A comprehensive review of uniparental systems in South Amerindians was undertaken. Variability in the Y-chromosome haplogroups were assessed in 68 populations and 1,814 individuals whereas that of Y-STR markers was assessed in 29 populations and 590 subjects. Variability in the mitochondrial DNA (mtDNA) haplogroup was examined in 108 populations and 6,697 persons, and sequencing studies used either the complete mtDNA genome or the highly variable segments 1 and 2. The diversity of the markers made it difficult to establish a general picture of Y-chromosome variability in the populations studied. However, haplogroup Q1a3a* was almost always the most prevalent whereas Q1a3* occurred equally in all regions, which suggested its prevalence among the early colonizers. The STR allele frequencies were used to derive a possible ancient Native American Q-clade chromosome haplotype and five of six STR loci showed significant geographic variation. Geographic and linguistic factors moderately influenced the mtDNA distributions (6% and 7%, respectively) and mtDNA haplogroups A and D correlated positively and negatively, respectively, with latitude. The data analyzed here provide rich material for understanding the biological history of South Amerindians and can serve as a basis for comparative studies involving other types of data, such as cultural data.

Key words: genetics, language and geography, mitochondrial DNA, Native Americans, South Amerindians, Y-chromosome.

Received: September 23, 2011; Accepted: January 12, 2012.

Introduction

Native Americans have been the subject of a large number of population genetic studies because of particular characteristics: (a) there are groups among them that until recently had a hunter-gatherer way of living with only incipient agriculture, typical of our ancestors, (b) they show considerable interpopulation but low intrapopulation variability, and (c) since until recently they could not write there is no written record of their history, except for those of non-Amerindian colonizers. Biological studies can therefore be used to investigate their past.

The first genetic studies examined the variability in blood groups and proteins and have been summarized in Salzano and Callegari-Jacques (1988) and Crawford (1998). The advent of modern molecular biology, which allows direct, detailed DNA analysis, has opened new possibilities for investigating these populations.

DNA studies can basically be divided into two groups: those involving autosomal markers and those involving uniparental (Y-chromosome, mitochondrial DNA) markers. The latter are important because they can provide a clear-cut pattern of historical events that is not clouded by recombination factors. For Amerindians, the number of reviews that have dealt with these markers is not large or comprehensive. For the Y-chromosome, Bortolini et al. (2003) considered 438 individuals from 23 Southern and one Northern Amerindian populations who were screened for eight single nucleotide polymorphisms (SNPs) and six short tandem repeat/microsatellite (STR) loci, and Zegura et al. (2004) studied 63 binary polymorphisms and 10 STR regions in 2,344 persons from 15 Northern and three Southern Amerindian groups. Only a few recent studies have used all known SNPs necessary to identify the major Native American Y-haplogroups and their sublineages in Amerindian populations (Geppert et al., 2011; Jota et al., 2011; Bisso-Machado et al., 2011).

The most recent mtDNA reviews were published four years ago and involved sequence variability in the hypervariable region 1 (Hunley et al., 2007; Lewis Jr et al., 2007). Schurr and Sherry (2004), on the other hand, associated data from Y-chromosome markers with mitochondrial DNA (mtDNA) results, providing a good picture of the information available at the time. No general review considering both data sets has been published since then.

This review provides a detailed, comprehensive survey of Y-chromosome haplogroup frequency variation in 68 populations involving 1,814 individuals. In addition,
specific information on Y-STR markers for 29 populations and 590 subjects is given. The haplogroup mtDNA data included 108 populations involving a total of 6,697 persons. Geographic and linguistic factors that may have influenced this variation were carefully considered, leading to a global, overview of the genetic pattern associated with these markers in South Amerindians. Information on mtDNA sequencing studies is also supplied.

Materials and Methods

The data used in this review were obtained from 17 primary surveys of the Y-chromosome and 66 primary surveys of mtDNA. These studies were retrieved through PubMed and by searching the reference lists of the corresponding papers. Haplogroup frequencies were obtained by direct counting. Intra- and inter-populational diversity was calculated with AMOVA (Weir and Cockerham, 1984; Excoffier et al., 1992; Weir, 1996) using Arlequin 3.5.1.2 software (Excoffier and Lischer, 2010). AMOVA was also used to estimate the level of differentiation between and within 17 pre-defined language and 7 geographical categories, respectively. The distribution patterns of the mtDNA haplogroup frequencies were established by generating isoline maps using IDRISI 16.0 software (IDRISI Taiga) (Eastman, 2006). Spearman’s correlation coefficients were calculated with PASW Statistics 18 software. Average heterozygosity (ah) was calculated with Arlequin 3.5.1.2 software.

Results and Discussion

Table 1 gives the distribution of the Q and non-Q chromosomes (defined by a set of SNPs), as well as linguistic and geographical information for the samples considered. The samples were distributed from latitude 11° North to 45° South and longitude 46° to 76° West, with the individuals involved speaking 23 languages. Sample sizes varied widely from 1 to 151 individuals. Twenty-two of the studies involved less than 10 persons. Unfortunately, there is no standardization on the number of SNPs studied and in most cases only the M242 and M3 markers (which define the Asian/Native American paragroup Q* and its autochthonous Native American sublineage Q1a3a*, respect-
| Populations (n) | Haplogroup (%) | Language | Geographical coordinates | References |
|----------------|----------------|----------|--------------------------|------------|
| Yagua (7)      | 100            | Peba-Yaguan | 0° 51' N; 72° 27' W | Bortolini et al. (2003) |
| Ingano (108)   | 80             | Quechuan | 0° 50' N; 77° W | Bortolini et al. (2003); Rojas et al. (2010) |
| Wai-Wai (9)    | 100            | Carib | 0° 40' S; 58° W | Bisso-Machado et al. (2011) |
| Urubu-Kaapor (16) | 100        | Tupi | 2° -3° S; 46° -47° W | Bortolini et al. (2003) |
| Huitoto (4)    | 75             | Witotoan | 2° 14' S; 72° 19' W | Bortolini et al. (2003) |
| Arara (15)     | 100            | Carib | 3° 30' -4° 20' S; 53° 0' -54° 10' W | Rodriguez-Delfin et al. (1997); Bianchi et al. (1998); Bisso-Machado et al. (2011) |
| Asurini (4)    | 100            | Tupi | 3° 35' -4° 12' S; 49° 40' -52° 26' W | Bortolini et al. (2003) |
| Ticuna (59)    | 93             | Ticuna | 4° S; 69° 58' W | Bortolini et al. (2003); Rojas et al. (2010) |
| Parakanã (20)  | 100            | Tupi | 5° 22' S; 51° 17' W | Bortolini et al. (2003) |
| Xikrin (14)    | 100            | Macro-Ge | 5° 55' S; 51° W | Bortolini et al. (2003); Bisso-Machado et al. (2011) |
| Suruí (24)     | 96             | Tupi | 5° 58' -10° 50' S; 48° 39' -61° 10' W | Underhill et al. (1996); Bisso-Machado et al. (2011) |
| Araweté (4)    | 100            | Tupi | 5° 9' S; 52° 22' W | Bisso-Machado et al. (2011) |
| Munduruku (1)  | 100            | Tupi | 6° 23' S; 59° 9' W | Bisso-Machado et al. (2011) |
| Jamamadi (3)   | 100            | Arauan | 7° 15' S; 66° 41' W | Bisso-Machado et al. (2011) |
| Gorotire (19)  | 100            | Macro-Ge | 7° 44' S; 51° 10' W | Bortolini et al. (2003); Bisso-Machado et al. (2011) |
| Krahó (15)     | 93             | Macro-Ge | 8° S; 47° 15' W | Lel et al. (2002); Bortolini et al. (2003) |
| Kuben-Kran-Kegn (9) | 100    | Macro-Ge | 8° 10' S; 52° 8' W | Bisso-Machado et al. (2011) |
| Tenharim (1)   | 100            | Tupi | 8° 20' S; 62° W | Bisso-Machado et al. (2011) |
| Mekranoti (9)  | 78             | Macro-Ge | 8° 40' S; 54° W | Bortolini et al. (2003) |
| Kayapó (10)    | 100            | Macro-Ge | 9° S; 53° W | Rodriguez-Delfin et al. (1997) |
| Karitiana (18) | 100            | Tupi | 9° 30' S; 64° 15' W | Underhill et al. (1996); Bisso-Machado et al. (2011) |
| Cinta-Larga (15) | 100        | Tupi | 9° 50' –12° 30' S; 59° 10' –60° 50' W | Bortolini et al. (2003) |
| Gavião (7)     | 100            | Tupi | 10° 10' S; 61° 8' W | Bisso-Machado et al. (2011) |
| Karipuna (1)   | 100            | Tupi | 10° 14' S; 64° 13' W | Bisso-Machado et al. (2011) |
| Zoró (6)       | 100            | Tupi | 10° 20' S; 60° 20' W | Bisso-Machado et al. (2011) |
| Matsiguenga (28) | 91          | Arawakan | 10° 47' -12° 51' S; 73° 17' -70° 44' W | Mazières et al. (2008) |
| Pacaís Novos (Warí) (29) | 100 | Chapacura-Wanham | 11° 8' S; 65° W | Bortolini et al. (2003) |
| Panoa (5)      | 100            | Pano | 12° 55' S; 65° 12' W | Lel et al. (2002) |
| Xavante (15)   | 100            | Macro-Ge | 14° S; 52° 30' W | Bisso-Machado et al. (2011) |
| Quechua (44)   | 73             | Quechuan | 14° 30' S; 69° W | Gayà-Vidal et al. (2011) |
| Aymara (59)    | 97             | Aymaran | 17° 68' S; 69° 16' W | Gayà-Vidal et al. (2011) |
| Ayoreo (9)     | 78             | Zamucoan | 19° S; 60° 30' W | Bailleil et al. (2009) |
| Wichí (Mataco) (151) | 48    | Mataco-Guaicuru | 22° 28' S; 62° 70' W | DeMarchi and Mitchell (2004); Bailleil et al. (2009) |
| Lengua (36)    | 97             | Mascoian | 22° 45' S; 58° 5' W | Bailleil et al. (2009); Bisso-Machado et al. (2011) |
| Chorote (9)    | 89             | Mataco-Guaicuru | 22° 90' S; 65° 40' W; 23° 30' –24° 10' S; 55° 50' –56° 30' W | Bortolini et al. (2009) |
| Aché (54)      | 98             | Tupi | 23° 30' –24° 10' S; 55° 50' –56° 30' W | Bortolini et al. (2003) |
Table 1 (cont).

| Populations (n)   | Haplogroup (%) | Language¹ | Geographical coordinates | References                  |
|------------------|----------------|-----------|--------------------------|-----------------------------|
|                  | Q lineages /   |           |                          |                             |
|                  | Amerindian     |           |                          |                             |
|                  | Non-Q          |           |                          |                             |
|                  | lineages       |           |                          |                             |
| Guarani (78)     | 77             | 23        | Tupi                     | 23° 6’ S; 55° 12’ W         | Bortolini et al. (2003); Marrero et al. (2007) |
| Pilagá (63)      | 47             | 53        | Mataco-Guaicuru          | 24° 8’ S; 59° 0’ W          | Demarchi and Mitchell (2004) |
| Colla (63)       | 35             | 65        | Quechuan                 | 24° 4’ - 24° 43’ S          | Blanco-Verea et al. (2010); Toscanini et al. (2011) |
| Toba (89)        | 88             | 12        | Mataco-Guaicuru          | 26° 4’ S; 58° 0’ W          | Demarchi and Mitchell (2004); Bailliet et al. (2009); Toscanini et al. (2011) |
| Kaingang (59)    | 69             | 31        | Macro-Ge                 | 28° 8’ S; 51° 20’ W         | Bortolini et al. (2003); Marrero et al. (2007); Bisso-Machado et al. (2011) |
| Diaguita (24)    | 37             | 63        | Quechuan¹               | 28° 20’ S; 67° 43’ W        | Blanco-Verea et al. (2010) |
| Mocovi (40)      | 60             | 40        | Mataco-Guaicuru          | 29° 51’ S; 59° 56’ W        | Bailliet et al. (2009) |
| Pehuencche (18)  | 83             | 17        | Araucanian               | 37° 43’ S; 71° 16’ W        | Bailliet et al. (2009) |
| Mapuche (105)    | 36             | 64        | Araucanian               | 39° 10’ - 41° 20’ S; 68° 37’ - 70° 22’ W | Bailliet et al. (2009); Blanco-Verea et al. (2010) |
| Huilliche (26)   | 50             | 50        | Araucanian               | 41° 16’ S; 73° W           | Bailliet et al. (2009) |
| Tehuelche (20)   | 65             | 35        | Chon                     | 45° S; 71° W               | Bailliet et al. (2009) |

¹Arranged according to latitude. ²Classification according to Lewis (2009). ³Original language is extinct. ⁴The Diaguita spoke originally Kakán, but this language became extinct and was substituted by Quechua.

The nature of some evolutionary and demographic scenarios, mediated by men, in native American populations has also been evaluated by using Y microsatellite markers (Y-STRs), which have a much faster evolutionary rate than SNPs. Y-STRs allow the retrieval of population and chromosome evolutionary histories. For example, STR data have been used to estimate that the mutations that gave rise to the Q1a3a1 and Q1a3a4 sublineages occurred 7,972 ± 2,916 and 5,280 ± 1,330 years ago, probably in northwest South America and the Andean region, respectively (Bortolini et al., 2003; Jota et al., 2011).

Table 2 shows the STR allele frequencies observed in 29 South Amerindian populations, based only on Q clade chromosomes. In this compilation, we considered only studies containing information on the allele frequencies for each population individually. There was considerable variation in the number of samples tested in each study, the number of tribes, and the number of individuals per tribe. Depending on the locus considered, the number of alleles observed ranged from one to eight, with some of them appearing in only one study while others were present in almost all populations. Based on the most prevalent alleles per locus we reconstructed a probable haplotype of the ancient Native American Q-clade chromosome (ANAQC) as: 13(DYS19)-12(DYS388)-14(DYS389I)-31(DYS389II)-2.
**Figure 1** - Y-chromosome phylogenetic tree considering only the Q derived lineages. Note: The letters and numbers in the branches indicate the name of the loci where the mutations occurred, leading to the haplogroup classification. The data for this tree were compiled from the references in Table 1, plus Santos et al. (1995), Underhill et al. (1997, 2001), Karafet et al. (1997, 1999, 2008), Carvalho-Silva et al. (1999), Vallinoto et al. (1999), Bortolini et al. (2002), The Y-Chromosome Consortium (2002) and Geppert et al. (2011).

**Table 2 (a)** - Y-Q-chromosome STR studies in distinct South Amerindian samples in which allele frequencies can be assessed (Part A).
| STR (allele) | Aché (48)
| Apalaí (9)
| Arara (8)
| Ayoreo (21)
| Barira (12)
| Diaguita (9)
| Guarani (47)²
| Ingano (8)
| Kaingang (16)
| Kayapó (10)
| Colla (22)²
| Lenguá (6)
| Mapuche (24)²
| Mekranoti (5) |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| DYS390 (20) | 0.230 | 0.120 | 0.070 | | | | | | | | | | |
| DYS390 (21) | 0.110 | | | | | | | | | | | | |
| DYS390 (22) | 1.000 | 0.330 | 0.880 | 0.560 | 0.080 | 0.330 | 0.080 | 0.250 | 0.100 | 0.640 | 0.330 | 0.420 | 0.400 |
| DYS390 (23) | 0.330 | 0.250 | 1.000 | 0.920 | 0.450 | 0.680 | 0.370 | 0.940 | 0.700 | 0.180 | 0.670 | 0.330 | 0.600 |
| DYS390 (24) | 0.120 | 0.220 | 0.090 | 0.250 | 0.100 | | | | | | | | 0.210 |
| DYS390 (25) | | | | | | | | | | | | | |
| DYS390 (26) | 0.020 | 0.130 | | | | | | | | | | | 0.140 |
| DYS390 (27) | | | | | | | | | | | | | |
| DYS390 total | | | | | | | | | | | | | |
| DYS391 (9) | 0.020 | | | | | | | | | | | | 0.210 |
| DYS391 (10) | 1.000 | 0.820 | 1.000 | 1.000 | 0.890 | 0.210 | 0.860 | | 0.820 | 1.000 | 0.750 | 1.000 |
| DYS391 (11) | 0.140 | 0.110 | 0.790 | 0.140 | | | | | | 0.180 | 0.040 |
| DYS391 (12) | 0.020 | | | | | | | | | | | | |
| DYS391 total | | | | | | | | | | | | | |
| DYS392 (11) | 0.060 | | | | | | | | | | | | |
| DYS392 (12) | 0.140 | | | | | | | | | | | | |
| DYS392 (13) | 1.000 | 1.000 | 0.920 | 0.070 | 0.120 | 0.350 | 0.700 | 0.050 | | 0.170 | 1.000 |
| DYS392 (14) | 1.000 | 0.470 | 0.500 | 0.080 | 0.450 | 0.720 | 0.880 | 0.530 | 0.300 | 0.360 | 0.830 | 0.710 |
| DYS392 (15) | 0.040 | 0.500 | 0.330 | 0.210 | | 0.060 | | 0.090 | 0.170 | 0.120 |
| DYS392 (16) | 0.470 | 0.220 | | | | | | | | | | | 0.360 |
| DYS392 (17) | 0.020 | | | | | | | | | | | | |
| DYS392 (18) | | | | | | | | | | | | | |
| DYS392 total | | | | | | | | | | | | | |
| DYS393 (11) | | | | | | | | | | | | | 0.480 |
| DYS393 (12) | 0.220 | 0.120 | 0.080 | 0.090 | 0.120 | 0.060 | 0.400 | 0.090 | | 0.400 |
| DYS393 (13) | 1.000 | 0.780 | 0.880 | 0.440 | 1.000 | 0.670 | 0.340 | 0.760 | 0.590 | 0.600 | 0.550 | 1.000 | 1.000 | 0.600 |
| DYS393 (14) | 0.560 | 0.920 | 0.330 | 0.090 | 0.120 | 0.350 | 0.360 | | | | | | | |
| DYS393 (15) | | | | | | | | | | | | | |
| DYS393 (16) | | | | | | | | | | | | | |
| DYS393 total | | | | | | | | | | | | | |
| DYS437 (8) | | | | | | | | | | | | | |
| DYS437 (9) | | | | | | | | | | | | | |
| DYS437 (11) | | | | | | | | | | | | | 0.040 |
| DYS437 (12) | 1.000 | 1.000 | 0.930 | 0.590 | 1.000 | | | | | | | 0.920 |
| DYS437 (15) | 0.070 | 0.410 | | | | | | | | | | | 0.040 |
| DYS437 total | | | | | | | | | | | | | |
| DYS438 (9) | | | | | | | | | | | | | |
| DYS438 (10) | 0.050 | 0.220 | | | | | | | | | | | 0.040 |
| DYS438 (11) | 0.930 | 0.670 | 0.860 | 0.590 | 1.000 | | | | | | | 0.920 |
| DYS438 (12) | 0.020 | 0.110 | 0.140 | | | | | | | | | | 0.410 |
| DYS438 (16) | | | | | | | | | | | | | |
| DYS438 total | | | | | | | | | | | | | |
| DYS439 (9) | | | | | | | | | | | | | |
| DYS439 (10) | | | | | | | | | | | | | 0.060 | 0.130 |
| DYS439 (11) | 0.160 | 0.220 | 0.070 | 0.230 | 0.050 | 0.220 | | | | | | | |
| DYS439 (12) | 0.240 | 0.450 | 0.640 | 0.590 | | 0.360 | 0.390 | | | | | | |
| DYS439 (13) | 0.370 | 0.330 | 0.290 | 0.060 | 0.270 | 0.260 | | | | | | | |

Table 2 (a) (cont.)
Table 2 (a) (cont.)

| STR (allele) | Aché (48) | Apalaí (9) | Arara (8) | Ayoreo (2) | Bartra (12) | Diaguita (9) | Guarani (47) | Ingano (8) | Kaingang (17) | Kayapó (10) | Colla (22) | Lengua (6) | Mapuche (24) | Mekranoti (5) |
|--------------|-----------|-----------|-----------|------------|------------|-------------|-------------|-----------|--------------|-------------|-----------|-----------|-------------|------------|
| DYS439 (14)  | 0.230     |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS439 total |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS448 (18)  |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS448 (19)  |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS448 (20)  |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS448 (21)  |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS448 (22)  |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS448 total |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS456 (11)  |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS456 (12)  |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS456 (13)  |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS456 (14)  |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS456 (15)  |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS456 (16)  |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS456 (17)  |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS456 total |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS458 (11)  |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS458 (12)  |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS458 (13)  |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS458 (14)  |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS458 (15)  |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS458 (16)  |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS458 (17)  |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS458 (18)  |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS458 (19)  |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS458 total |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS635 (21)  |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS635 (22)  |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS635 (23)  |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS635 (24)  |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS635 (26)  |           |           |           |            |            |             |             |           |              |             |           |           |             |            |
| DYS635 total |           |           |           |            |            |             |             |           |              |             |           |           |             |            |

Table 2 (b) – Y-Q-chromosome STR studies in distinct South Amerindian samples in which allele frequencies can be assessed (Part B).

Ref. (n)

| STR (allele) | Mocoví (22) | Pacais Novus (15) | Parakanã (4) | Pilagá (9) | Quechuana (58) | Ticuna (36) | Toba (70) | Warao (12) | Wayampi (10) | Wayuu (14) | Wichí (27) | Yanomama (9) | Yukpa (12) | Zenu (28) | Total (590) |
|--------------|-------------|------------------|-------------|------------|----------------|-------------|-----------|------------|--------------|------------|-----------|---------------|-----------|-----------|-------------|
| DYS19 (12)   | 0.030       |                  |             |            |                |             |           |            |              |            |           |               |           |           | 0.010       |
| DYS19 (13)   | 0.500       |            0.790 | 0.950      | 0.850      | 0.870          | 1.000       |            | 0.790      | 0.640        | 0.550      | 0.650      | 0.890          |           |           |             |
| DYS19 (14)   | 0.500       | 1.000          | 0.210       | 0.030      | 0.090          | 0.090       |            | 0.140      | 0.230        | 0.450      | 0.270      | 0.050          |           |           |             |
| DYS19 (15)   |             |                | 0.020       | 0.060      | 0.010          |            |           | 0.070      | 0.100        |            |            | 0.080          | 0.040     |           |             |
| DYS19 (16)   |             |                  |             |            |                |             |           |            |              |            |           |               |           |           | 0.010       |
| DYS19 total  |             |                  |             |            |                |             |           |            |              |            |           |               |           |           | 474         |
| DYS388 (12)  | 0.860       |                  | 1.000       |            | 0.780          | 0.750       |            | 0.570      |              | 0.500      | 0.500      | 0.780          |           |           |             |
| DYS388 (13)  | 0.140       |                  |            |            | 0.220          | 0.250       |            | 0.430      |              | 0.500      | 0.430      | 0.200          |           |           |             |
| DYS388 (14)  |             |                  |             |            |                |             |           |            |              |            |            | 0.010          |           |           |             |
| DYS388 (17)  |             |                  |             |            |                |             |           |            |              |            |            | 0.070          | 0.040     |           |             |
| DYS388 total |             |                  |             |            |                |             |           |            |              |            |            | 189           |           |           |             |
| DYS389I (12) |             |                  |             |            |                |             |           |            |              |            |            | 0.090          |           | 0.080     |             |
| DYS389I (13) |             |                  |             |            |                |             |           |            |              |            |            | 1.000          | 0.240     | 0.800     | 0.440       |

1Rodriguez-Delfín et al. (1997); 2Bortolini et al. (2003); 3Demarchi and Mitchell (2004); 4Altuna et al. (2006); 5Leite et al. (2008); 6Toscanini et al. (2008); 7Baillet et al. (2009); 8Blanco-Veréa et al. (2010); 9Gayà-Vidal et al. (2011); 10Jota et al. (2011).
Table 2 (b) (cont.)

| STR (allele) | Ref. (n) |
|--------------|----------|
| DYS389I (14) | 0.670 0.200 0.450 |
| DYS389I (15) | 0.030 |
| DYS389I total | 288 |
| DYS389II (17) | 0.270 0.020 0.390 |
| DYS389II (18) | 0.530 0.160 |
| DYS389II (19) | 0.200 0.100 |
| DYS389II (26) | 0.010 |
| DYS389II (27) | 0.010 |
| DYS389II (28) | 0.090 0.030 |
| DYS389II (29) | 0.070 0.110 |
| DYS389II (30) | 0.210 0.500 0.290 |
| DYS389II (31) | 0.500 0.320 0.390 |
| DYS389II (32) | 0.500 0.160 0.080 |
| DYS389II (33) | 0.020 0.010 0.010 |
| DYS389II (34) | 0.020 0.010 0.010 |
| DYS389II total | 288 |
| DYS390 (20) | 0.020 0.010 |
| DYS390 (21) | 0.240 0.070 |
| DYS390 (22) | 0.020 0.080 0.030 0.780 0.070 |
| DYS390 (23) | 0.500 0.210 0.450 0.060 0.110 0.080 1.000 0.220 0.070 |
| DYS390 (24) | 0.500 0.050 0.550 1.000 0.790 0.220 0.150 0.730 0.920 0.560 0.740 0.110 1.000 0.290 0.400 |
| DYS390 (25) | 0.050 0.760 0.080 0.220 0.130 0.420 0.110 0.100 |
| DYS390 (26) | 0.030 0.010 |
| DYS390 (27) | 0.030 0.110 0.040 0.010 |
| DYS390 total | 659 |
| DYS391 (9) | 0.070 0.030 0.030 |
| DYS391 (10) | 1.000 1.000 0.890 0.840 0.930 0.900 0.250 0.570 0.870 1.000 0.640 0.800 |
| DYS391 (11) | 1.000 0.110 0.160 0.070 0.100 0.750 0.360 0.100 0.360 0.160 |
| DYS391 (12) | 0.010 |
| DYS391 total | 512 |
| DYS392 (11) | 0.030 0.570 0.060 0.020 |
| DYS392 (12) | 1.000 0.070 0.130 0.890 |
| DYS392 (13) | 1.000 0.890 0.100 0.100 0.110 0.130 0.890 0.180 0.060 0.170 |
| DYS392 (14) | 0.110 0.800 0.520 0.380 0.670 0.890 0.290 0.420 0.110 0.460 0.560 |
| DYS392 (15) | 0.100 0.070 0.620 0.190 0.070 0.390 0.360 0.250 0.150 |
| DYS392 (16) | 0.290 0.100 |
| DYS392 (17) | 0.020 0.100 |
| DYS392 (18) | 0.010 |
| DYS392 total | 535 |
| DYS393 (11) | 1.000 0.060 0.010 0.100 0.060 0.040 0.040 |
| DYS393 (12) | 1.000 0.780 1.000 0.550 0.710 0.980 0.750 0.800 0.840 0.890 0.500 0.200 0.670 |
| DYS393 (13) | 0.220 0.450 0.170 0.010 0.250 1.000 0.100 0.100 0.110 0.500 0.240 0.220 |
| DYS393 (15) | 0.030 0.480 0.020 |
| DYS393 (16) | 0.030 0.040 0.010 |
| DYS393 total | 584 |
## Table 2 (b) (cont.)

| STR (allele) | Mocoví (n) | Pacaás Novos (n) | Parakanã (n) | Pilagá (n) | Quechua (n) | Ticuna (n) | Toba (n) | Wayampi (n) | Wayuu (n) | Wichí (n) | Yanomama (n) | Yukpa (n) | Zenu (n) | Total (n) |
|--------------|------------|-----------------|-------------|------------|-------------|------------|----------|-------------|-----------|-----------|--------------|----------|---------|----------|
| DYS437 (8)   | 0.420      | 0.360           | 1.000       |             |             |            |          |              |           |           |               |          |         | 0.200    |
| DYS437 (9)   | 0.580      | 0.030           |             |            |             |            |          |              |           |           |               |          |         | 0.040    |
| DYS437 (11)  |             |                 |             |            |             |            |          |              |           |           |               |          |         | 0.010    |
| DYS437 (14)  | 1.000      | 0.500           |             |            |             |            |          |              |           |           |               |          |         | 0.700    |
| DYS437 (15)  |             | 0.110           |             |            |             |            |          |              |           |           |               |          |         | 0.050    |
| DYS437 total |             |                 |             |            |             |            |          |              |           |           |               |          |         | 322      |
| DYS438 (9)   |             |                 |             |            |             |            |          |              |           |           |               |          |         | 0.010    |
| DYS438 (10)  |             |                 |             | 0.050      |             |            |          |              |           |           |               | 0.160    |         | 0.050    |
| DYS438 (11)  | 0.790      | 0.930           | 0.850       |             | 0.840       |            |          |              |           |           |               |          |         | 0.850    |
| DYS438 (12)  | 0.210      | 0.050           | 0.100       |             |             |            |          |              |           |           |               |          |         | 0.080    |
| DYS438 (16)  |             |                 |             |            |             |            |          |              |           |           |               |          |         | 0.010    |
| DYS438 total |             |                 |             |            |             |            |          |              |           |           |               | 322      |         |          |
| DYS439 (9)   |             |                 |             |            |             |            |          |              |           |           |               | 0.010    |         |          |
| DYS439 (10)  |             |                 |             |            |             |            |          |              |           |           |               | 0.010    |         |          |
| DYS439 (11)  | 0.090      | 0.130           |             | 0.230      |             |            |          |              |           |           |               | 0.140    |         |          |
| DYS439 (12)  | 0.320      | 0.240           | 0.500       |             | 0.710       |            |          |              |           |           |               | 0.400    |         |          |
| DYS439 (13)  | 0.680      | 0.500           | 0.290       |             | 0.060       |            |          |              |           |           |               | 0.330    |         |          |
| DYS439 (14)  | 0.170      | 0.080           |             |            |             |            |          |              |           |           |               | 0.110    |         |          |
| DYS439 total |             |                 |             |            |             |            |          |              |           |           |               | 322      |         |          |
| DYS448 (18)  |             |                 |             |            |             |            |          |              |           |           |               | 0.010    |         |          |
| DYS448 (19)  | 0.140      | 0.020           |             |            |             |            |          |              |           |           |               | 0.090    |         |          |
| DYS448 (20)  | 0.550      | 0.930           |             |            |             |            |          |              |           |           |               | 0.740    |         |          |
| DYS448 (21)  | 0.240      | 0.050           |             |            |             |            |          |              |           |           |               | 0.140    |         |          |
| DYS448 (22)  | 0.070      |                 |             |            |             |            |          |              |           |           |               | 0.020    |         |          |
| DYS448 total |             |                 |             |            |             |            |          |              |           |           |               | 169      |         |          |
| DYS456 (11)  |             |                 |             |            |             |            |          |              |           |           |               | 0.010    |         |          |
| DYS456 (13)  |             |                 |             |            |             |            |          |              |           |           |               | 0.020    |         |          |
| DYS456 (14)  | 0.230      | 0.020           |             |            |             |            |          |              |           |           |               | 0.090    |         |          |
| DYS456 (15)  | 0.720      | 0.730           |             |            |             |            |          |              |           |           |               | 0.750    |         |          |
| DYS456 (16)  | 0.050      | 0.250           |             |            |             |            |          |              |           |           |               | 0.120    |         |          |
| DYS456 (17)  |             |                 |             |            |             |            |          |              |           |           |               | 0.010    |         |          |
| DYS456 total |             |                 |             |            |             |            |          |              |           |           |               | 169      |         |          |
| DYS458 (13)  |             |                 |             |            |             |            |          |              |           |           |               | 0.020    |         |          |
| DYS458 (15)  | 0.030      | 0.040           |             |            |             |            |          |              |           |           |               | 0.040    |         |          |
| DYS458 (16)  | 0.360      | 0.180           |             |            |             |            |          |              |           |           |               | 0.380    |         |          |
| DYS458 (17)  | 0.520      | 0.550           |             |            |             |            |          |              |           |           |               | 0.410    |         |          |
| DYS458 (18)  | 0.090      | 0.230           |             |            |             |            |          |              |           |           |               | 0.140    |         |          |
| DYS458 (19)  |             |                 |             |            |             |            |          |              |           |           |               | 0.010    |         |          |
| DYS458 total |             |                 |             |            |             |            |          |              |           |           |               | 169      |         |          |
| DYS635 (22)  | 0.950      | 0.950           |             |            |             |            |          |              |           |           |               | 0.890    |         |          |
| DYS635 (23)  | 0.020      | 0.050           |             |            |             |            |          |              |           |           |               | 0.090    |         |          |
| DYS635 (24)  | 0.030      |                 |             |            |             |            |          |              |           |           |               | 0.010    |         |          |
| DYS635 (26)  |             |                 |             |            |             |            |          |              |           |           |               | 0.010    |         |          |
| DYS635 total |             |                 |             |            |             |            |          |              |           |           |               | 169      |         |          |

1Rodriguez-Delfin *et al.* (1997); 2Bortolini *et al.* (2003); 3Demarchi and Mitchell (2004); 4Altuna *et al.* (2006); 5Leite *et al.* (2008); 6Toscanini *et al.* (2008); 7Bailliet *et al.* (2009); 8Blanco-Verea *et al.* (2010); 9Gayà-Vidal *et al.* (2011); 10Jota *et al.* (2011).
Fagundes et al. complete Amerindian genomes, Fagundes interethnic or interhaplogroup comparisons. Based on 86 Amerindian relationships and were mostly concerned with the analyses performed did not consider the within South American individual (Kaqchiquel).

Table 3 shows the results of the molecular analysis of variance for populations structured by language or geography based on the data in Table 2. The estimates were calculated for each STR locus because testing heterogeneity prevented haplotype identification. As expected, most of the diversity was attributable to intrapopulation variation, with one exception (DYS347) that was explained by the fixation of allele 14 in 40% of the populations, whereas only allele 8 was found in the Wichí. In contrast, significant variation among subdivisions was detected for only six loci (DYS398I, DYS391, DYS392, DYS393, DYS437 and DYS456) and in five out of these six it was attributable to geography. There was also considerable inter-population/within subdivision variability (significant in 28 of 30 evaluations), with the average percentage being 16% for geography and 21% for language.

Table 4 summarizes the information on sequencing studies of mitochondrial DNA. The mtDNA genome of representative individuals from 35 populations has been entirely sequenced, as reported in six publications (Ingman et al., 2000; Kivisild et al., 2006; Tamm et al., 2007; Fagundes et al., 2008; Perego et al., 2009, 2010). However, the analyses performed did not consider the within South Amerindian relationships and were mostly concerned with interethnic or interhaplogroup comparisons. Based on 86 complete Amerindian genomes, Fagundes et al. (2008) concluded that the prehistoric colonization of the Americas involved a single founding population, with an initial differentiation from Asia occurring in Beringia that ended around 19,000-23,000 years ago, with a moderate bottleneck. Expansion into the New World would have occurred about 18,000 years ago. An extensive 5.76 kb analysis by Dornelles et al. (2005) established that haplogroup X is not present in extant South American Indians. The most extensive set of data involves the highly variable segment 1 (HVS-I) that has been studied in 92 populations and reported in 30 papers; surveys that have included the HVS-II region are much less common (10 articles) (Table 4). For HVS-I, Merriwether et al. (2000) provided an excellent example of how intrapopulation variability in the Yanomamí could be interpreted in a historical and demographical context and relating it to other Amerindian and Asian data. They studied 129 Yanomamí sequences from individuals in eight villages and compared their haplotypes with those of other Asian and New World populations, in a total of 482 unique haplotypes. Interestingly, the pairwise inter-population gene flow estimates were lower between some pairs of Yanomamí villages than between them and four other South Amerindian groups.

With regard to intrapopulation variability, as measured by $\Theta_a$, Fuselli et al. (2003) and Corella et al. (2007) reported extensive variation for 14 and 27 Central and

Table 3 - Analysis of molecular variance of the distinct alleles of the Y-Q STRs in relation to the language and geography of the populations tested.

| STR loci (structured by) | Among subdivisions | Among populations within subdivisions | Within populations |
|--------------------------|--------------------|--------------------------------------|--------------------|
| DYS19 (Language)¹ | 0.311* | 0.689* |
| DYS19 (Geography)² | 0.332* | 0.668* |
| DYS388 (Language)¹ | 0.145 | 0.317* | 0.638* |
| DYS388 (Geography)² | 0.091 | 0.240* | 0.669* |
| DYS391 (Language)¹ | 0.227* | 0.030 | 0.743* |
| DYS391 (Geography)² | 0.116 | 0.148* | 0.736* |
| DYS391I (Language)¹ | 0.036 | 0.077* | 0.887* |
| DYS391I (Geography)² | 0.125* | 0.875 |
| DYS390 (Language)¹ | 0.326* | 0.666* |
| DYS390 (Geography)² | 0.359* | 0.642* |
| DYS391 (Language)¹ | 0.385* | 0.615* |
| DYS391 (Geography)² | 0.131* | 0.616* |
| DYS392 (Language)¹ | 0.005 | 0.319* | 0.676* |
| DYS392 (Geography)² | 0.007* | 0.262* | 0.661* |
| DYS393 (Language)¹ | 0.331* | 0.669* |
| DYS393 (Geography)² | 0.177* | 0.167* | 0.656* |
| DYS437 (Language)¹ | 0.289 | 0.273* | 0.438* |
| DYS437 (Geography)² | 0.390* | 0.213* | 0.397* |
| DYS438 (Language)¹ | 0.048 | 0.025 | 0.927* |
| DYS438 (Geography)² | 0.023 | 0.054* | 0.923* |
| DYS439 (Language)¹ | 0.019 | 0.054* | 0.929* |
| DYS439 (Geography)² | 0.055 | 0.034* | 0.911* |
| DYS448 (Language)¹ | 0.148* | 0.852* |
| DYS448 (Geography)² | 0.073* | 0.927* |
| DYS456 (Language)¹ | 0.078* | 0.922* |
| DYS456 (Geography)² | 0.017* | 0.044* | 0.939* |
| DYS458 (Language)¹ | 0.144* | 0.856* |
| DYS458 (Geography)² | 0.043 | 0.069* | 0.888* |
| DYS635 (Language)¹ | 0.278* | 0.722* |
| DYS635 (Geography)² | 0.128* | 0.872* |

¹Language: Tupi: Aché, Guaraní, Parakanã, Wayampí; Carib: Apalai, Arara, Yukpa; Macro-Ge: Kaingang, Kayapó, Mekranoti; Quechua: Diaigua, Quechua, Ingano; Mataco-Guaicuru: Mocoví, Toba, Wichí, Pilagá; Isolated languages and others with only one population were included as a sixth group.

²Geography: Amazonia/Central Brazilian Plateau: Apalai, Arara, Kayapó, Mekranoti, Pacaís Novos, Parakanã, Ticuna, Warao, Wayampí, Yanomamí; Southern Brazil: Guarani and Kaingang; Chaco: Ache, Ayoreo, Lenguá, Mocovi, Pilagá, Toba, Wichí; Andes: Aymara, Bari, Diaigua, Kolla, Quechua, Wayuu, Yukpa, Zenu.

*Significant values ($p < 0.05$). Negative values were adjusted to zero.
Table 4 - Mitochondrial DNA sequencing studies in South Amerindian populations.

| Population         | HVS-I | HVS-II | Complete mt genome | References                                                                 |
|--------------------|-------|--------|---------------------|---------------------------------------------------------------------------|
| Aché               | X     | X      | X                   | Schmitt et al. (2004); Dornelles et al. (2005); Fagundes et al. (2008); Yang et al. (2010) |
| Ancash             | X     |        |                     | Lewis Jr et al. (2005); Perego et al. (2009)                               |
| Andean             | X     |        |                     | García-Bour et al. (2004)                                                 |
| Apalai             | X     |        |                     | Lobato-da-Silva et al. (2001); Mazières et al. (2008)                     |
| Arara              | X     | X      |                     | Santos et al. (1996); Lobato-da-Silva et al. (2001); Ribeiro-dos-Santos et al. (2001); Silva Jr et al. (2002, 2003); Fagundes et al. (2008) |
| Araweté            | X     |        |                     | Lobato-da-Silva et al. (2001)                                             |
| Arequipa           | X     |        |                     | Fuselli et al. (2003); Perego et al. (2009)                                |
| Arhuaco            | X     | X      | X                   | Melton et al. (2007); Tamm et al. (2007); Yang et al. (2010)               |
| Arsario            | X     |        | X                   | Melton et al. (2007); Tamm et al. (2007)                                   |
| Asurini            | X     | X      |                     | Lobato-da-Silva et al. (2001); Dornelles et al. (2005)                    |
| Auca               |       |        | X                   | Kivisild et al. (2006)                                                    |
| Awa-Guajá          | X     |        |                     | Santos et al. (1996); Lobato-da-Silva et al. (2001)                       |
| Awa-Juriti         | X     |        |                     | Lobato-da-Silva et al. (2001)                                             |
| Aymara             | X     | X      |                     | Corella et al. (2007); Lewis Jr et al. (2007); Yang et al. (2010); Barbieri et al. (2011) |
| Ayoreco            | X     | X      |                     | Dornelles et al. (2004, 2005)                                             |
| Catamarca          |       |        | X                   | Tamm et al. (2007)                                                        |
| Cayapa             | X     | X      | X                   | Rickards et al. (1999); Tamm et al. (2007)                                |
| Chimane            | X     |        |                     | Corella et al. (2007)                                                     |
| Chilean North Coast| X     |        |                     | Moraga et al. (2005)                                                      |
| Cinta Larga        | X     | X      |                     | Lobato-da-Silva et al. (2001); Dornelles et al. (2005)                    |
| Coreguaje          |       |        | X                   | Tamm et al. (2007)                                                        |
| Coya               | X     | X      |                     | Alvarez-Iglesias et al. (2007)                                            |
| Cuboe              | X     |        | X                   | Torres et al. (2006)                                                      |
| Curripaco          | X     | X      |                     | Torres et al. (2006)                                                      |
| Desano             | X     | X      |                     | Torres et al. (2006)                                                      |
| Diaguita           |       |        | X                   | Perego et al. (2010)                                                      |
| Embera             | X     | X      | X                   | Torres et al. (2006); Tamm et al. (2007)                                  |
| Emerillon          | X     |        |                     | Mazières et al. (2008)                                                    |
| Gavião             | X     |        | X                   | Ward et al. (1996); Fagundes et al. (2008)                                |
| Gorotire           | X     | X      |                     | Dornelles et al. (2005)                                                   |
| Guaíibo            | X     | X      |                     | Vona et al. (2005); Torres et al. (2006)                                  |
| Guarani            | X     | X      | X                   | Ingman et al. (2000); Silva Jr et al. (2002, 2003); Dornelles et al. (2005); Kivisild et al. (2006); Marrero et al. (2007); Fagundes et al. (2008); Sala et al. (2010); Yang et al. (2010) |
| Huilliche          | X     | X      |                     | Yang et al. (2010)                                                        |
| Huitoto            | X     | X      |                     | Monsalve et al. (1994); Torres et al. (2006)                               |
| Içana River Indians| X     | X      |                     | Dornelles et al. (2005)                                                   |
| Ignaciano          | X     |        |                     | Bert et al. (2004)                                                        |
| Inga               | X     | X      |                     | Torres et al. (2006); Yang et al. (2010)                                  |
| Jaqaru             | X     |        |                     | Lewis Jr et al. (2007)                                                    |
| Jamamadi           | X     |        |                     | Lobato-da-Silva et al. (2001)                                             |
| Jebero             | X     | X      |                     | Monsalve et al. (1994); Torres et al. (2006)                               |
| Kaingang           | X     | X      |                     | Dornelles et al. (2005); Marrero et al. (2007); Yang et al. (2010)        |
| Kall'a             | X     |        |                     | Mazières et al. (2008)                                                    |
| Karitiana          | X     |        |                     | Lobato-da-Silva et al. (2001); Kivisild et al. (2006)                      |
Table 4 (cont.)

| Population       | HVS-I | HVS-II | Complete mt genome | References |
|------------------|-------|--------|--------------------|------------|
| Katuena          | X     | X      | Santos et al. (1996); Lobato-da-Silva et al. (2001); Silva Jr et al. (2002, 2003); Fagundes et al. (2008) |
| Kayapó           | X     |        | Lobato-da-Silva et al. (2001); Silva Jr et al. (2002, 2003); Garcia-Bour et al. (2004) |
| Kikretun         | X     |        | Santos et al. (1996); Fagundes et al. (2008) |
| Kubenkokre       | X     | X      | Santos et al. (1996); Fagundes et al. (2008) |
| Mapuche          | X     | X      | Dornelles et al. (2005) |
| Mekranoti        | X     | X      | Dornelles et al. (2005) |
| Mocovi           | X     |        | Tamm et al. (2007) |
| Movima           | X     |        | Bert et al. (2004); Melton et al. (2007) |
| Munduruku        | X     |        | Lobato-da-Silva et al. (2001); Dornelles et al. (2005) |
| Ocaina           | X     | X      | Monsalve et al. (1994); Torres et al. (2006) |
| Paez             | X     | X      | Dornelles et al. (2005) |
| Palikur          | X     |        | Lobato-da-Silva et al. (2001); Mazieres et al. (2008) |
| Pehuenche        | X     | X      | Merriwether et al. (1994, 1995); Merriwether and Ferrell (1996); Moraga et al. (1999, 2000); Garcia et al. (2006) |
| Peruvian Andes   | X     | X      | Shinoda et al. (2006); Fehren-Schmitz et al. (2011) |
| Peruvian Southern Coast | X | | Fehren-Schmitz et al. (2010) |
| Piapoco          | X     | X      | Torres et al. (2006) |
| Pilagá           | X     |        | Cabana et al. (2006) |
| Poturujara       | X     | X      | Santos et al. (1996); Lobato-da-Silva et al. (2001); Silva Jr et al. (2002, 2003); Fagundes et al. (2008) |
| Puinave          | X     | X      | Torres et al. (2006) |
| Puno             | X     |        | Lewis Jr et al. (2007) |
| Quechua          | X     | X      | Monsalve et al. (1994); Silva Jr et al. (2002, 2003); Fuselli et al. (2003); Dornelles et al. (2005); Kivisild et al. (2006); Corella et al. (2007); Lewis Jr et al. (2007); Dornelles et al. (2005); Fagundes et al. (2008); Yang et al. (2010); Barbieri et al. (2011) |
| Saliva           | X     | X      | Torres et al. (2006) |
| Salta            | X     |        | Tamm et al. (2007) |
| Saterê Mawé      | X     | X      | Dornelles et al. (2005) |
| Selknam          | X     |        | Garcia-Bour et al. (2004) |
| Sicán            | X     |        | Shimada et al. (2004) |
| Surui            | X     | X      | Lobato-da-Silva et al. (2001); Fagundes et al. (2008) |
| Tayacaja         | X     |        | Bert et al. (2004) |
| Ticuna           | X     | X      | Monsalve et al. (1994); Torres et al. (2006); Rojas et al. (2010); Yang et al. (2010) |
| Tiriyo           | X     | X      | Santos et al. (1996); Lobato-da-Silva et al. (2001); Silva Jr et al. (2002, 2003); Dornelles et al. (2005); Fagundes et al. (2008) |
| Toba             | X     |        | Cabana et al. (2006) |
Southern Amerindian populations, respectively (e.g., from 0.659 for the Quechua of Peru to 0.011 for the Xavante of the Brazilian Mato Grosso). Intra- and intergroup nucleotide diversity was calculated by Melton et al. (2007) for 20 of these Amerindian groups, whereas Barbieri et al. (2011) compared the sources of variation among North, Central and South Amerindians in 51 populations; the latter authors observed 3% variation among the three sets, 21% variation among populations within the subcontinent and 76% variation within populations.

To explore the mtDNA data further we compiled the prevalences of haplogroups A-D for 109 populations, in a total of 6,697 individuals distributed between latitude 11° North and 54° South, and longitude 46° to 78° West (Table 5). Sample sizes varied widely, from only one subject tested (Jebero) up to 491 (Yanomámi). The haplogroup frequencies reported in 52 articles also varied widely. The presence of mtDNA genomes of probable non-Amerindian origin was rare in all regions and populations, in contrast to the Y-SNP data (Table 1). Asymmetrical sex-mediated admixture was common during the first centuries of South American colonization, and involved mostly European men and Amerindian/African women. The main consequences of this historical contact was the formation of mestizos and the present-day national societies; the former are characterized by a composite genome, with the majority of Y-chromosomes being of European origin, while their mtDNA derives from Amerindian or African sources (Bortolini et al., 1999; Alves-Silva et al., 2000; Carvalho-Silva et al., 2001; Salzano and Bortolini, 2002). Asymmetrical mating could also explain the introduction of non-Amerindian Y-chromosomes into the tribes, while the autochthonous mtDNA genomes were preserved. However, the admixture dynamics are probably different from those observed in urban groups since they normally involve Amerindian women who live on reservations and men who live near the border of the reservations. In this situation, the children normally remain with their mothers. This phenomenon has been described for Guarani Indians (Marrero et al., 2007), but the data presented here indicate that it could be much more common than previously thought.

Table 4 (cont.)

| Population       | HVS-I | HVS-II | Complete mt genome | References                                                                 |
|------------------|-------|--------|--------------------|---------------------------------------------------------------------------|
| Trinitario       | X     |        |                    | Bert et al. (2004)                                                         |
| Tucuman          |       | X      |                    | Tamm et al. (2007)                                                        |
| Tupe             | X     |        |                    | Lewis Jr et al. (2007)                                                    |
| Txukahinae       | X     | X      |                    | Dornelles et al. (2005)                                                   |
| Uro              | X     |        |                    | Barbieri et al. (2011)                                                    |
| Urubu Kaapor     | X     | X      |                    | Lobato-da-Silva et al. (2001); Dornelles et al. (2005)                    |
| Vaupe            |       | X      |                    | Tamm et al. (2007)                                                        |
| Wai-wai          | X     |        |                    | Fagundes et al. (2008)                                                    |
| Wayampi          | X     | X      |                    | Santos et al. (1996); Lobato-da-Silva et al. (2001); Silva Jr et al. (2002, 2003); Dornelles et al. (2005); Fagundes et al. (2008); Mazieres et al. (2008) |
| Warao            |       |        |                    | Ingman et al. (2000)                                                      |
| Waunana          | X     |        |                    | Tamm et al. (2007)                                                        |
| Wayuu            | X     | X      |                    | Torres et al. (2006); Melton et al. (2007); Tamm et al. (2007); Yang et al. (2010) |
| Wichí            | X     |        |                    | Cabana et al. (2006)                                                      |
| Xavante          | X     | X      |                    | Ward et al. (1996); Dornelles et al. (2005); Fagundes et al. (2008)       |
| Xikrin           | X     | X      |                    | Lobato-da-Silva et al. (2001); Dornelles et al. (2005)                    |
| Yagua            | X     | X      |                    | Monsalve et al. (1994); Torres et al. (2006)                             |
| Yámana           | X     |        |                    | Garcia-Bour et al. (2004)                                                  |
| Yanomami         | X     | X      |                    | Easton et al. (1996); Santos et al. (1996); Merriwether et al. (2000); Lobato-da-Silva et al. (2001); Silva Jr et al. (2002, 2003); Williams et al. (2002); Dornelles et al. (2005); Fagundes et al. (2008) |
| Yungay           | X     |        |                    | Lewis Jr et al. (2007)                                                    |
| Yuracare         | X     |        |                    | Bert et al. (2004)                                                        |
| Zenú             | X     | X      |                    | Torres et al. (2006)                                                      |
| Zóro             | X     |        |                    | Ward et al. (1996); Fagundes et al. (2008)                                |

1Ancient DNA. 2Sequencing included almost half of the genome (sites 7,148-15,976).
Table 5 - Mitochondrial DNA haplogroup and linguistic and geographical information for the samples considered.

| Population (n) | A | B | C | D | Others | Language | Geographical coordinates | References |
|---------------|---|---|---|---|--------|----------|--------------------------|------------|
| Wayuu (89)    | 26| 28| 45|0 | 1      | Arawakan | 11° N; 73° W              | Mesa et al. (2000); Keyeux et al. (2002); Melton et al. (2007) |
| Kogi (153)    | 67| 0 | 33|0 | 0      | Chibchan | 11° N; 74° W              | Keyeux et al. (2002); Melton et al. (2007); Rojas et al. (2010) |
| Arsario (Wiwa) (76) | 63| 0 | 37|0 | 0      | Chibchan | 10° 25' N; 73° 05' W      | Keyeux et al. (2002); Melton et al. (2007); Rojas et al. (2010) |
| Chimila (35)  | 88| 0 | 3 |6 | 3      | Chibchan | 10° 16' N; 74° 4' W        | Keyeux et al. (2002) |
| Arhuaco (Ijka) (134) | 87| 1 | 12|0 | 0      | Chibchan | 9° 04' N; 73° 59' W        | Keyeux et al. (2002); Melton et al. (2007); Rojas et al. (2010) |
| Yukpa (88)    | 0 | 100|0 |0 | 0      | Carib    | 8° 40' N; 72° 41' W        | Keyeux et al. (2002) |
| Zenu (107)    | 19| 36| 5 |2 | 2      | Spanish  | 8° 30' N; 76° W            | Mesa et al. (2000); Keyeux et al. (2002); Torres et al. (2006) |
| Embera (43)   | 53| 35| 2 |5 | 5      | Choco    | 7° N; 76° 30' W            | Mesa et al. (2000); Keyeux et al. (2002) |
| Tule-Cuna (30)| 50| 27| 20|0 | 3      | Chibchan | 6° 56' N; 76° 45' W        | Keyeux et al. (2002) |
| Guane-Butaregua (33) | 12| 64| 0 |24| 0     | Guahiban | 5° N; 69° W                | Keyeux et al. (2002); Vona et al. (2005); Torres et al. (2006) |
| Makiritare (10) | 20| 0 | 70|10| 0     | Carib    | 5° 33' N; 65° 33' W        | Torroni et al. (1993) |
| Kali’ na (Galibi) (29) | 7 | 41| 38|7 | 7     | Carib    | 5° 31' N; 53° 47' W        | Mazières et al. (2008) |

This table continues with similar entries for other populations, detailing their haplogroups percentages, languages, and geographical coordinates. Each entry is referenced with multiple sources, indicating a comprehensive study approach to understanding mitochondrial DNA distribution and linguistic and geographical information.
Table 5 (cont.)

| Population (n) | Haplogroups (%) | Language | Geographical coordinates | References |
|----------------|------------------|----------|--------------------------|------------|
|                | A  | B  | C  | D  | Others |            |                            |                         |
| Pasto (9)      | 67 | 33 | 0  | 0  | 0      | Barbacoan | 0° 58’ N; 77° 44’ W       | Keyeux et al. (2002)    |
| Yagua (12)     | 25 | 0  | 67 | 8  | 0      | Peba-Yaguan | 0° 51’ N; 72° 27’ W    | Torres et al. (2006)    |
| Ingano (111)   | 35 | 38 | 42 | 2  | 0      | Quechuan  | 0° 50’ N; 77° W         | Mesa et al. (2000); Keyeux et al. (2002); Torres et al. (2006); Rojas et al. (2010) |
| Tucano (17)    | 10 | 18 | 47 | 35 | 0      | Tucanoan  | 0° 42’ N; 69° 53’ W     | Keyeux et al. (2002)    |
| Coregaaje (69) | 4  | 20 | 66 | 6  | 4      | Tucanoan  | 0° 38’ N; 76° 8’ W      | Keyeux et al. (2002); Tamm et al. (2007) |
| Awa-Juriti (18)| 0  | 72 | 11 | 0  | 17     | Tucanoan  | 0° 16’ N; 70° 45’ W     |                        |
| Muinane (19)   | 11 | 21 | 37 | 26 | 5      | Witotoan  | 0° 11’ N; 73° 25’ W     | Keyeux et al. (2002)    |
| Poturujara (23)| 44 | 0  | 26 | 30 | 0      | Tucanoan  | 0° 18’ S; 55° 18’ W     | Lobato-da-Silva et al. (2001); Silva Jr et al. (2003) |
| Katuena (23)   | 26 | 9  | 35 | 30 | 0      | Carib     | 0° 40’ S; 57° 30’ W      | Lourenco et al. (2001); Silva Jr et al. (2003) |
| Wai-wai (26)   | 15 | 15 | 43 | 27 | 0      | Carib     | 0° 40’ S; 57° 30’ W      | Bonatto and Salzano (1997) |
| Urubu Kaapor (42)| 21 | 14 | 29 | 5  | 0      | Tucanoan  | 2° -3° S; 46° -47° W     | Torroni et al. (1992, 1993); Lobato-da-Silva et al. (2001); Dornelles et al. (2005) |
| Huitoto (35)   | 23 | 3  | 25 | 46 | 3      | Witotoan  | 2° 14’ S; 72° 19’ W     | Keyeux et al. (2002); Torres et al. (2006) |
| Arara (70)     | 54 | 20 | 26 | 0  | 0      | Carib     | 3° 30’ -4° 20’ S; 53° 0’ -54° 10’ W | Lobato-da-Silva et al. (2001); Ribeiro-dos-Santos et al. (2001); Silva Jr et al. (2003); Bisso-Machado (2010, MSc Dissertation, Universidade Federal do Rio Grande do Sul, Porto Alegre, RS, Brazil.) |
| Awa-Guajá (53)| 13 | 87 | 0  | 0  | 0      | Tucanoan  | 3° 30’ S; 46° 40’ W      | Lobato-da-Silva et al. (2001) |
| Asurini (24)   | 4  | 54 | 17 | 21 | 4      | Tucanoan  | 3° 35’ -4° 12’ S; 49° 40’ -52° 26’ W | Lobato-da-Silva et al. (2001) |
| Piapoco (39)   | 18 | 3  | 15 | 59 | 0      | Arawakan  | 3° 36’ S; 70° 23’ W      | Torres et al. (2006)    |
| Puinave (19)   | 5  | 16 | 58 | 16 | 5      | Arawakan  | 3° 36’ S; 70° 23’ W      | Torres et al. (2006)    |
| Saliba (13)    | 15 | 0  | 55 | 15 | 15     | Tucanoan  | 3° 49’ S; 70° 9’ W       | Torres et al. (2006)    |
| Ticuna (371)   | 20 | 11 | 35 | 33 | 1      | Ticuna    | 4° S; 69° 18’ W          | Scharf et al. (1990); Mesa et al. (2000); Torres et al. (2006); Mendes-Junior and Simões (2009); Rojas et al. (2010) |
| Parakanã (31)  | 6  | 39 | 32 | 23 | 0      | Tucanoan  | 5° 22’ S; 51° 17’ W      | Lobato-da-Silva et al. (2001); Bisso-Machado (2010, MSc Dissertation) |
| Xikrin (33)    | 30 | 64 | 3  | 3  | 0      | Macro-Ge  | 5° 55’ S; 51° W          | Lobato-da-Silva et al. (2001); Dornelles et al. (2005); Bisso-Machado (2010, MSc Dissertation) |
| Suruí (44)     | 7  | 4  | 0  | 89 | 0      | Tucanoan  | 5° 58’ -10° 10’ S; 48° 39’ -61° 10’ W | Bonatto and Salzano (1997); Lobato-da-Silva et al. (2001) |
| Araweté (18)   | 39 | 0  | 50 | 11 | 0      | Tucanoan  | 5° 9’ S; 52° 22’ W       | Lobato-da-Silva et al. (2001) |
| Munduruku (92) | 12 | 17 | 9  | 58 | 4      | Tucanoan  | 6° 23’ S; 59° 9’ W       | Torroni et al. (1992, 1993); Lobato-da-Silva et al. (2001); Marrero et al. (2007); Bisso-Machado (2010, MSc Dissertation) |
| Marubo (10)    | 10 | 0  | 60 | 30 | 0      | Panoan    | 6° 47’ S; 72° 80’ W      | Torroni et al. (1993)    |
| Jamamadi (23)  | 0  | 0  | 96 | 4  | 0      | Arawakan  | 7° 15’ S; 66° 41’ W      | Lobato-da-Silva et al. (2001); Bisso-Machado (2010, MSc Dissertation) |
| Yungay (38)    | 5  | 45 | 34 | 16 | 0      | Quechuan  | 7° 26’ S; 77° 4’ W       | Lewis Jr et al. (2007)   |
| Ancash (33)    | 9  | 52 | 18 | 21 | 0      | Quechuan  | 7° 41’ S; 77° 6’ W       | Lewis Jr et al. (2005)   |
| Gorotire (11)  | 28 | 18 | 36 | 0  | 0      | Macro-Ge  | 7° 44’ S; 51° 10’ W      | Bisso-Machado (2010, MSc Dissertation) |
| Krahó (14)     | 29 | 57 | 14 | 0  | 0      | Macro-Ge  | 8° S; 47° 15’ W          | Torroni et al. (1993)    |
| Kuben-Kran-Kegn (19)| 58 | 26 | 6  | 10 | 0      | Macro-Ge  | 8° 10’ S; 52° 8’ W       | Bisso-Machado (2010, MSc Dissertation) |
| Population | Haplogroups (%) | Language | Geographical coordinates | References |
|------------|----------------|----------|-------------------------|------------|
|            | A | B | C | D | Others |                  |            |
| Mekranoti (19) | 26 | 63 | 11 | 0 | 0 | Macro-Ge | 8° 40' S; 54° W | Dornelles et al. (2005); Bisso-Machado (2010, MSc Dissertation) |
| Kubenkokre (4) | 0 | 100 | 0 | 0 | 0 | Macro-Ge | 8° 43' S; 53° 23' W | Marrero et al. (2007) |
| Kayapó (13) | 46 | 54 | 0 | 0 | 0 | Macro-Ge | 9° S; 53° W | Lobato-da-Silva et al. (2001) |
| Karitiana (19) | 0 | 11 | 0 | 89 | 0 | Tupi | 9° 30' S; 64° 15' W | Lobato-da-Silva et al. (2001) |
| Cinta-Larga (45) | 25 | 0 | 20 | 53 | 2 | Tupi | 9° 50' –12° 30' S; 59° 10' –60° 50' W | Lobato-da-Silva et al. (2001); Dornelles et al. (2005); Bisso-Machado (2010, MSc Dissertation) |
| Gavião (27) | 15 | 15 | 0 | 70 | 0 | Tupi | 10° 10' S; 61° 8' W | Ward et al. (1996) |
| Tupe (16) | 0 | 69 | 31 | 0 | 0 | Aymaran | 10° 16' S; 75° 47' W | Lewis Jr et al. (2007) |
| Txukahamãe (2) | 100 | 0 | 0 | 0 | 0 | Macro-Ge | 10° 20' S; 53° 5' W | Dornelles et al. (2005) |
| Zoró (30) | 20 | 7 | 13 | 60 | 0 | Tupi | 10° 20' S; 60° 20' W | Ward et al. (1996) |
| Kayapó (13) | 46 | 54 | 0 | 0 | 0 | Macro-Ge | 10° 47' -12° 51' S; 73° 17' -70° 44' W | Lobato-da-Silva et al. (2001); Dornelles et al. (2005); Bisso-Machado (2010, MSc Dissertation) |
| Kokraimoro (2) | 50 | 50 | 0 | 0 | 0 | Macro-Ge | 10° 49' S; 55° 27' W | Marrero et al. (2007) |
| Pacaís Novos (Wari) (30) | 40 | 30 | 27 | 3 | 0 | Chapacura-Wanhãm | 11° 8' S; 65° W | Bisso-Machado (2010, MSc Dissertation) |
| Tayacaja (61) | 21 | 33 | 13 | 30 | 3 | Quechuan | 12° 24' S; 74° 34' W | Fuselli et al. (2003) |
| Arequipa (22) | 9 | 68 | 14 | 9 | 0 | Quechua | 13° 13' S; 72° 11' W | Fuselli et al. (2003) |
| Trinitario (35) | 14 | 40 | 37 | 3 | 6 | Arawakan | 14° S; 65° W | Bert et al. (2001) |
| Xavante (25) | 16 | 84 | 0 | 0 | 0 | Macro-Ge | 14° S; 52° 30' W | Ward et al. (1996) |
| Movima (22) | 9 | 9 | 64 | 18 | 0 | Movima | 14° 26' S; 65° 53' W | Bert et al. (2001) |
| Quechua (232) | 14 | 62 | 15 | 9 | 0 | Quechuan | 14° 30' S; 69° W | Merriwether et al. (1995); Bert et al. (2001); Silva Jr et al. (2003); Lewis Jr et al. (2007); Corella et al. (2007); Barbieri et al. (2011); Gayá-Vidal et al. (2011) |
| Chimane (Moseten) (71) | 39 | 54 | 3 | 0 | 4 | Chimane | 14° 41' S; 66° 50' W | Bert et al. (2001); Corella et al. (2007); Mazières et al. (2008) |
| Ignaciano (22) | 18 | 36 | 1 | 0 | 5 | Arawakan | 15° 1' S; 66° 47' W | Bert et al. (2001) |
| Uro (64) | 11 | 69 | 9 | 11 | 0 | Uru-Chipaya | 17° 45' S; 69° 53' W | Barbieri et al. (2011) |
| Yuracare (28) | 39 | 32 | 21 | 4 | 4 | Yuracare | 17° S; 65° W | Bert et al. (2001) |
| Aymara (411) | 4 | 76 | 8 | 11 | 1 | Aymaran | 17° 68' S; 69° 16' W | Merriwether et al. (1995); Easton et al. (1996); Bert et al. (2001); Lewis Jr et al. (2007); Corella et al. (2007); Barbieri et al. (2011); Gayá-Vidal et al. (2011) |
| Ayoreo (91) | 0 | 0 | 83 | 17 | 0 | Zamucoan | 19° S; 60° 30' W | Dornelles et al. (2004) |
| Wichí (199) | 12 | 51 | 7 | 29 | 1 | Mataco-Guaicurú | 22° 28' S; 62° 70' W | Tolroni et al. (1993); Bianchi et al. (1995); Bravi et al. (1995); Demarchi et al. (2001); Cabana et al. (2006) |
| Chorote (34) | 15 | 44 | 23 | 18 | 0 | Mataco-Guaicurú | 22° 90' S; 65° 40' W | Bianchi et al. (1995); Bravi et al. (1995) |
| Humahuaaca (46) | 11 | 68 | 17 | 4 | 0 | Spanish | 23° 11' S; 65° 20' W | Dipierri et al. (1998) |
| Aché (63) | 10 | 90 | 0 | 0 | 0 | Tupi | 23° 30' –24° 10' S; 55° 50' –56° 30' W | Schmitt et al. (2004) |
| Atacamecho (79) | 13 | 73 | 10 | 4 | 0 | Atacama | 23° 50' S; 68° W | Bailleul et al. (1994); Merriwether et al. (1995); Merriwether and Ferrell (1996) |
| Guaraní (249) | 77 | 6 | 9 | 6 | 2 | Tupi | 23° 6' S; 55° 12' W | Silva Jr et al. (2003); Marrero et al. (2007); Garcia and Demarchi (2009) |
| Pilagá (41) | 5 | 37 | 27 | 29 | 2 | Mataco-Guaicurú | 24° S; 59° W | Demarchi et al. (2001); Cabana et al. (2006) |
| Coya (60) | 13 | 57 | 23 | 5 | 2 | Coya | 25° 30' S; 67° 28' W | Álvarez-Iglesias et al. (2007) |
Table 6 summarizes the influence of geography. In the seven regions that were defined, 74% of the variation occurred within populations, 6% among geographic divisions and 20% among populations within divisions. To analyze this variability further, the isolated frequencies of haplogroups A to D were plotted as shown in Figure 2. High frequencies of haplogroup C were observed in specific regions along the northwestern portion of the continent, with additional high spots in southern Brazil and northern Argentina. The prevalences of haplogroups B and D showed a clear east-west separation, while for haplogroup A there were three main high prevalence nuclei in the north, center and south of the continent. Spearman’s correlation coefficient between haplogroup frequencies and latitude yielded a positive value (0.27; p < 0.01) for haplogroup A, with a corresponding negative one (-0.25; p < 0.01) for haplogroup D. The coefficients for haplogroups B and C were not significant.

Table 6 - Mitochondrial DNA haplogroup frequencies by geography.

| Geographic divisions          | No. of populations | No. of individuals | A  | B  | C  | D  | Other |
|------------------------------|--------------------|--------------------|----|----|----|----|-------|
| Amazonia                     | 55                 | 2410               | 20 | 21 | 31 | 25 | 3     |
| Central Plateau              | 2                  | 39                 | 21 | 74 | 5  | 0  | 0     |
| Southern Brazil              | 2                  | 328                | 70 | 6  | 18 | 4  | 2     |
| Chaco                        | 6                  | 479                | 10 | 43 | 22 | 24 | 1     |
| Southern South America       | 3                  | 726                | 4  | 20 | 31 | 43 | 2     |
| Tierra del Fuego             | 5                  | 111                | 0  | 5  | 39 | 55 | 1     |
| Andes                        | 35                 | 2604               | 27 | 45 | 20 | 7  | 1     |
| Total                        | 108                | 6697               |  |    |    |    |       |

1AMOVA results: (a) Among geographic divisions: 6.2%; (b) Among populations within geographic divisions: 19.5%; (c) Within populations: 74.3%. The three values are statistically significant.
Table 7 summarizes the influence of language. Sixteen main language groups were considered, plus a composite set of “others”. The AMOVA results indicated that 73% of the haplogroup prevalence variability occurred within populations, with 7% of it being attributable to languages. However, there was considerable heterogeneity (20%) within the language categories established. Overall, the variability was similar to that obtained for geography.

**Table 7 - Mitochondrial DNA hapgroup frequencies by language**

| Language      | No. of populations | No. of individuals | Haplogroups (%) | Other |
|---------------|--------------------|--------------------|-----------------|-------|
|               |                    |                    | A   | B   | C   | D   | |
| Tupi          | 16                 | 889                | 38  | 24  | 10  | 27  | 1   |
| Macro-Ge      | 11                 | 221                | 37  | 38  | 21  | 3   | 1   |
| Carib         | 9                  | 408                | 24  | 32  | 25  | 18  | 1   |
| Chibchan      | 6                  | 461                | 69  | 6   | 22  | 2   | 1   |
| Mataco-Guaicuru | 5             | 359                | 13  | 46  | 11  | 29  | 1   |
| Arawakan      | 8                  | 321                | 16  | 38  | 24  | 13  | 9   |
| Araucanian    | 3                  | 726                | 4   | 20  | 31  | 43  | 2   |
| Choco         | 2                  | 204                | 28  | 46  | 13  | 12  | 1   |
| Chon          | 3                  | 60                 | 0   | 10  | 33  | 55  | 2   |
| Tucanoan      | 6                  | 140                | 14  | 26  | 48  | 8   | 4   |

Figure 2 - Isoline map distribution showing the geographic pattern of the four (A-D) mtDNA haplogroups in South Amerindians. The dots indicate the locations of the populations sampled. As indicated in the scales given at right of each map, the colors represent the haplogroup frequencies, from dark blue (0.00) to red (1.00).
Conclusion

South Amerindians have been extensively studied with regard to the Y-chromosome, as well as and especially so for mtDNA markers. In agreement with studies from other regions, by far most of the mtDNA variability (73%-74%) is intrapopulational. Geographical and linguistic factors influenced the patterns of mtDNA diversity to a similar extent, while geography was apparently more important than language in explaining the data for the Y chromosome Q clade-STRs. Additional factors that may have influenced these results include distinct male and female migration patterns, as well as cultural and other characteristics. The fact that most studies have generally dealt with small populations, in which genetic drift may be important, could also have influenced the results.

Acknowledgments

This work was supported by Coordenação de Aperfeiçoamento de Pessoal de Nível Superior (CAPES), Conselho Nacional de Desenvolvimento Científico e Tecnológico (CNPq) and Fundação de Amparo a Pesquisa do Rio Grande do Sul, Programa de Apoio a Núcleos de Excelência (FaperGS/PRONEX). We thank Sidia M. Callegari-Jacques and Luciana Tovo-Rodrigues for their help with early aspects of the statistical analyses.

References

Altuna ME, Modesti NM and Demarchi DA (2006) Y-chromosomal evidence for a founder effect in Mbyá-guaraní Amerindians from northeast Argentina. Hum Biol 78:635-639.

Alvarez-Iglesias V, Jaime JC, Carracedo A and Salas A (2007) Coding region mitochondrial DNA SNPs: Targeting East Asian and Native American haplogroups. Forensic Sci Int Genet 1:44-55.

Alves-Silva J, da Silva Santos M, Guimarães PE, Ferreira AC, Bandelt HJ, Penna SD and Prado VF (2000) The ancestry of Brazilian mtDNA lineages. Am J Hum Genet 67:444-461.

Baillet G, Rothhammer F, Carnese FR, Bravi CM and Bianchi NO (1994) Founder mitochondrial haplotypes in Amerindian populations. Am J Hum Genet 54:27-33.

Baillet G, Ramallo V, Muzzio M, Garcia A, Santos MR, Alfiaro EL, Dipierr E, Salc S, Carnese FR, Bravi CM, et al. (2009) Brief communication: Restricted geographic distribution for Y-Q* paragroup in South America. Am J Phys Anthropol 140:578-582.

Barbieri C, Heggarty P, Casti L, Luissell D and Pettener D (2011) Mitochondrial DNA variability in the Iticaca basin: Matches and mismatches with linguistics and ethnohistory. Am J Hum Biol 23:89-99.

Bert F, Corella A, Gené M, Pérez-Pérez A and Turbón D (2001) Major mitochondrial DNA haplotype heterogeneity in highland and lowland Amerindian populations from Bolivia. Hum Biol 73:1-16.

Bert F, Corella A, Gene M, Pérez-Pérez A and Turbón D (2004) Mitochondrial DNA diversity in the Llanos de Moxos: Moxo, Movima and Yuracare Amerindian populations. Ann Hum Biol 31:9-28.

Bianchi NO, Baillet G and Bravi CM (1995) Peopling of the Americas as inferred through the analysis of mitochondrial DNA. Bact J Genet 18:161-168.

Bianchi NO, Canesiti CI, Baillet G, Martinez-Marignac VL, Bravi CM, Vidal-Rioja LB, Herrera RJ and López-Camelo JS (1998) Characterization of ancestral and derived Y-chromosome haplotypes of New World native populations. Am J Hum Genet 63:1862-1871.

Bissu-Machado R, Jota MS, Ramallo V, Paixão-Côrtes VR, Lacerda DR, Salzano FM, Bonatto SL, Santos FR and Bertolini MC (2011) Distribution of Y-chromosome Q lineages in Native Americans. Am J Hum Biol 23:563-566.

Blanco-Vere AC, Jaime JC, Brion M and Carracedo A (2010) Y-chromosome lineages in native South American populations. Forensic Sci Int Genet 4:187-193.

Bonatto SL and Salzano FM (1997) A single and early migration for the peopling of the Americas supported by mitochondrial DNA sequence data. Proc Natl Acad Sci USA 94:1866-1871.

Bortolini MC, Da Silva Junior WA, De Guerra DC, Remonatto G, Miranda R, Hutz MH, Weimer TA, Silva MC, Zago MA and Salzano FM (1999) African-derived South American populations: A history of symmetrical and asymmetrical
matings according to sex revealed by bi- and uni-parental genetic markers. Am J Hum Biol 11:551-563.

Bortolini MC, Salzano FM, Bau CHD, Layrisse Z, Petzl-Erler ML, Tsuneto LT, Hill K, Hurtado AM, Castro-de-Guerra D, Bedoya G, et al. (2002) Y-chromosome biallelic polymorphisms and Native American population structure. Ann Hum Genet 66:255-259.

Bortolini MC, Salzano FM, Thomas MG, Stuart S, Nasanen SPK, Bau CHD, Hutz MH, Layrisse Z, Petzl-Erler ML, Tsuneto LT, et al. (2003) Y-Chromosome evidence for differing ancient demographic histories in the Americas. Am J Hum Genet 73:524-539.

Bravi CM, Cejas S, Baillet G, Goicoechea AS, Carnese FR and Bianchi NO (1995) Haplotipos mitocondriales en Amerindios. Abstractos do XXVI Congreso Argentino de Genética, San Carlos de Bariloche, pp 152.

Cabana GS, Merriwether DA, Hunley K and Demarchi DA (2006) Is the genetic structure of Gran Chaco populations unique? Interregional perspectives on Native South American mitochondrial DNA variation. Am J Phys Anthropol 131:108-119.

Carvalho-Silva DR, Santos FR, Hutz MH, Salzano FM and Pena SDJ (1999) Divergent human Y-chromosome microsatellite evolution rates. J Mol Evol 49:204-214.

Carvalho-Silva DR, Santos FR, Rocha J and Pena SD (2001) The phylogeography of Brazilian Y-chromosome lineages. Am J Hum Genet 68:281-286.

Corella A, Bert F, Pérez-Pérez A, Gené M and Turbón D (2007) Mitochondrial DNA diversity of the Amerindian populations living in the Andean Piedmont of Bolivia: Chimane, Mosetén, Aymara and Quechua. Ann Hum Biol 34:34-55.

Crawford MH (1998) The Origins of Native Americans. Evidence from Anthropological Genetics. Cambridge University Press, Cambridge, 308 pp.

Demarchi DA and Mitchell RJ (2004) Genetic structure and gene flow in Gran Chaco populations of Argentina: Evidence from Y-Chromosome markers. Hum Biol 76:413-429.

Demarchi DA, Panzetta-Dutari GM, Colontonio SE and Marcelino AJ (2001) Absence of the 9-bp deletion of mitochondrial DNA in pre-Hispanic inhabitants of Argentina. Hum Biol 73:575-582.

Dipierri JE, Alfaro E, Martínez-Marignac VL, Baillet G, Bravi CM, Cejas S and Bianchi NO (1998) Paternal directional mating in two Amerindian subpopulations located at different altitudes in northwestern Argentina. Hum Biol 70:1001-1010.

Dornelles CL, Battilana J, Fagundes NJ, Freitas LB, Bonatto SL and Salzano FM (2004) Mitochondrial DNA and Alu insertions in a genetically peculiar population: The Ayoreo. Am J Hum Biol 16:479-488.

Dornelles CL, Bonatto SL, Freitas LB and Salzano FM (2005) Is haplogroup X present in extant South American Indians? Am J Phys Anthrop 127:439-448.

Eastman JR (2006) IDRISI 15.0: The Andes edition. Clark University, Worcester.

Easton RD, Merriweather DA, Crews DE and Ferrell RE (1996) mtDNA variation in the Yanomami: Evidence for additional New World founding lineages. Am J Hum Genet 59:213-225.

Excoffier L and Lischer HE (2010) Arlequin suite ver 3.5: A new series of programs to perform population genetics analyses under Linux and Windows. Mol Ecol Resour 10:564-567.

Excoffier L, Smouse PE and Quattro JM (1992) Analysis of molecular variance inferred from metric distances among DNA haplotypes: Application to human mitochondrial DNA restriction data. Genetics 131:479-491.

Fagundes NJR, Kanitz R, Eckert R, Valls ACS, Bogo MR, Salzano FM, Smith DG, Silva Jr WA, Zago MA, Ribeiro-dos-Santos AK, et al. (2008) Mitochondrial population genomics supports a single pre-Clovis origin with a coastal route for the peopling of the Americas. Am J Hum Genet 82:583-592.

Fehren-Schmitz L, Reindel M, Cagigao ET, Hummel S and Herrmann B (2010) Pre-Columbian population dynamics in coastal southern Peru: A diachronic investigation of mtDNA patterns in the Palpa region by ancient DNA analysis. Am J Phys Anthropol 141:208-221.

Fehren-Schmitz L, Warnberg O, Reindel M, Seidenberg V, Tomasto-Cagigao E, Isla-Cuadrado J, Hummel S and Herrmann B (2011) Diachronic investigations of mitochondrial and Y-chromosomal genetic markers in pre-Columbian Andean highlanders from South Peru. Ann Hum Genet 75:266-283.

Fuselli S, Tarazona-Santos S, Dupanloup I, Soto A, Luiselli D and Pettener D (2003) Mitochondrial DNA diversity in South America and the genetic history of Andean Highlanders. Mol Biol Evol 20:1682-1691.

García A and Demarchi DA (2009) Incidence and distribution of Native American mtDNA haplogroups in central Argentina. Hum Biol 81:59-69.

García F, Moraga M, Vera S, Henríquez H, Llop E, Aspillaga E and Rothhammer F (2006) mtDNA microevolution in Southern Chile’s archipelagos. Am J Phys Anthropol 129:473-481.

García-Bour J, Pérez-Pérez A, Álvarez S, Fernández E, López-Parrama AM, Arroyo-Pardo E and Turbón D (2004) Early population differentiation in extinct aborigines from Tierra Del Fuego – Patagonia: Ancient mtDNA sequences and Y-chromosome STR characterization. Am J Phys Anthropol 123:361-370.

Gayá-Vidal M, Moral P, Saenz-Ruales N, Gerbault P, Tonasso L, Villena M, Vasquez R, Bravi CM and Dugoujon JM (2011) mtDNA and Y-chromosomal diversity in Aymaras and Quechuas from Bolivia: Different stories and special genetic traits of the Andean Altiplano populations. Am J Phys Anthropol 145:215-230.

Geppert M, Baeta M, Núñez C, Martínez-Jarreta B, Zweynert S, Cruz OW, González-Andrade F, González-Solorzano J, Nagy M and Roewer L (2011) Hierarchical Y-SNP assay to study the hidden diversity and phylogenetic relationship of native populations in South America. Forensic Sci Int Genet 5:100-104.

Ginther C, Corach D, Penacino GA, Rey JA, Carnese FR, Hutz MH, Anderson A, Just J, Salzano FM and King M-C (1993) Genetic variation among the Mapuche Indians from the Patagonian region of Argentina: Mitochondrial DNA sequence variation and allele frequencies of several nuclear genes. In: Pena SDJ, Chakraborty R, Epplen JT and Jeffreys AJ (eds) DNA Fingerprinting: State of the Science. Birkhäuser Verlag, Berlin, pp 211-219.
Goicoechea AS, Carnese FR, Dejean C, Avena SA, Weimer TA, Estalote AC, Simões ML, Palatnik M, Salamoni SP, Salzano FM, et al. (2001) New genetic data on Amerindians from the Paraguayan Chaco. Am J Hum Biol 13:660-667.

Horai S, Kondo S, Nakagawa-Hattori Y, Hayashi S, Sonoda S and Tajima K (1993) Peopling of the Americas, founded by four major lineages of mitochondrial DNA. Mol Biol Evol 10:23-47.

Hunley KL, Cabana GS, Merriwether DA and Long JC (2007) A formal test of linguistic and genetic coevolution in native Central and South America. Am J Phys Anthropol 132:622-631.

Ingman M, Kaessmann H, Pääbo S and Gyllensten U (2000) Mitochondrial genome variation and the origin of modern humans. Nature 408:708-713.

Jota MS, Lacerda DR, Sandoval JR, Vieira PPR, Santos-Lopes SS, Bisso-Machado R, Paixão-Cortes VR, Revollo S, Paz-y-Miño C, Fujita R, et al. (2011) A new subclade of Native American Y chromosomes from the Andes. Am J Phys Anthropol 146:553-559.

Karafet T, Segura SL, Vuturo-Brady J, Posukh O, Ospivova L, Wiebe V, Romero F, Long JC, Harihara S, Jin F, et al. (1997) Y chromosome markers and trans-Bering Strait dispersals. Am J Phys Anthropol 102:301-314.

Karafet TM, Zegura SL, Posukh O, Ospivova L, Bergen A, Long J, Goldman D, Klitz W, Harihara S, de Knijff P, et al. (1999) Ancestral Asian source(s) of New World Y-chromosome founder haplotypes. Am J Hum Genet 64:817-831.

Karafet TM, Mendez FL, Meilerman MB, Underhill PA, Zegura SL and Hammer MF (2008) New binary polymorphisms reshape and increase resolution of the human Y chromosomal haplogroup tree. Genome Res 18:830-838.

Keyeux G, Rodas C, Gelvez N and Carter D (2002) Possible migration routes into South America deduced from mitochondrial DNA studies in Colombian Amerindian populations. Hum Biol 74:211-233.

Kivisild T, Shen P, Wall D, Do B, Sung R, Davis K, Passarino G, Underhill PA, Scharfe C, Torroni A, et al. (2006) The role of selection in the evolution of human mitochondrial genomes. Genetics 172:373-387.

Leite FP, Callegari-Jacques SM, Carvalho BA, Kommers T, Matte CH, Raimann PE, Schwenger SP, Sortica VA, Tsuneto LT, Petzl-Erler ML, et al. (2008) Y-STR analysis in Brazilian and South Amerindian populations. Am J Hum Biol 20:359-363.

Lell JT, Sukernik RI, Starikovskaya YB, Su B, Jin L, Schurr TG, Underhill PA and Wallace DC (2002) The dual origin and Siberian affinities of Native American Y chromosomes. Am J Hum Genet 70:192-206.

Lewis Jr CM, Tito RY, Lizárraga B and Stone AC (2005) Land, language, and loci: mtDNA in Native Americans and the genetic history of Peru. Am J Phys Anthropol 127:351-360.

Lewis Jr CM, Buikstra JE and Stone AC (2007) Ancient DNA and genetic continuity in the South Central Andes. Lat Am Antiq 18:1-48.

Lobato-da-Silva DF, Ribeiro-dos-Santos AKC and Santos SEB (2001) Diversidade genética de populações humanas na Amazônia. In: Guimarães Vieira IC, Cardoso da Silva JM, Oren DC and D’Ineoa MA (eds) Diversidade Humana e Cultural na Amazônia. Museu Paraense Emílio Goeldi, Belém, pp 167-193.
Schurr TG and Sherry ST (2004) Mitochondrial DNA and Y chromosome diversity and the peopling of the Americas: An integrated analysis of pre-Hispanic mortuary practices. Curr Anthropol 45:369-402.

Shinoda K, Adachi N, Guillon S and Shimada I (2006) Mitochondrial DNA analysis of ancient Peruvian highlanders. Am J Phys Anthropol 131:98-107.

Silva Jr WA, Bonatto SL, Holanda AJ, Ribeiro-dos Santos AK, Paixão BM, Goldman GH, Abe-Sandes K, Rodriguez-Del-fín L, Barbosa M, Pacó-Larson ML, et al. (2002) Mitochondrial genome diversity of Native Americans supports a single early entry of founder populations into America. Am J Hum Genet 71:187-192.

Silva Jr WA, Bonatto SL, Holanda AJ, Ribeiro-dos-Santos AK, Paixão BM, Goldman GH, Abe-Sandes K, Rodriguez-Del-fín L, Barbosa M, Pacó-Larson ML, et al. (2003) Correction: Mitochondrial DNA variation in Amerindians. Am J Hum Genet 72:1346-1348.

Tamm E, Kivisild T, Reilda M, Metspalu M, Smith DG, Mulligan CJ, Bravi CM, Rickards O, Martinez-Labarga C, Khusnutdinova EK, et al. (2007) Beringian standstill and spread of Native American founders. PLoS One 2:e829.

The Y Chromosome Consortium (2002) A nomenclature system for the tree of human Y-chromosomal binary haplogroups. Genome Res 12:339-348.

Torres MM, Bravi CM, Bortolini MC, Duque C, Callegari-Jacques S, Ortiz D, Bedoya G, Groot de Restrepo H and Ruiz-Linares A (2006) A revertant of the major founder Native American haplogroup C common in populations from northern South America. Am J Hum Biol 18:59-65.

Torroni A, Schurr TG, Yang C-C, Szathmary EJE, Williams RC, Schanfield MS, Troup GA, Knowler WC, Lawrence DN, Weiss KM, et al. (1992) Native American mitochondrial DNA analysis indicates that the Amerind and the Nadene populations were founded by two independent migrations. Genetics 130:153-162.

Torroni A, Schurr TG, Cabell MF, Brown MD, Neel JV, Larsen M, Smith DG, Vullo CM and Wallace DC (1993) Asian affinities and continental radiation of the four founding Native American mtDNAs. Am J Hum Genet 53:563-590.

Toscanini U, Gusmão L, Berardi G, Amarim A, Carracedo A, Salas A and Raimondi E (2008) Y chromosome microsatellite genetic variation in two Native American populations from Argentina: Population stratification and mutation data. Forensic Sci Int Genet 2:274-280.

Toscanini U, Gusmão L, Berardi G, Gomes V, Amarim A, Salas A and Raimondi E (2011) Male lineages in South American native groups: Evidence of M19 traveling south. Am J Phys Anthropol 146:188-196.

Underhill PA, Jin L, Zemans R, Oefner PJ and Cavalli-Sforza LL (1996) A pre-Columbian Y chromosome-specific transition and its implications for human evolutionary history. Proc Natl Acad Sci USA 93:196-200.

Underhill PA, Jin L, Lin AA, Mehdi SQ, Jenkins T, Vollrath D, Davis RW, Cavalli-Sforza LL and Oefner PJ (1997) Detection of numerous Y chromosome biallelic polymorphisms by denaturing high-performance liquid chromatography. Genome Res 7:996-1005.
Underhill PA, Passarino G, Lin AA, Shen P, Mirazón Lahr M, Foley RA, Oefner PJ and Cavalli-Sforza LL (2001) The phylogeography of Y chromosome binary haplotypes and the origins of modern human populations. Ann Hum Genet 65:43-62.

Vallinoto AC, Cayres-Vallinoto IM, Ribeiro Dos Santos ÂKC, Zago MA, Santos SE and Guerreiro JF (1999) Heterogeneity of Y chromosome markers among Brazilian Amerindians. Am J Hum Biol 11:481-487.

Vona G, Falchi A, Moral P, Calò CM and Varesi L (2005) Mitochondrial sequence variation in the Guahibo Amerindian population from Venezuela. Am J Phys Anthropol 127:361-369.

Ward RH, Salzano FM, Bonatto SL, Hutz MH, Coimbra CEA and Santos RV (1996) Mitochondrial DNA polymorphism in three Brazilian Indian tribes. Am J Hum Biol 8:317-323.

Weir BS (1996) The second National Research Council report on forensic DNA evidence. Am J Hum Genet 59:497-500.

Weir BS and Cockerham CC (1984) Estimating F-statistics for the analysis of population structure. Evolution 38:1358-1370.

Williams SR, Chagnon NA and Spielman RS (2002) Nuclear and mitochondrial genetic variation in the Yanomamo: A test case for ancient DNA studies of prehistoric populations. Am J Phys Anthropol 117:246-259.

Yang NN, Mazières S, Bravi C, Ray N, Wang S, Burley MW, Bedoya G, Rojas W, Parra MV, Molina JA, et al. (2010) Contrasting patterns of nuclear and mtDNA diversity in Native American populations. Ann Hum Genet 74:525-538.

Zegura SL, Karafet TM, Zhivotovsky LA and Hammer MF (2004) High-resolution SNPs and microsatellite haplotypes point to a single, recent entry of Native American Y chromosomes into the Americas. Mol Biol Evol 21:164-175.

Internet Resources

PASW Statistics 18, http://www.spss.com (June 10, 2011).
Pubmed, http://www.ncbi.nlm.nih.gov/pubmed/ (June 10, 2011).
Y Chromosome Haplotype Reference Database, http://www.yhrd.org/ (June 10, 2011).
Lewis MP (2009) Ethnologue: Languages of the World. 16th edition. SIL International, Dallas, http://www.ethnologue.com/.

Associate Editor: Mara H. Hutz

License information: This is an open-access article distributed under the terms of the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited.