaniola (1492), as the 39 men he left on the island had sexual intercourse with the local Native American women (Mörner, 1967). Therefore, a maximum of roughly 21 generations of admixing may be established, with some variance due to region. Wang et al. (2008), based on 13 mixed Latin American populations, estimated that the average time since first admixture allowed for six to 14 generations, but these estimations excluded the Caribbean region, where the process began. Based on pedigrees, Heyer et al. (1997) identified up to 19 generations, descended from males who lived in the 17th century.

While admixture at first involved primarily Spanish (or European) men and Native American women, it shortly expanded to include European or “criollo” (European descendants born in America) men and mixed women or mixed men and women. The process was complicated by the introduction of African slaves into America at the beginning of the 16th century. The legal status of Africans delayed their admixture with other ethnic groups, but sexual intercourse between African men and Native American or mixed women, and later between African or African-descendant women and European or criollo men, was relatively frequent (Mörner, 1967).

The social, cultural, and economic characteristics of native populations, aspects of the European Conquest and Colonization, and the nature of the Europeans and Africans arriving at the continent undoubtedly influenced the admixture process. For example, prehistoric America had great cultural and demographic heterogeneity. Salzano and Callegari-Jacques (1988) classified these populations into three stages: hunters and gatherers with incipient agriculture, with low fertility and mortality; sedentary and more advanced agriculturalists, with high fertility and high mortality; and populations living in densely inhabited areas, with high fertility and low mortality. Accordingly, the consequences of the Conquest and Colonization were different in relation to each stage; the last two were more affected by diseases and were more involved in the admixture process.

Clearly, more males than females migrated to Latin America, and the Spanish Crown encouraged unions between European men and Native American women as a way to evangelize and/or obtain economic advantages; unions between Africans and any other group were prohibited. As Mörner (1967) has stated, “In a way, the Spanish Conquest of the Americas was a conquest of women”.

More recently, Latin American countries have accepted intermarriage as legal, with some exceptions; the law against marriages with Chinese in Mexico is among the most recent bans (Mörner, 1967). “Mixed blood” offspring generally became an integral part of European family life, and a long tradition of contact exists between Africans and both Spanish and Portuguese persons. Moreover, a distinct Mestizo (mixed Native American and Iberian) identity emerged in some parts of Latin America, including Mexico and Brazil (Yinger, 1985). In the latter country, interethnic unions were even favored. The Marquis of Pombal, who governed Brazil in the middle of the 18th century, established that mixed citizens should receive equal treatment as unmixed ones regarding employment, honor and dignity (Rosenblat 1954).

Individually, people from the first generation of an admixture process will have entire chromosomes of a single origin, while the second generation will have chromosomes containing blocks of different ancestry. As the process persists, these blocks of different origins will become smaller. Consequently, the chromosomes of people living in the present day show a complex mix of ancestry, depending on the number of generations of admixture, marriage patterns, and the characteristics of the mixed people involved in them.

The Genetic/Genomic Approach to Interethnic Admixture

Bernstein (1931) and Ottensooser (1944) were the first to use allele frequencies in admixed and parental populations to estimate the accumulated proportional contributions of the parental groups to a given admixed population. From that modest beginning, a vast array of methods and computing programs have been developed to analyze the problem. All of these techniques depend on two basic assumptions: (a) there is no error in the choice of parental
groups or in their genetic/genomic frequencies, and (b) the
changes are due mainly to gene flow, not other evolutionary
factors which may influence the estimates. Examples in-
clude those of Chakraborty (1975, 1985), who used the
gene identity method to incorporate fluctuations due to the
size of the hybrid population; Bertorelle and Excoffier
(1998), who added the changes that may have occurred in
parentals and hybrids after the event of admixture; Wang
(2003), who considered gene flow after the first event of ad-
mixture; and McKeigue et al. (2000), who employed a
Bayesian approach to incorporate the effects of linkage and
population structure.

Molecular approaches now allow researchers to sepa-
rate maternal and paternal contributions (mitochondrial
DNA, Y chromosome); to identify variation in all DNA re-
gions (coding and noncoding, introns, pseudogenes, repeat
sequences, regulatory elements); and to determine the ori-
gin of chromosome segments depending on the ancestors’
origin. The degree of divergence among alleles (Bertorelle
and Excoffier, 1998; Dupanloup and Bertorelle, 2001) and
the genetic drift since admixture calculated using different
approaches (Bayesian: Chikhi et al., 2001; maximum like-
lihood: Wang, 2003; and coalescence: Excoffier et al.,
2005) can now be considered. Molecular techniques have
also improved the estimation of individual admixture, an
approach initially proposed by Hanis et al. (1986) to avoid
the variability among individuals due to recombination and
independent loci assortment.

Ancestry Informative Markers (AIMs), that is, those
with high discriminatory power due to large interethnic dif-
fferences in frequencies, are also now being used, as sug-
gested by Shriver et al. (1997, 2003) and Collins-Schramm
et al. (2002).

Whole genome data sets are revealing complex sto-
ries of divergence and admixture that are impossible to ob-
tain using other approaches. For example, Harris and
Nielsen (2013), using parent-offspring trios from the 1000
Genomes Project, were able to detect extensive gene flow
between Africa and Europe after their populations di-
verted; together with the ancient admixture into Europe,
the results reveal a population structure that must be recon-
sidered to construct more realistic models of the gene pools
in these regions. In a more restricted analysis, Wang et al.
(2008) employed 678 autosomal and 29 X-chromosomal
microsatellites to differentiate the Native American ances-
try among Mestizos from 13 Latin American populations.

In the following sections, we provide detailed and
specific information about continental ancestries. Because
uniparental and X chromosome estimates generally overes-
timate non-European contributions, and Y chromosomes
the European influence, we concentrated our attention on
autosomes. With the exception of the data for Uruguay, for
which we wanted to construct a complete picture, protein
markers, which have been adequately surveyed in previous
reviews (Sans, 2000; Salzano and Bortolini, 2002), are not
considered here.

Actual Data, Middle America

Mexico

A total of 19 reports regarding molecular autosome
estimates of parental continental ancestry in Mexican pop-
ulations are presented in Table 1. Three of them concern the
country in general, while 27 concern specific populations,
with the large population of Mexico City heavily repre-
sented (seven estimates). Amerindian ancestry is most
prevalent (51% to 56%) in the three general estimates, fol-
lowed by European ancestry (40% to 45%); the African
share represents only 2% to 5%. The Amerindian contribu-
tion is the highest in 22 (81%) of the 27 estimates.

Many cases of repeated sampling in the same popula-
tion were recorded: for the general evaluations, the percent-
age differences are minimal (at most 6%), but for Nuevo
León, Veracruz, Guerrero, and Yucatan (all sampled
twice), the highest differences, generally involving the Eu-
ropean fraction, are 17% to 28%. In Mexico City, the Euro-
pean contribution was estimated as 21% to 32% in six of the
seven reports, with the anomalous value of 57% obtained in
a single sample of 19 subjects. European ancestry is most
prevalent in the north (Chihuahua, 50%; Sonora, 62%;
Nuevo León, 55%), but in a recent sample from Nuevo
León and elsewhere in the country, Amerindian ancestry is
dominant. The general conclusion, therefore, is that the
Amerindian genes were victorious in the battle of survival
over those of the Spanish Conquistadores!

Other Middle American countries

Table 2 presents the Middle American data excepting
those of Mexico. Clear differences may be observed be-
tween the areas: some show considerable African influence
(Carib as a whole, 77%; Haiti, 96%; Jamaica, 78%-82%),
others Spanish (Cuba, 73%-86%; Puerto Rico, 60%-76%,
with some interregional variability; Nicaragua, 69%; Costa
Rica, 58%-67%), and another Amerindian (Guatemala,
53%). In the Carib, the exception is Dominica, whose es-
sentially trihybrid structure reflects the French (as opposed
to Anglophone) influence throughout its history.

Actual Data, South America

Colombia

As shown in Table 3, the Antioquia region has been
extensively studied (five Mestizo samples, one Afro-
derived sample), with variable results (European fraction
estimated from 46% to 79%); however, the three surveyed
studies of its main urban center, Medellín, showed similar
values for the European contribution (60%-66%). The two
independent estimates from North Santander and Valle del
Cauca are also rather similar, with lower (39%-42%) Euro-
pean fractions.
When the specific populations from the six considered regions are examined, a wide array of continental percentage frequencies are observed, preventing an arrival at general conclusions. The same is true for the three African-derived samples, which showed African contributions ranging from 89% (Antioquia) to 46% (Mulaló). This variability may be real, but sampling biases, as well as the types and numbers of markers tested, should also be considered.

New, more specific and directed investigations should be undertaken.

Brazil

Brazil is the country from which the largest number of parental ethnic estimates were obtained. Table 4 lists 13 uniparental references (mtDNA only: 1; mtDNA+Y chromosome: 4; Y chromosome only: 8), two X-linked references.

### Table 1 - Molecular autosome estimates of parental continental ancestry in different segments of the Mexican population.

| Type and no. of markers | Population or region | No. indiv. studied | % Ancestry | References |
|-------------------------|----------------------|--------------------|------------|------------|
|                         |                      |                    | European | African | Amerindian |
| 44 AIMs                 | General              | 181                | 45        | 4       | 51        | 1 |
| 1814 AIMs               | General              | 300                | 42        | 2       | 56        | 2 |
| 446 AIMs                | General              | 312                | 40        | 5       | 55        | 3 |
| 13 STRs                | Chihuahua            | 161                | 50        | 12      | 38        | 4, 5 |
| 1814 AIMs               | Sonora               | 60                 | 62        | 2       | 36        | 2 |
| 10 STRs                | Nuevo León           | 143                | 55        | 5       | 40        | 6 |
| 74 AIMs                | Nuevo León           | 100                | 38        | 6       | 56        | 7 |
| 1814 AIMs               | Zacatecas            | 60                 | 46        | 3       | 51        | 2 |
| 13 STRs                | General              | 211                | 52        | 10      | 38        | 5, 8 |
| 13 STRs                | Jalisco              | 309                | 31        | 16      | 53        | 5, 9 |
| 1814 AIMs               | Guanajuato           | 60                 | 40        | 1       | 59        | 2 |
| 13 STRs                | Hidalgo              | 106                | 25        | 11      | 64        | 5, 10 |
| 1814 AIMs               | Veracruz             | 60                 | 36        | 2       | 62        | 2 |
| 13 STRs                | Veracruz             | 130                | 9         | 17      | 74        | 5 |
| 69 AIMs                | Mexico City          | 286                | 30        | 5       | 65        | 11 |
| 15 STRs                | Mexico City          | 378                | 26        | 5       | 69        | 12 |
| 678 STRs               | Mexico City          | 19                 | 57        | 3       | 40        | 13 |
| 128 AIMs               | Mexico City          | 66                 | 37        | 2       | 61        | 14 |
| 13 STRs                | Mexico City          | 242                | 21        | 15      | 64        | 5, 15 |
| 550 Kb                 | Mexico City          | 984                | 31        | 3       | 65        | 16 |
| 446 AIMs               | Mexico City          | 1310               | 32        | 4       | 64        | 17 |
| 13 STRs                | Puebla               | 313                | 17        | 11      | 72        | 5, 9 |
| 24 AIMs                | Guerrero             | 156                | 4         | 1       | 95        | 18 |
| 1814 AIMs               | Guerrero             | 60                 | 28        | 4       | 67        | 2 |
|                       |                      |                    |           |         |           |   |
| 1814 AIMs               | Yucatan              | 60                 | 39        | 1       | 60        | 2 |
| 13 STRs                | Yucatan              | 262                | 19        | 11      | 70        | 5, 9 |
| 13 STRs                | Campeche             | 106                | 8         | 16      | 76        | 5, 19 |

1. Chowdhry et al. (2006), Mexican recruited in the San Francisco Bay Area, USA; 2. Silva-Zolezzi et al. (2009); 3. Galanter et al. (2012); 4. Martínez-Gonzalez et al. (2005); 5. Rubi-Castellanos et al. (2009a); 6. Cerda-Flores et al. (2002); 7. Martínez-Fierro et al. (2009); 8. Hernández-Gutiérrez et al. (2005); 9. Rubi-Castellanos et al. (2009b); 10. Gorostiza et al. (2007); 11. Martínez-Marignac et al. (2007); 12. Juárez-Cedillo et al. (2008); 13. Wang et al. (2008); 14. Kosoy et al. (2009), 26 individuals from Mexico City and 40 Mexicans from California, USA; 15. Luna-Vazquez et al. (2005); 16. Johnson et al. (2011); 17. Galanter et al. (2012); 18. Bonilla et al. (2005); and 19. Sánchez et al. (2005).
ences, and 27 references involving autosome markers. The data are subdivided into three sets: sociogeographic regions, Afro-Brazilian communities, and specific populations.

Five sociogeographic regions are generally recognized by official censuses, according to a large number of criteria, and their ethnic ancestries vary as may be generally expected from Brazilian history. More European influence is observed in the southeast and south (up to 89%), while the African contribution predominates in the northeast (maximum estimate 30%) and the Amerindian in the north (up to 19%). The center-west estimates show the highest resemblance to the northern values (Table 4).

| Type and no. of markers | Population or region | No. indiv. studied | % Ancestry       | References² |
|------------------------|----------------------|--------------------|------------------|-------------|
| 105 AIMS               | Eight Caribbean islands | 420 | 15 | 77 | 8 | 1 |
| 13 STRs               | New Providence, Bahamas | 221 | 4 | 96 | 0 | 2 |
| 17 AIMS               | Havana, Cuba, Spanish-descendants | 79 | 86 | 14 | 0 | 3 |
| 17 AIMS               | Havana, Cuba, Afro-derived | 50 | 23 | 77 | 0 | 3 |
| 17 AIMS               | Havana, Cuba, Mulattos | 77 | 60 | 40 | 0 | 3 |
| 16 AIMS               | Havana, Cuba, Random sample | 129 | 73 | 26 | 1 | 4 |
| 60 AIMS               | Havana and Matanzas, Cuba | 584 | 81 | 16 | 3 | 5 |
| 13 STRs               | Haiti | 111 | 4 | 96 | 0 | 2 |
| 44 AIMS               | Puerto Rico | 181 | 60 | 21 | 19 | 6 |
| 15 STRs               | Puerto Rico | 192 | 76 | 17 | 7 | 7 |
| 93 AIMS               | Puerto Rico, West | 99 | 69 | 16 | 15 | 8 |
| 93 AIMS               | Puerto Rico, South | 75 | 65 | 19 | 16 | 8 |
| 93 AIMS               | Puerto Rico, North | 115 | 64 | 19 | 17 | 8 |
| 93 AIMS               | Puerto Rico, Central | 87 | 69 | 17 | 14 | 8 |
| 93 AIMS               | Puerto Rico, Metropolitan | 129 | 64 | 21 | 15 | 8 |
| 93 AIMS               | Puerto Rico, East | 137 | 55 | 32 | 13 | 8 |
| 93 AIMS               | Puerto Rico, Total | 642 | 64 | 21 | 15 | 8 |
| 13 STRs               | Jamaica | 119 | 16 | 78 | 6 | 2 |
| 105 AIMS               | Jamaica | 44 | 10 | 82 | 8 | 1 |
| 105 AIMS               | St. Thomas | 99 | 17 | 77 | 6 | 1 |
| 105 AIMS               | St. Kitts | 47 | 8 | 86 | 6 | 1 |
| 105 AIMS               | Dominica | 37 | 28 | 56 | 16 | 1 |
| 105 AIMS               | St. Lucia | 50 | 18 | 75 | 7 | 1 |
| 105 AIMS               | St. Vincent | 51 | 13 | 81 | 6 | 1 |
| 105 AIMS               | Grenada | 48 | 12 | 81 | 7 | 1 |
| 105 AIMS               | Trinidad | 43 | 16 | 75 | 9 | 1 |
| 678 STRs               | Guatemala, East | 20 | 40 | 7 | 53 | 9 |
| 15 STRs               | Nicaragua | 165 | 69 | 20 | 11 | 10 |
| 678 STRs               | Costa Rica, Central Valley | 20 | 67 | 4 | 29 | 9 |
| 39 AIMS               | Costa Rica, Central Valley | 1998 | 58 | 4 | 38 | 11 |

1As indicated, articles reporting protein markers (for instance, Arias et al., 2002; Morera et al., 2003) or uniparental markers only (Martinez-Cruzado et al., 2001; McLean Jr et al., 2005; Castri et al., 2007; Benn Torres et al., 2007; Gaieski et al., 2011) were not included. No specific prevalences were given by Bryc et al. (2010) or Moreno-Estrada et al. (2013).

21. Benn Torres et al. (2013); 2. Simms et al. (2008, 2010) East Asian, not Amerindian frequencies were employed in the analyses; 3. Cintado et al. (2009); 4. Diaz-Horta et al. (2010); 5. Teruel et al. (2011), includes individuals with dementia (40%); 6. Chowdhry et al. (2006); 7. Tang et al. (2007); 8. Via et al. (2011); 9. Wang et al. (2008); 10. Nuñez et al. (2010); 11. Ruiz-Narváez et al. (2010).
Table 3 - Molecular autosome estimates of parental continental ancestry in different segments of the Colombian populations.

| Type and no. of markers | Population or region \(^2\) | No. indiv. studied | % Ancestry | References \(^3\) |
|-------------------------|-----------------------------|--------------------|-------------|-----------------|
|                         | Geographic regions          |                    | European    | African         | Amerindian      |
| 8 AIMs                  | Antioquia, NW               | 80                 | 79          | 6              | 16             | 1             |
| 11 AIMs                 | Antioquia, NW               | 80                 | 63          | 11             | 26             | 2             |
| 5 Alu insertions        | Antioquia, NW, Mestizos     | 34                 | 64          | 17             | 19             | 3             |
| 5 Alu insertions        | Antioquia, NW, Afro-derived| 64                 | 0           | 89             | 11             | 3             |
| 75 AIMs                 | Antioquia, NW               | 849                | 60          | 12             | 28             | 4             |
| 52 AIMs                 | Antioquia, NW               | 25                 | 46          | 20             | 34             | 5             |
| 52 SNPs                 | North of Santander, NE      | 32                 | 42          | 18             | 40             | 5             |
| 11 AIMs                 | North of Santander, NE      | 35                 | 42          | 5              | 53             | 2             |
| 52 SNPs                 | Coffee area, CE             | 66                 | 45          | 20             | 35             | 5             |
| 11 AIMs                 | Vale de Cauca, SW           | 124                | 39          | 22             | 39             | 2             |
| 52 SNPs                 | Vale de Cauca, SW           | 28                 | 42          | 23             | 35             | 5             |
|                         | Specific populations        |                    |             |                |                |               |
| 11 AIMs                 | Caribbean area              |                    |             |                |                |               |
|                         | Cartagena                   | 80                 | 23          | 44             | 33             | 2             |
| 11 AIMs                 | Santa Marta                 | 26                 | 50          | 28             | 22             | 2             |
| 678 STRs                | Pasto                       | 19                 | 39          | 4              | 57             | 6             |
|                         | Northwest                   |                    |             |                |                |               |
| 678 STRs                | Medellín                    | 20                 | 66          | 9              | 25             | 6             |
| 11 AIMs                 | Medellín                    | 80                 | 63          | 11             | 26             | 2             |
| 75 AIMs                 | Medellín                    | 849                | 60          | 12             | 28             | 4             |
| 678 STRs                | Peque                       | 20                 | 37          | 5              | 58             | 6             |
| 11 AIMs                 | Peque                       | 163                | 32          | 6              | 62             | 2             |
| 11 AIMs                 | Manizales                   | 203                | 59          | 4              | 37             | 2             |
|                         | Northeast                   |                    |             |                |                |               |
| 11 AIMs                 | Bucaramanga                 | 82                 | 56          | 1              | 43             | 2             |
| 52 SNPs                 | Arauca                      | 73                 | 40          | 22             | 38             | 5             |
|                         | Central                     |                    |             |                |                |               |
| 11 AIMs                 | Armenia                     | 58                 | 57          | 5              | 38             | 2             |
| 11 AIMs                 | Bogotá                      | 24                 | 45          | 3              | 52             | 2             |
| 52 SNPs                 | Boyacá                      | 80                 | 42          | 20             | 38             | 5             |
| 678 STRs                | Cundinamarca                | 19                 | 47          | 2              | 51             | 6             |
| 11 AIMs                 | Yopal                      | 20                 | 24          | 1              | 75             | 2             |
| 52 SNPs                 | Huila                       | 82                 | 41          | 19             | 40             | 5             |
| 52 SNPs                 | Tolima                      | 26                 | 41          | 21             | 38             | 5             |
|                         | Southwest                   |                    |             |                |                |               |
| 11 AIMs                 | Pasto                       | 201                | 32          | 3              | 65             | 2             |
| 11 AIMs                 | Popayan                     | 61                 | 20          | 23             | 57             | 2             |
| 11 AIMs                 | Neiva                       | 24                 | 39          | 0              | 61             | 2             |
| 52 SNPs                 | Huila                       | 82                 | 41          | 19             | 40             | 5             |
| 52 SNPs                 | Nariño                      | 78                 | 30          | 19             | 51             | 5             |
| 34 AIMs                 | Cauca                       | 306                | 48          | 11             | 41             | 7             |
| 52 SNPs                 | Mulaló, Afro-derived        | 33                 | 28          | 46             | 26             | 5             |
|                         | Pacific coast               |                    |             |                |                |               |
| 11 AIMs                 | Quibdo, Mestizos            | 170                | 47          | 8              | 45             | 2             |
| 11 AIMs                 | Quibdo, Afro-derived        | 72                 | 21          | 68             | 11             | 2             |
| 52 SNPs                 | Chocó                       | 93                 | 23          | 54             | 23             | 5             |

\(^1\) As indicated, articles reporting uniparental markers only (for instance Rodas et al., 2003; Yunis and Yunis, 2013) were not included. No specific prevalences were given by Bryc et al. (2010).

\(^2\) NW: Northwest; NE: Northeast; CE: Central; SW: Southwest.

\(^3\) 1. Bedoya et al. (2006); 2. Rojas et al. (2010); 3. Gómez-Pérez et al. (2010); 4. Duque et al. (2012); 5. Ibarra et al. (2014), Porras et al. (2009); 6. Wang et al. (2008); 7. Córdoba et al. (2012).
Table 4 - Autosome estimates of parental continental ancestry in different segments of the Brazilian population.

| Sampling criteria | Type and no. of markers | Population or region | No. indiv. studied | % Ancestry | European | African | Amerindian | Reference |
|-------------------|--------------------------|-----------------------|--------------------|------------|----------|---------|-----------|-----------|
|                   |                          |                       |                    |            |          |         |           | 1         |
| Sociogeographic regions |                          |                       |                    |            |          |         |           | 1         |
| North            | 12 STRs                  | 253                   | 68                 | 14         | 18       | 1       |           | 1         |
|                  | 28 AIMs                  | 40                    | 71                 | 18         | 11       | 2       |           | 2         |
|                  | 40 AIMs                  | 203                   | 70                 | 11         | 19       | 3       |           | 3         |
| Northeast        | 12 STRs                  | 164                   | 75                 | 15         | 10       | 1       |           | 1         |
|                  | 28 AIMs                  | 40                    | 77                 | 14         | 9        | 2       |           | 2         |
|                  | 40 AIMs                  | 82                    | 61                 | 30         | 9        | 3       |           | 3         |
| Center-West      | 12 STRs                  | 286                   | 71                 | 18         | 11       | 1       |           | 1         |
|                  | 28 AIMs                  | 40                    | 69                 | 19         | 12       | 2       |           | 2         |
| Southeast        | 12 STRs                  | 109                   | 75                 | 18         | 7        | 1       |           | 1         |
|                  | 28 AIMs                  | 40                    | 80                 | 14         | 6        | 2       |           | 2         |
|                  | 40 AIMs                  | 264                   | 74                 | 19         | 7        | 3       |           | 3         |
| South            | 12 STRs                  | 226                   | 81                 | 11         | 8        | 1       |           | 1         |
|                  | 28 AIMs                  | 40                    | 88                 | 7          | 5        | 2       |           | 2         |
|                  | 40 AIMs                  | 189                   | 78                 | 13         | 9        | 3       |           | 3         |
|                  | 48 AIMs                  | 81                    | 89                 | 3          | 8        | 4       |           | 4         |
| Afro-Brazilian communities | 3 VNTRs, 3 STRs          | 2 communities, N      | 64                 | 18         | 47       | 35      | 5         | 5         |
|                  | 48 AIMs                  | 5 communities, N      | 103                | 15         | 69       | 16      | 4         | 4         |
|                  | 48 AIMs                  | 7 communities, N      | 294                | 29         | 48       | 23      | 6         | 6         |
|                  | 10 AIMs                  | 3 communities, NE     | 207                | 39         | 49       | 12      | 7         | 7         |
|                  | 14 STRs                  | Marinhas, SE          | 60                 | 33         | 67       | 0       | 8         | 8         |
|                  | 48 AIMs                  | 10 communities, SE    | 307                | 39         | 40       | 21      | 9         | 9         |
| Specific populations | 12 STRs                  | Macapá, N             | 307                | 46         | 19       | 35      | 10        | 10        |
|                  | 48 AIMs                  | Macapá, N             | 130                | 50         | 29       | 21      | 11        | 11        |
|                  | 13 STRs                  | Belém, N              | 325                | 46         | 34       | 20      | 12        | 12        |
|                  | 48 AIMs                  | Belém, N              | 196                | 54         | 15       | 31      | 4         | 4         |
|                  | 6 VNTRs                  | São Luís, N           | 161                | 33         | 67       | 0       | 13        | 13        |
|                  | 2 STRs, 2 VNTRs          | São Luís, N           | 177                | 42         | 19       | 39      | 14        | 14        |
|                  | 9 STRs                   | Macéio, NE            | 598                | 56         | 27       | 17      | 15        | 15        |
|                  | 12 STRs                  | Brasília, CW          | 153                | 67         | 21       | 12      | 16        | 16        |
|                  | 28 AIMs                  | Brasília, CW          | 200                | 77         | 14       | 9       | 17        | 17        |
|                  | 11 STRs                  | Belo Horizonte, SE    | 234                | 65         | 34       | 1       | 18        | 18        |
| 3                | 3 VNTRs, 2 STRs          | Rio de Janeiro, SE    | 3 blood groups     | 81         | 67       | 21      | 12        | 19        |
|                  | 40 AIMs                  | Euro-Brazilians       | 107                | 86         | 7        | 7       | 20        | 20        |
| 3                | 3 VNTRs, 2 STRs          | Afro-Brazilians       | 3 blood groups     | 69         | 39       | 49      | 12        | 19        |
| 4                | 40 AIMs                  | Afro-Brazilians       | 228                | 55         | 37       | 8       | 20        | 20        |

1. Salzano and Sans.
Table 4 - cont.

| Sampling criteria | Type and no. of markers | Population or region | No. indiv. studied | % Ancestry | Reference |
|-------------------|------------------------|----------------------|--------------------|-------------|-----------|
|                   |                        |                      |                    | European    | African   | Amerindian |
| 4                 | 46 AIMs                | Afro-Brazilians      | 113                | 39          | 52        | 9         | 21        |
| 1                 | 46 AIMs                | General              | 280                | 59          | 30        | 11        | 21        |
|                   |                        | Ribeirão Preto, SE   |                    |             |           |           |           |
| 3                 | 8 STRs                 | Euro-Brazilians      | 400                | 79          | 14        | 7         | 22        |
| 3                 | 7 STRs                 | Afro-Brazilians      | 220                | 50          | 88        | 12        | 22        |
| 1                 | 6 VNTRs                | Campinas, SE         | 206                | 64          | 36        | 0         | 13        |
| 1                 | 15 STRs                | São Paulo, SE        | 294                | 52          | 34        | 14        | 24        |
|                   |                        | São Paulo, SE        |                    |             |           |           |           |
| 4                 | 48 AIMs                | Euro-Brazilians      | 367                | 63          | 22        | 15        | 27        |
| 4                 | 48 AIMs                | Afro/Euro-Brazilians | 68                 | 45          | 41        | 14        | 27        |
| 4                 | 48 AIMs                | Afro-Brazilians      | 51                 | 32          | 57        | 11        | 27        |
| 1                 | 9 STRs                 | Porto Alegre, S      | 104                | 86          | 3         | 11        | 25        |
| 1                 | 678 STRs               | Bagé and Alegrete, S | 20                 | 70          | 10        | 20        | 26        |

1 Additional information based on uniparental or X-linked markers can be found as follows: (a) mtDNA only: Carvalho et al. (2008); (b) mtDNA plus Y-chromosome: RibeiroDosSantos et al. (2002), Marrero et al. (2005, 2007), Hnemenier et al. (2007), Guerreiro-Junior et al. (2009); (c) Y-chromosome only: Carvalho-Silva et al. (2001), Ferreira et al. (2006), Silva et al. (2006), Ribeiro et al. (2009), Carvalho et al. (2010), Palha et al. (2011), Ribeiro et al. (2011), Francez et al. (2012); (d) X-linked only: Ribeiro-Rodrigues et al. (2009), Resque et al. (2010).

2 Key to sampling criteria: 1. Random; 2. Total ancestry, proportion of a given ancestry in a given color category multiplied by the official census information about the proportion of that color category in the specified region; 3. Morphological evaluation; and 4. Self-reported ethnicity.

3 N: North; NE: Northeast; CW: Center-West; SE: Southeast; and S: South.

4 References: 1. Callegari-Jacques et al. (2003); 2. Lins et al. (2010); 3. Pena et al. (2011); 4. Santos et al. (2010); 5. Vairinoto et al. (2003); 6. Maciel et al. (2011); 7. Amorim et al. (2009); 8. Scliar et al. (2009); 9. Kimura et al. (2013); a subset of these communities was also studied for two Alu insertions, and the values used to estimate parental contributions (Cotrim et al., 2004), but the results obtained showed inconsistencies among the populations and were not considered; 10. Francez et al. (2011a); 11. Francez et al. (2011b); 12. Ribeiro-Rodrigues et al. (2007); 13. Ramos et al. (2004); 14. Ferreira et al. (2005); 15. Ferreira da Silva et al. (2002); 16. Godinho et al. (2008); 17. Lins et al. (2011); 18. Scliar et al. (2009); 19. Palhatik et al. (2002); 20. Suarez-Kurtz et al. (2007); 21. Manta et al. (2013); 22. Ferreira et al. (2006); 23. Muniz et al. (2008); 24. Sao-Bento et al. (2008); 25. Leite et al. (2003); 26. Wang et al. (2008); 27. Cardenas et al. (2013).

As a result of the present emphasis of the Brazilian government on positive actions in favor of previously discriminated African-derived people, many rural areas where escaped slaves found refuge are now being demarcated to ensure property rights for their descendants. Several of these communities have been studied regarding parental markers (Table 4), and the results show high heterogeneity among them. These communities have not remained isolated from persons of other ethnic backgrounds, as illustrated by the fact that in four of the six estimates, their African contributions amount to less than half of the total parental ancestry.

Twelve specific populations are listed in Table 4. The main results can be summarized as follows: (a) repeated sampling in both Macapá and Brasília yielded less than 10% average differences; the discrepant values obtained in São Luís are most likely due to the nature and number of the markers used; (b) in Rio de Janeiro, Ribeirão Preto and São Paulo, Afro-Brazilians and Euro-Brazilians were considered separately; the sampling differences in Rio de Janeiro were not high, but the differences between Rio de Janeiro, Ribeirão Preto and São Paulo in terms of the African ancestry in Afro-Brazilians are large; and (c) differences in the specific and global regional ethnic parental estimates were most marked for São Luís [probable reason indicated in (a)].

Although in population terms the morphological and genetic/genomic evaluations generally agree, wide variability exists for individual, personal estimates. This observation should be expected due to the high frequency of interethnic unions that occurred in the past and that are likely even more prevalent in the present. The implication for the implementation of positive actions is obvious: it is impossible to establish objective, specific criteria of ethnic classification to screen potential candidates for a program. These programs must therefore rely on self-classification, disregarding possible errors in favor of the general socio-economic improvement of the minorities considered.

Peru

The extensive study conducted by Sandoval et al. (2013) using 40 AIMs provides a good overall estimate of the degree of interethnic admixture present in the populations of the three main geographical regions of the country (Table 5). Overall, non-Amerindian European ancestries varied from 1% to 31%, while the African contribution was only 1% to 3%. The two coastal populations sampled contain 14% to 15% European admixture, while much lower...
frequencies occur in the Amazon (2% to 9%). Some heterogeneity is observed in the Andes, with three of the four northern populations (San Marcos, Cajamarca, Chogo) presenting only 67%, 76%, and 82% Amerindian ancestry, respectively. In the extreme south, the results from Characato also show evidence of more marked admixture (73% Amerindian heritage); but the 14 other populations sampled yielded high values (90% to 98%) for Amerindian gene pools.

**Argentina**

Nine studies involving autosomal molecular markers were considered for Argentina, and the results are given in Table 6. For the country as a whole, the European influence (65%-79%) is dominant, but the Amerindian contribution (17%-31%) is also important. The African influence, however, is minimal (2%-4%). Considering the four geographical regions of the country, somewhat different estimates were obtained for the northeast and northwest, but the evaluations for the center and south were in general agreement.

Turning to specific populations, a geographical pattern emerges in relation to Amerindian parentage, with higher frequencies observed in the northwest and south. Within the northwest region, there is ample variability (33%-100%) of the Amerindian contribution in the Province of Jujuy, but a rough gradient of decreasing Amerindian influence is generally observed from north to south. The values in the northeast and center are similar; in the Buenos Aires megalopolis, the Amerindian percentages are modest (16%-17%) in the inner and first urban belt, but the contribution increases to 29% in the second urban belt, where migrants and people of generally low socioeconomic status live. The African contribution is low but detectable in most of the regions and populations studied, reaching its maximum in Santiago del Estero and La Rioja.

**Uruguay**

Uruguay is unique among Latin American countries in that it has no Native American or African-descendant communities. This fact has shaped its national identity, which at least until the 1980s was considered to be almost strictly European. This view has also affected population genetics studies. In 1986, however, the first analysis of the Mongolian spot trait showed a frequency that was much higher (42%) than that expected in a mostly European population (Sans et al., 1986). This initial study encouraged additional enquiries into the Uruguayan identity. One of the first of these studies indicated that Montevideo (in the south) and Tacuarembó (in the northeast) had differences between them: while the African and Amerindian contributions in Montevideo were estimated as 7% and 1%, respectively, the same contributions in Tacuarembó were 15% and 20%, respectively (Sans et al., 1997). These data demonstrated not only a clear non-European contribution to ancestry in the northeast but also population heterogeneity in this small country (176,215 km², 3,286,314 inhabitants, according to the 2011 National Census, INE, 2012). This estimation was based on classical (blood groups, electrophoretic proteins) markers, as was another study in Cerro Largo, in the northeast, that showed 10% Native American and 8% African ancestry contributions (Sans et al., 2006). These data are in agreement with those obtained from a study using nDNA, which estimated 6% African and 10% Native American contributions for the whole country (Hidalgo et al., 2005).

Studies in self-defined African descendants, however, have presented very different results: African genes contribute nearly half of the ancestry to these populations, while Native American contributions vary from 15% in the

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**Table 5** - Molecular autosome estimates of parental continental ancestry in different segments of the Peruvian population.

| Population or region | No. indiv. studied | % Ancestry |
|----------------------|--------------------|------------|
|                      |                    | European   | African  | Amerindian |
| Amazon               |                    |            |          |            |
| Andoa                | 71                 | 4          | 1        | 95         |
| Iquitos              | 8                  | 2          | 2        | 96         |
| Chachapoyas          | 15                 | 5          | 2        | 93         |
| Lamas                | 18                 | 9          | 2        | 89         |
| Pucallpa             | 10                 | 8          | 1        | 91         |
| Coast                |                    |            |          |            |
| Lambayeque           | 31                 | 15         | 3        | 82         |
| Lima                 | 43                 | 14         | 2        | 84         |
| Andes                |                    |            |          |            |
| Cajamarca            | 34                 | 21         | 3        | 76         |
| San Marcos           | 19                 | 31         | 2        | 67         |
| Ocopon               | 11                 | 4          | 2        | 94         |
| Chogo                | 14                 | 15         | 3        | 82         |
| Huarochiri           | 15                 | 2          | 1        | 97         |
| Huancayo             | 29                 | 6          | 3        | 91         |
| Ayacucho             | 31                 | 8          | 2        | 90         |
| Kaquiabamba          | 9                  | 2          | 1        | 97         |
| Andahuaylas          | 19                 | 2          | 1        | 97         |
| Cabauaconde          | 20                 | 3          | 1        | 97         |
| Yanque               | 10                 | 1          | 1        | 98         |
| Chivay               | 25                 | 2          | 1        | 97         |
| Characato            | 8                  | 24         | 3        | 73         |
| Mollebaya            | 8                  | 3          | 1        | 96         |
| Amantani             | 31                 | 1          | 1        | 98         |
| Uros                 | 25                 | 2          | 1        | 97         |
| Taquile              | 23                 | 1          | 1        | 98         |
| Anapia               | 24                 | 1          | 1        | 98         |

1 All of them were studied with 40 AIMs by Sandoval et al. (2013). It was assumed that the strictly non-European and non-African ancestries (labelled as Oceanian and East Asian) were remote ancestral Amerindian ancestries.
Several studies have focused on uniparental markers, especially mtDNA. The Native American maternal contri-

Table 6 - Molecular autosome estimates of parental continental ancestry in different segments of the Argentinian population.

| Type and no. of markers | Population or region | No. indiv. studied | % Ancestry | References |
|-------------------------|----------------------|--------------------|------------|------------|
|                         |                      |                    | European   | African    | Amerindian |
| Geographic regions      |                      |                    |            |            |            |
| 99 AIMS                 | Northeast            | 33                 | 54         | 5          | 41         | 1          |
| 24 SNPs                 | Northeast            | 61                 | 79         | 4          | 17         | 2          |
| HLA-A, B                | Northwest            | 1,293              | 55         | 10         | 35         | 3          |
| 99 AIMS                 | Northwest            | 37                 | 33         | 3          | 64         | 1          |
| 24 SNPs                 | Central              | 153                | 81         | 4          | 15         | 2          |
| 99 AIMS                 | Province of Buenos Aires | 263         | 76         | 4          | 20         | 1          |
| 24 SNPs                 | South                | 32                 | 68         | 4          | 28         | 2          |
| 99 AIMS                 | South                | 108                | 54         | 3          | 43         | 1          |
| 99 AIMS                 | General              | 441                | 65         | 4          | 31         | 1          |
| 100 K+149 AIMS          | General              | 94                 | 78         | 2          | 20         | 4          |
| 24 SNPs                 | General              | 246                | 79         | 4          | 17         | 2          |
| Specific populations    |                      |                    |            |            |            |
| HLA-A, B                | Jujuy, NW            | 273                | 47         | 0          | 53         | 3          |
| 8 Alu insertions        | Jujuy, La Puna, NW   | 47                 | 0          | 0          | 100        | 5          |
| 8 Alu insertions        | Jujuy, Quebrada Baja, NW | 36            | 16         | 7          | 87         | 5          |
| 8 Alu insertions        | Jujuy, Quebrada Alta, NW | 36            | 5          | 3          | 92         | 5          |
| 8 Alu insertions        | Jujuy, Selva, NW     | 45                 | 23         | 0          | 77         | 5          |
| 8 Alu insertions        | Jujuy, Valle, NW     | 62                 | 16         | 7          | 77         | 5          |
| HLA-A, B                | Salta, NW            | 241                | 56         | 3          | 41         | 3          |
| 678 STRs                | Salta, NW            | 19                 | 25         | 3          | 72         | 6          |
| HLA-A, B                | Catamarca, NW        | 81                 | 53         | 10         | 37         | 3          |
| 678 STRs                | Catamarca, NW        | 14                 | 53         | 3          | 44         | 6          |
| HLA-A, B                | Tucuman, NW          | 418                | 67         | 9          | 24         | 3          |
| 678 STRs                | Tucuman, NW          | 19                 | 65         | 4          | 31         | 6          |
| HLA-A, B                | Santiago del Estero, NW | 156         | 46         | 24         | 30         | 3          |
| HLA-A, B                | La Rioja, NW         | 124                | 50         | 19         | 31         | 3          |
| 24 SNPs                 | Formosa, NE          | 11                 | 75         | 3          | 22         | 2          |
| 24 SNPs                 | Misiones, NE         | 28                 | 83         | 4          | 13         | 2          |
| 24 SNPs                 | Corrientes, NE       | 21                 | 77         | 5          | 18         | 2          |
| 24 SNPs                 | Buenos Aires, CE1    | 150                | 81         | 4          | 15         | 2          |
| 99 AIMS                 | Buenos Aires, inner city, CE | 98           | 79         | 4          | 17         | 1          |
| 99 AIMS                 | Buenos Aires, 1st urban belt, CE | 47           | 80         | 3          | 16         | 1          |
| 15 STRs                 | Mar del Plata, CE    | 180                | 77         | 1          | 22         | 7          |
| 19 Alu insertions       | Bahia Blanca, CE     | 119                | 79         | 0          | 21         | 8          |
| 15 STRs                 | Bahia Blanca, CE     | 85                 | 68         | 4          | 28         | 7          |
| 24 SNPs                 | Rio Negro, S         | 31                 | 68         | 4          | 28         | 2          |
| 9 blood polymorphisms   | Puerto Madryn, S     | 82                 | 68         | 3          | 29         | 9          |

1Earlier studies by Avena and coworkers that included protein markers only were not included here.

2NE: Northeast; NW: Northwest; CE: Central; S: South.

31. Avena et al. (2012); 2. Corach et al. (2010); 3. Alfaro et al. (2005); 4. Seldin et al. (2008); 5. Gómez-Pérez et al. (2011); 6. Wang et al. (2008); 7. Parolin et al. (2013a); 8. Resano et al. (2007); 9. Parolin et al. (2013b).

4Using 12 autosomal markers Fejerman et al. (2005) estimated as 2% the African ancestry of a sample of 90 individuals from the city of Buenos Aires.
bution estimated in all of these studies is higher than that estimated by paternal genes (Y chromosome) or autosomes, reaching 62% in Tacuarembó (Bonilla et al., 2004) and decreasing to 21% in Montevideo (Gascue et al., 2005). For the whole country, the maternal Native American ancestry was determined as 34% (recalculated from Pagano et al., 2005a). Self-identified Basque descendants with Basque paternal surnames living in Trinidad, in the southwest, also showed high levels (20%) of Native American maternal ancestry (Sans et al., 2011). The African contribution is relatively less, but the maternal contribution of this ancestry was estimated as 17% in Tacuarembó (Bonilla et al., 2004) and 21% in Cerro Largo (Sans et al., 2006). Self-defined African descendants presented 52% African and 29% Native ancestry (Sans et al., 2002). Y-chromosomal data indicated much lower African (less than 1%) and Native American (less than 8%) contributions (Bertoni et al., 2005, Pagano et al., 2005b).

Genealogical data have also been used to better understand parental contributions for Basque descendants (Sans et al., 2011), people from Santo Domingo de Soriano in the west (Barreto, 2011), and Canarians in Canelones, close to Montevideo (Barreto, 2008).

Recently, genetic or multifactorial diseases related to or influenced by ethnic ancestry have attracted attention. For hemoglobinopathies, the presence of 0.8% (2.2% in individuals with declared African ancestry) HbS, 1.2% of the 0-codon in α-thalassemia, and 0.25% of the β9 codon in β-thalassemia in Montevideo indicated the influence of African and Mediterranean ancestries (Da Luz et al., 2013). Other diseases for which gene frequencies are being investigated are diabetes (Mimbacas et al., 2004), breast cancer and melanoma (Cappetta et al., unpublished, and studies in progress).

The genetic studies performed in Uruguay have had two important outcomes. First was their contribution to the change in Uruguayan national identity, a process that started in the 1980s based on historical, demographic, and anthropological information (Verdesio, 1992; Viñar, 1992; Demasi, 1995), as well as ethnic movements (associations such as “Mundo Afro” or “Asociación de Descendientes de Indígenas Charrúas,” both founded in 1989, and others, which appeared more recently). This “new” identity currently relates Uruguay to other Latin American countries rather than to Europe exclusively (Sans, 2011). Second was their contribution to the search for diseases associated with non-European ancestries, linking alleles to population origins. This latter contribution is now demonstrating its full potential, and ancestry studies are being incorporated to bolster analyses of disease presence, frequency and prognosis.

Other South American countries

The results for three previously undiscovered South American countries are displayed in Table 7. For Venezuela, the data of the geographic regions indicated a basically trihybrid distribution, with similar contributions of European, African and Amerindian parentages. However, differences emerge when specific populations are considered, with higher (52% to 73%) European fractions. The exception is an African-derived isolate in the state of Zulia (100% African). Additionally, a sample from people of lower socioeconomic status in Caracas shows higher frequencies of African and Amerindian parentages than one from people of a higher socioeconomic level (27% vs 8% and 40% vs 17%, respectively). This result reflects the stratification that has occurred over centuries of ethnic discrimination across Latin America. Two samples from Ecuador show a high (73%) Amerindian contribution to Mestizos, while the Afro-derived subjects present only an approximately half (56%) African parentage. The two Chilean populations surveyed indicate a basically dihybrid (European/Amerindian) composition.

Actual data - Overview

The data surveyed generally confirm previous historical and nonmolecular evaluations. A marked Amerindian influence exists in Mexican, Guatemalan, Peruvian, and Ecuadoran populations, while European ancestries are more prevalent in Cuba, Puerto Rico, Nicaragua, Costa Rica, Argentina, and Uruguay. For Venezuela, the European, Amerindian, and African fractions are similar, while for Colombia, high interpopulation variability is observed. The ethnic distribution in Brazil follows a geographical pattern, with European influence more prevalent in the southeast and south, African in the northeast, and Amerindian in the north. For Chile, the Amerindian and European contributions are equivalent.

The data from which these general conclusions were obtained, however, are uneven and of varying quality. The following problems were noted: (a) dissimilar amounts of coverage for the different countries, some of which were not represented at all; (b) sample representativeness in nature and size; (c) types and number of markers used; (d) phenotypic characterization of the subjects sampled; and (e) methods of quantitative ancestry determination. Only a systematic, comprehensive approach, ideally involving multinational teams of researchers, will yield a more detailed picture of the highly complex process of admixture and its social implications.

Application - Admixture Mapping

Continental parentage estimates are not only of historical interest. Admixture mapping is a tool that is increasingly being used to localize disease genes in populations of recently mixed ancestry in which the ancestral populations have differing genetic risks. For example, Native American and Latino populations show higher frequencies for type 2 diabetes, obesity, gallbladder disease, and rheumatoid arthritis, as well as lower prevalences for asthma and prostate
cancer, in comparison to populations of European ancestry (Price et al., 2007; Winkler et al., 2010).

This method has both advantages and disadvantages in comparison to dense, whole-genome scans. The advantages include (a) lower genotyping costs; (b) the use of disease cases only, avoiding the noise introduced by inadequate controls; and (c) the characteristics of the admixture signal, which reduce the number of hypotheses tested. The disadvantages include (a) possible differences between the estimated ancestry and the frequencies of a given disease allele; (b) imperfect power to estimate local ancestry; and (c) the need for the fine mapping of the fraction of admixture that successfully identifies a disease locus.

Panels for these analyses involving Mexican Americans have been developed by Collins-Schramm et al. (2004; 100 AIMs) and Tian et al. (2007; 5,287 AIMs), and similar panels have been developed for Hispanic/Latino populations in general by Price et al. (2007; 1,649 AIMs) and Mao et al. (2007; 2,120 AIMs). Examples of the specificity of this approach include: (a) Price et al. (2007), who estimated that in Latinos and Mexicans from Los Angeles, where Native American ancestry is close to 50%, admixture mapping should be 15% to 30% more powerful per sample than in Colombians or Brazilians, who have lower percentages of this ancestry; and (b) Fejerman et al. (2012), who identified a region in chromosome 6 related to breast cancer susceptibility in Latinas. A detailed review of the studies in this area, however, is outside the scope of the present work.

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