Genetic signatures of parental contribution in black and white populations in Brazil

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Abstract

Two hundred and three individuals classified as white were tested for 11 single nucleotide polymorphisms plus two insertion/deletions in their Y-chromosomes. A subset of these individuals (n = 172) was also screened for sequences in the first hypervariable segment of their mitochondrial DNA (mtDNA). In addition, complementary studies were done for 11 of the 13 markers indicated above in 54 of 107 black subjects previously investigated in this southern Brazilian population. The prevalence of Y-chromosome haplogroups among whites was similar to that found in the Azores (Portugal) or Spain, but not to that of other European countries. About half of the European or African mtDNA haplogroups of these individuals were related to their places of origin, but not their Amerindian counterparts. Persons classified in these two categories of skin color and related morphological traits showed distinct genomic ancestries through the country. These findings emphasize the need to consider in Brazil, despite some general trends, a notable heterogeneity in the pattern of admixture dynamics within and between populations/groups.

Key words: admixture dynamics, mtDNA, Y-chromosome markers.

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Introduction

The great maritime expeditions and colonial expansion by Europeans during the 15th and 16th centuries considerably changed their history and those of the colonized peoples. The cultural impact of colonization has been known for a long time, but only more recently have its genetic aspects been investigated. Genetic studies of Brazilian populations have shown, for example, that the extent of admixture among Europeans, Africans and Native Americans is higher than perceived by phenotypic characteristics, and that marriages during the Colonial Era were mainly asymmetric, with most of the Y-chromosomes present in contemporary white Brazilian populations being of European origin, but a significant portion of the mtDNA lineages being of Amerindian or African origin (Alves-Silva et al., 2000; Carvalho-Silva et al., 2001). However, marked regional differences have been reported for white Brazilian populations, indicating that there can be considerable variation in the general trends (Callegari-Jacques et al., 2003; Marrero et al., 2005, 2007a; Zembrzuski et al., 2006). DNA studies with urban black populations have been less common but have also identified important regional differences (Bortolini et al., 1999; Hümeier et al., 2007; Gonçalves et al., 2008).

In this work, we investigated the genetic background of people classified as white or black living in the same city (Porto Alegre, the capital of the southernmost state of Brazil). Our results reveal details about the history of Porto Alegres human population and indicate the extent of genetic admixture, as well as the nature of this process within and between these two major color groups in Brazil's four distinct geographical regions. These findings also show how it is possible to discover details of the ancestral history of a contemporary population based on its current genetic characteristics.

Subjects and Methods

Population samples and DNA extraction

The history of Porto Alegre (30°5’ S, 5°10’ W), the capital of the Brazilian state of Rio Grande do Sul, dates from 1730 when Jerônimo de Ornellas, an immigrant from Madeira Island, received a large plot of land (sesmaria) in this area from the Portuguese crown. The little village that was subsequently formed showed significant population growth only after the arrival of ~500 couples from the Azores archipelago (another Portuguese colony), from 1752 to 1754 (Flores, 1996). The importance of the Azorian
migrations to Rio Grande do Sul, especially Porto Alegre, is illustrated by Laytano (1974), who stated that in 1780 about 55% of the 18,000 inhabitants of the town were Azorian or of Azorian-descendant. The pre-Columbian inhabitants, the Guarani Indians, were displaced, although in other regions of the state they, as well as other natives, were still present (Noelli et al., 1997). Subsequent waves of European migration, especially from Portugal, Spain, Italy and Germany, contributed to the growth of Porto Alegre and nearby cities. The presence of African slaves is also mentioned in historical records (Nogueról, 2005), although their proportion relative to the total population is unknown. Currently, the urban complex formed by the capital and neighboring cities has 3,152,596 inhabitants, 7% and 88% of whom are classified as blacks (pretos, in Portuguese) and whites (brancos), respectively (Brazilian Institute of Geography and Statistics-IBGE, 2000). In Brazil, skin color rather than ancestry is used to define an equivalent to “race”, and in the present study the word “black” will be used to refer to “race”, and in the present study the word “black” will be used to refer to pretos and any person identified and/or self-identified with some other term that suggests African ancestry, such as mulato or pardos. “White” will be used to define those who, based on their physical traits and information, show no admixture with non-Europeans.

Two hundred and three unrelated individuals living in Porto Alegre and its metropolitan region, phenotypically classified as white by the interviewer of the research group, were studied. DNA was extracted from whole blood according to Lahiri and Nurnberger (1991) or from saliva using QIAamp® kits (QIAGEN), according to the manufacturer's instructions. This investigation was approved by the Brazilian National Ethics Commission (CONEP Resolution no. 1333/2002).

Y-chromosome markers

Hünemeier et al. (2007) recently presented and discussed the African portion of the Y-chromosomes and mtDNA lineages of a sample of 107 blacks from the same city. Here, we investigated additional Y-chromosome polymorphisms (see below) to characterize the non-African portion of the Y-chromosomes of this sample, and also considered the mtDNA lineages classified as Amerindian or European. This procedure standardized the set of uniparental markers investigated in the two samples, thereby allowing comparative analyses.

Two hundred and three white men were genotyped for 13 binary loci (11 single nucleotide polymorphisms or SNPs: 92R7, M3, M242, M9, M17, M170, M173, M213, M269, SRY2627, M2, and two insertion/deletions, Yap ALU and 12f2) located in the non-recombinant region of the Y-chromosome (NRY). These markers define well the most common and widespread African, European and Native American Y-chromosomes. Genotyping was done according to the hierarchical order of markers presented by Jobling and Tyler-Smith (2003) using the primers and conditions described in Hammer and Horai (1995), Underhill et al. (1996, 2001), Hurles et al. (1999), Thomas et al. (1999), Rosser et al. (2000), Bortolini et al. (2003), Flores et al. (2003), Montiel et al. (2005), and Kayser et al. (2005). Haplogroups P*(xQ3,R1), Q3*, K(xP), Q*(xQ3), R1a1*(xDE), I*(xP,K,J), R1*(xR1a1,R1b3), F(xI,J,K,P), R1b3*(xR1b3f), R1b3f, E3a*, DE*(E3a) and J* were established using the markers indicated above in the order given. According to Jobling and Tyler-Smith (2003), designations such as P*(xQ3, R1) indicate partial typing of markers in a haplogroup and, in the present case, describes all chromosomes in clade P* except Q3* and R1*. We also investigated 11 of the biallelic loci indicated above (92R7, M3, M242, M9, M17, M170, M173, M213, M269, SRY2627 and 12f2) to characterize the non-African portion of the Y-chromosomes in the black sample previously studied by Hünemeier et al. (2007).

Mitochondrial DNA

The first hypervariable segment (HVS-I) of the mtDNA control region was sequenced in a subset of 172 white individuals using primers and conditions previously described by Marrero et al. (2005). The sequence reactions were run in an automated ABI 310 sequencer. Both DNA strands were sequenced from positions 16050 to 16391, since this is the region for which most of the comparative information is available. Individuals with the “C-stretch” between positions 16183-16193, which is caused by the 16189C substitution, were re-sequenced in each direction so that each base was determined twice.

The sequences were checked manually, and validated by using the CHROMAS LITE 2.0 program. Alignment relative to the revised Cambridge Reference Sequence (Andrews et al., 1999) was done using the BIOEDIT software (Hall, 1999). The filtering procedure described by Bandelt et al. (2002) was used to check the quality of the sequences and eliminate artifacts introduced during sequencing and editing. After filtering, the relationships between the lineages were examined with the NETWORK 4.2.0.0. program using the median-joining algorithm (Bandelt et al., 1999). Weight networks showing star tree patterns, together with other criteria such as those suggested by Yao et al. (2004), ensured that the data were essentially free of artifacts. The HVS-I sequences were classified into haplogroups according to recommended criteria (Bandelt et al., 2002; Salas et al., 2002, 2004; Kivisild et al., 2002; Torroni et al., 2006).

Data analysis

Y-SNP and mtDNA haplogroup frequencies were obtained by counting. Estimates of parental continental contributions in the paternal and maternal data sets were obtained directly since the major haplogroups of mtDNA and Y-SNP are geographically specific. Population relationships were estimated through F_st genetic distances us-
ing Arlequin version 3.01 (Schneider et al., 2000; Excoffier et al., 2005) and their statistical significance was assessed by permutation using 10,000 runs.

Results and Discussion

Y-chromosome biallelic polymorphisms (SNPs)

Based on the Y-chromosome distribution (Table 1), the most frequent haplogroup in Porto Alegre whites was R1b3* (51%), which was also the most common haplogroup found in another Brazilian city (Rio de Janeiro), as well as in the Azores and Portugal. The frequency of this haplogroup was lower (~13%) in Porto Alegre blacks.

To test the hypothesis of random haplogroup distribution among populations we computed FST values based on the major hierarchical clustering for these analyses. There were no significant differences between Porto Alegre whites and those of the Azores, Portugal or Spain (FST = 0.00162, p = 0.6126; FST = 0.00235, p = 0.1622; FST = 0.00345, p = 0.1261, respectively), although the Porto Alegre white prevalences differ significantly from those of the other European countries considered. The difference between Porto Alegre and Rio de Janeiro was also significant (FST = 0.0120, p = 0.0090), but when the African (E3a, B*, and DE) and Amerindian (Q* and Q3*) haplogroups are excluded this significance disappeared (FST = 0.0056, p = 0.1441). Surprisingly, the haplogroups of European origin detected in Porto Alegre blacks were significantly different from those found in whites of the same city (FST = 0.1562, p = 0.000), and from those observed in Iberian populations. However, since the number of blacks studied was small (30), it is probable that these differences reflect sampling error rather than any phenomenon of the admixture process. The African portion of the Y-chromosomes of this black sample has been described in detail by Hünemeier et al. (2007).

Mitochondrial DNA

Multiple alignment with the reference sequence (Andrews et al., 1999) allowed the identification of 105 mtDNA lineages in white individuals (Table 2). This table also shows the non-African lineages identified in blacks, as well as those shared by the two samples.

For 35 of the 69 European lineages (51%), identical matches were found for data in the literature from countries with an important history of migration to Rio Grande do Sul (Portugal/Azores, Spain, Italy, and Germany; data from Crespiollo et al., 2000; Pereira et al., 2000; Mogental-Profizi et al., 2001; Brehm et al., 2003; Poetsch et al., 2003; Picornell et al., 2005; Pichler et al., 2006). Most of these haplotypes (25; 71%) perfectly matched more than one of the European populations. Four (lineages #12, #13, #38 and #68), three (#36, #62 and #63), two (#2 and #38), and one (#14) of the haplotypes found in Porto Alegre (Table 2) exclusively matched those found in the Azores, Portugal.
Table 2 - mtDNA haplotypes and haplogroups in Porto Alegre residents classified as black or white.

| Lineage | Number of sequences | HVS-I mutations$^a$ | Haplogroup$^b$ | Origin   |
|---------|---------------------|---------------------|----------------|----------|
|         | White               | Black               |                |          |
| #       |                     |                     |                |          |
| 1       | 22                  | 2                   | rCRS           | H        | European |
| 2       | 4                   | 126                 | H              | European |
| 3       | 3                   | 153                 | H              | European |
| 4       | 2                   | 261                 | H              | European |
| 5       | 2                   | 354                 | H              | European |
| 6       | 2                   | 311                 | H              | European |
| 7       | 1                   | 83                  | H              | European |
| 8       | 1                   | 93                  | H              | European |
| 9       | 1                   | 114                 | H              | European |
| 10      | 1                   | 168                 | H              | European |
| 11      | 1                   | 189                 | H              | European |
| 12      | 1                   | 240                 | H              | European |
| 13      | 1                   | 124 354             | H              | European |
| 14      | 1                   | 93 304              | H              | European |
| 15      | 1                   | 209                 | H              | European |
| 16      | 1                   | 233                 | H              | European |
| 17      | 1                   | 189 300 325         | H              | European |
| 18      | 1                   | 361                 | H              | European |
| 19      | 2                   | 162                 | H1a            | European |
| 20      | 1                   | 304                 | H5             | European |
| 21      | 1                   | 129 223             | I              | European |
| 22      | 2                   | 69 126 192          | J              | European |
| 23      | 1                   | 69 126 193 300 309  | J              | European |
| 24      | 1                   | 69 126 366          | J              | European |
| 25      | 1                   | 69 126 222          | J              | European |
| 26      | 2                   | 69 126 145 231 261  | J2a            | European |
| 27      | 1                   | 69 93 126 261 274 355 | J2a         | European |
| 28      | 1                   | 69 126 261          | J2a            | European |
| 29      | 1                   | 69 126 145 172 222 261 | J1b1         | European |
| 30      | 1                   | 69 126 145 172 222 261 305T | J1b1       | European |
| 31      | 3                   | 69 126 193 278      | J1c            | European |
| 32      | 1                   | 69 111 126          | J1c            | European |
| 33      | 1                   | 188 224 311         | K              | European |
| 34      | 3 1                 | 224 311             | K              | European |
| 35      | 1                   | 83 224 311          | K              | European |
| 36      | 1                   | 86 224 311          | K              | European |
| 37      | 1 1                 | 93 224 290 311      | K              | European |
| 38      | 1                   | 93 224 311          | K              | European |
| 39      | 1                   | 129 223 291 298     | M or I         | European |
| 40      | 1                   | 129 183C 189 249 311 | M1 or U1a     | European |
| 41      | 1                   | 126 145 176G 223 260 | N1b          | European |
| 42      | 1                   | 145 176G 223        | N1b            | European |
| 43      | 1                   | 126 292 294         | T              | European |
| 44      | 1                   | 51 126 294          | T              | European |
| 45      | 3                   | 126 163 186 189 284 294 | T1          | European |
| Lineage # | White Number of sequences | HVS-I mutations<sup>a</sup> | Haplogroup<sup>b</sup> | Origin |
|-----------|--------------------------|-----------------------------|---------------------|--------|
| 46        | 1                        | 126 254 294 296 304         | T2b                 | European |
| 47        | 3                        | 126 294 296 304             | T2b                 | European |
| 48        | 1                        | 126 294 296 304 360         | T2b                 | European |
| 49        | 1                        | 126 193 294 296 304 357     | T2b                 | European |
| 50        | 1                        | 126 294 296 311             | T2b                 | European |
| 51        | 1                        | 126 193 294 296 304         | T2b                 | European |
| 52        | 2                        | 126 220 292 294             | T3                  | European |
| 53        | 1                        | 126 163 186 189 294         | T3                  | European |
| 54        | 1                        | 51 129C 183C 189            | U2e                 | European |
| 55        | 1                        | 189 319 356                 | U4                  | European |
| 56        | 2                        | 270                         | U5                  | European |
| 57        | 2                        | 192 256 270                 | U5                  | European |
| 58        | 1                        | 192 270                     | U5                  | European |
| 59        | 1                        | 256 270                     | U5                  | European |
| 60        | 4                        | 167 192 256 270 311 318     | U5a                 | European |
| 61        | 1                        | 189 256 362                 | U5a                 | European |
| 62        | 1                        | 256 270 342                 | U5a                 | European |
| 63        | 2                        | 172 183C 189 219 278        | U6a                 | European |
| 64        | 1                        | 172 183C 189 278            | U6a                 | European |
| 65        | 3                        | 153 298                     | V                   | European |
| 66        | 3                        | 298                         | V                   | European |
| 67        | 1                        | 187 298 311                 | V                   | European |
| 68        | 1                        | 291 298                     | V                   | European |
| 69        | 1                        | 189 223 278                 | X                   | European |
| 70        | 3                        | 126 223 278 290 319 362     | A                   | Amerindian |
| 71        | 1                        | 111 223 290 319 362         | A                   | Amerindian |
| 72        | 1                        | 111 223 290 319             | A                   | Amerindian |
| 73        | 1                        | 111 129 223 290 319 362     | A                   | Amerindian |
| 74        | 1                        | 111 126 223 259 290 319 362 | A                   | Amerindian |
| 75        | 1                        | 111 223 266 290 319 362     | A                   | Amerindian |
| 76        | 1                        | 111 223 269 290 319 360 362 | A                   | Amerindian |
| 77        | 1                        | 92 111 223 290 319 362      | A                   | Amerindian |
| 78        | 2                        | 178 183C 189 217            | B                   | Amerindian |
| 79        | 1                        | 189 217                     | B                   | Amerindian |
| 80        | 1                        | 178 183C 189 217 311        | B                   | Amerindian |
| 81        | 1                        | 178 183C 189 217            | B                   | Amerindian |
| 82        | 1                        | 83 189 217                  | B                   | Amerindian |
| 83        | 1                        | 183C 189 217                | B                   | Amerindian |
| 84        | 1                        | 189 217 249 312 344         | B                   | Amerindian |
| 85        | 1                        | 189 217 311 319             | B                   | Amerindian |
| 86        | 1                        | 189 217 311                 | B                   | Amerindian |
| 87        | 10                       | 223 298 325 327             | C                   | Amerindian |
| 88        | 1                        | 114 123 298 325 327         | C                   | Amerindian |
| 89        | 1                        | 223 224 298 311 325 327 356 | C                   | Amerindian |
| 90        | 1                        | 223 270 298 325 327         | C                   | Amerindian |
Spain, and Italy, respectively. Only two lineages of European origin (#49 and #50) present in Porto Alegre blacks were not observed in whites from the same city or in any of the European populations considered here. As with the Y markers, the mtDNA data also revealed that the Azorian presence in Porto Alegre was clearly detectable regardless of the demographic and cultural changes that occurred after the initial foundation/colonization of the city.

The same analysis with the 14 lineages of African origin found in the white sample revealed that 57% of them had perfect matches with those found in the regions of slave importation to Brazil (West, West-Central, and Southeast Africa; Salas et al., 2002; Plaza et al., 2004; Rosa et al., 2004; Beleza et al., 2005; Coia et al., 2005; Jackson et al., 2005). Three lineages (# 103, #104, and #116) exclusively matched those found in Angola, Cabinda, and Mozambique, countries inhabited by peoples who speak Bantu languages (Salas et al., 2002; Plaza et al., 2004; Beleza et al., 2005). Another lineage (#110) was also probably of Bantu origin since it occurs in Mozambique and in the Bassa ethnic group of Cameroon. Lineage #113 matched one found only in Guinea Bissau, whereas # 106 matched another found in Sierra Leone, both of which are West African countries (Rosa et al., 2004; Jackson et al., 2005). Two lineages (#108 and #109) had a geographical origin that was difficult to define since they occur in all sub-Saharan

| Lineage Number | Number of sequences | HVS-I mutations | Haplogroup | Origin |
|---------------|---------------------|-----------------|------------|--------|
| 91            | 1                   | 126 207 223 298 325 327 | C          | Amerindian |
| 92            | 1                   | 051 172 223 295 298 325 327 335 | C          | Amerindian |
| 93            | 1                   | 185 209 223 327 | C          | Amerindian |
| 94            | 1                   | 187 223 290 325 | C          | Amerindian |
| 95            | 2                   | 051 223 298 325 327 | C          | Amerindian |
| 96            | 1                   | 223 298 325 327 336 | C          | Amerindian |
| 97            | 1                   | 051 223 287 298 311 325 327 | C          | Amerindian |
| 98            | 1                   | 223 298 325 327 362 | C          | Amerindian |
| 99            | 1                   | 223 325 327 | C          | Amerindian |
| 100           | 1                   | 223 239 288 325 362 | D          | Amerindian |
| 101           | 1                   | 189 223 325 362 | D          | Amerindian |
| 102           | 1                   | 223 325 362 | D          | Amerindian |
| 103           | 3                   | 148 172 187 188G 189 223 230 311 320 | L0a2      | African |
| 104           | 1                   | 129 148 168 172 187 188G 189 223 230 278 293 311 320 | L0a1      | African |
| 105           | 1                   | 66 129 179 187 189 218 223 230 243 290 311 | L0d       | African |
| 106           | 2                   | 111 126 187 189 223 239 270 278 293 311 | L1b       | African |
| 107           | 1                   | 83 126 187 189 215 223 264 270 278 293 311 | L1b       | African |
| 108           | 1                   | 126 187 189 223 264 270 278 293 311 | L1b       | African |
| 109           | 1                   | 126 187 189 223 264 270 278 311 | L1b       | African |
| 110           | 1                   | 129 187 189 223 274 278 293 294 311 360 | L1c1      | African |
| 111           | 1                   | 134 187 213 223 265C 274 278 286G 360 | L1c2      | African |
| 112           | 1                   | 189 223 265C 274 286G 294 311 343T 360 | L1c2      | African |
| 113           | 1                   | 193 213 223 239 278 294 309 390 | L2aβ1      | African |
| 114           | 1                   | 223 292 320 | L3e2      | African |
| 115           | 1                   | 86 149T 152A 223 248 320 355 | L3e2      | African |
| 116           | 1                   | 209 223 311 | L3f       | African |

*The nucleotide positions considered for the analyses were from 16,050 to 16,391. Note that a value of 16,000 has been subtracted from each nucleotide position to make this column easier to read. Sequences were aligned with the revised reference sequence (rCRS; Andrews et al., 1999). *Haplogroup nomenclature is that recommended in the literature (see text). Those cases in which HVS-I information alone did not allow the identification of specific haplogroups were classified based on probabilities. Complete information about the African lineages observed in Porto Alegre blacks can be found in Hünemeier et al. (2007).
Africa. Only four African lineages were shared between the black and white Porto Alegre groups, with all four occurring in Africa and/or other Brazilian populations (Salas et al., 2002; Plaza et al., 2004; Rosa et al., 2004; Beleza et al., 2005; Coia et al., 2005; Jackson et al., 2005; Gonçalves et al., 2008).

The four major Amerindian haplogroups were detected among Porto Alegre whites (A = 28%; B = 22%, C = 47%, D = 3%), whereas haplogroup A was absent among blacks (B = 19%, C = 69% and D = 12%). Lineage #87 was the most common in both samples. Since this lineage contained the mutations that defined the C nodal branch, it matched several sequences found in admixed populations from southern Brazil, and in individuals from the Tupian and Jêan tribes; these different matches precluded identification of the precise origin of this lineage (Alves-Silva et al., 2000; Marrero et al., 2005, 2007). On the other hand, the Guarani contribution was clearly detected through the presence of lineages such as #75 (Marrero et al., 2007). However, most of the Amerindian lineages found in Porto Alegre did not match and/or cluster with Guarani mtDNA sequences. These results support the idea that the present Guarani mitochondrial genome may be a poor representative of that found at colonial times (Marrero et al., 2007a,b). On the other hand, the Guarani contribution was clearly detected through the presence of lineages such as #75 (Marrero et al., 2007b). However, most of the Amerindian lineages found in Porto Alegre did not match and/or cluster with Guarani mtDNA sequences. These results support the idea that the present Guarani mitochondrial genome may be a poor representative of that found at colonial times (Marrero et al., 2007a,b). This finding also suggests that other tribes may have made a more significant contribution, through their women, to the formation of the contemporary admixed Porto Alegre population.

Ancestral contributions

Table 3 summarizes the continental origins of the two Porto Alegre samples based on the mtDNA and Y-chromosome haplogroups. Although there was a significant introduction of non-European mtDNA sequences (Amerindian = 21%; African = 10%) among whites, Europe was still the major contributor in both genetic systems (mtDNA = 69%; Y-chromosome = 99%). This finding supported the general correspondence between physical appearance and maternal or paternal ancestry at the population level. In contrast, a completely different picture emerged when the black group was considered. Most of the mtDNA sequences (79%) had an African origin, but 56% of the Y-chromosomes were of European origin, while the Amerindian contribution involved both paternal and maternal inheritance.

These results for the black group can be partly explained by the fact that mulatos and pardos, who showed visible signs of admixture, were included in this sample. However, history and the dynamics of admixture could also have played an important role. In the early centuries of colonization, almost only European men migrated to Brazil, and for different reasons, African males were brought preferentially to Brazil during the slave trade (Matoso, 1982). This initial demographic asymmetry, and compulsory restrictions to the African male slaves reproduction, determined that the first Brazilians were born mostly from the union between European males and Amerindian or African females. Later, social practices determined that a child with more pronounced physical African features would be considered black, while those with more pronounced European features would be considered white. This situation created ample opportunity for the introduction of African mtDNA lineages and European Y-chromosomes into the white and black segments, respectively (Parra et al., 2003; Gonçalves et al., 2007). A second major European migratory movement during the 19th century that was particularly important for southern Brazil involved couples and families, not just males. This migration resulted in many white persons/populations with complete European genomes, whereas others are phenotypically white but show non-European admixture signs at the genome level (Marrero et al., 2005; Zembrzuski et al., 2006).

To investigate whether these differences between the two skin color groups were peculiar to Porto Alegre or whether they represented a more general tendency, we compiled the literature data for all of the estimates of African, European and Amerindian contributions in black and white Brazilian populations from different geographical regions. Table 4 shows that the pattern described above occurs throughout southern Brazil, but not in other regions of the country. In blacks from the Southeast, for example, the African component predominates in both paternal and maternal data sets, whereas among whites in this region the European contribution is particularly frequent when only Y chromosome markers are considered. Table 4 also show

Table 3 - Estimates of parental contribution based on mtDNA and Y-chromosome markers in whites and blacks from Porto Alegre.

| Samples and markers | Parental contribution (%) | Reference |
|---------------------|---------------------------|-----------|
|                     | European | Amerindian | African |         |
| White               |          |           |         |
| mtDNA               | 69       | 21        | 10      | Present study |
| Y-chromosome        | 99       | 0         | < 1     | Present study |
| Black               |          |           |         |
| mtDNA               | 5        | 16        | 79      | Present study; Hünemeier et al. (2007) |
| Y-chromosome        | 56       | 6         | 38      | Present study; Hünemeier et al. (2007) |
that the whites from the North and South present large differences, basically related to the Amerindian and European women contribution; the extensive admixture between Indians and non-Indians ended in southern Brazil at least 170 years ago, whereas in northern Brazil, especially in the Amazon basin, Amerindian genes are still being introduced into non-native urban and rural populations. The results of these discontinuous vs. continuous patterns of gene flow can be seen by comparing the mtDNA results mentioned above with the biparental loci admixture values (21% vs. 54% and 13% vs. 44% for the Amerindian component in white samples from the South and North, respectively). In

Table 4 - Estimates of parental contribution using three set of markers and considering persons classified as white or black in different regions of Brazil.

| Region and samples | N | Genetic system | Parental contribution (%)<sup>a</sup> | Reference |
|--------------------|---|----------------|-------------------------------------|-----------|
| Whitess<br>North   | 48 mtDNA<sup>b</sup> | 31 15 54 | Alves-Silva et al. (2000) |
|                   | 49 Y-chromosome<sup>c</sup> | 98 2 0 | Carvalho-Silva et al. (2001) |
|                   | 48-2,054 Bi-parental loci<sup>d</sup> | 53 3 44 | Salzano and Bortolini (2002) |
| Northeast<br>      | 50 mtDNA<sup>b</sup> | 34 44 22 | Alves-Silva et al. (2000) |
|                   | 49 Y-chromosome<sup>c</sup> | 96 4 0 | Carvalho-Silva et al. (2003) |
|                   | 64-27,607 Bi-parental loci<sup>d</sup> | 72 23 5 | Salzano and Bortolini (2002) |
| Southeast<br>      | 99 mtDNA<sup>b</sup> | 31 35 34 | Alves-Silva et al. (2000) |
|                   | 50 Y-chromosome<sup>c</sup> | 96 4 0 | Carvalho-Silva et al. (2001) |
|                   | 89-60,270 Bi-parental loci<sup>d</sup> | 56 39 5 | Salzano and Bortolini (2002) |
| South              | 328 mtDNA<sup>b</sup> | 63 16 21 | Alves-Silva et al. (2000); Marrero et al. (2005); present study |
|                   | 255 Y-chromosome<sup>c</sup> | 99 0 < 1 | Carvalho-Silva et al. (2001); present study |
|                   | 107-5,527 Bi-parental loci<sup>d</sup> | 73 14 13 | Salzano and Bortolini (2002) |
| Blackss<br>North   | 270 mtDNA<sup>b</sup> | 3 57 40 | Silva-Junior et al. (2006); Ribeiro-dos-Santos et al. (2006) |
|                   | ND Y-chromosome<sup>c</sup> | ND ND ND | |
|                   | 38-482 Bi-parental loci<sup>d</sup> | 25 33 42 | Salzano and Bortolini (2002) |
| Northeast<br>      | 39 mtDNA<sup>b</sup> | 21 69 10 | Bortolini et al. (1997); Silva-Junior et al. (2006) |
|                   | 89 Y-chromosome<sup>c</sup> | 34 64 2  | Abe-Sandes et al. (2004) |
|                   | 30-38,898 Bi-parental loci<sup>d</sup> | 38 55 7 | Salzano and Bortolini (2002) |
| Southeast<br>      | 233 mtDNA<sup>b</sup> | 2 89 9 | Silva-Junior et al. (2006); Gonçalves et al. (2007); Hünemeier et al. (2007); present study |
|                   | 288 Y-chromosome<sup>c</sup> | 43 56 1  | Abe-Sandes et al. (2004); Gonçalves et al. (2007); Hünemeier et al. (2007); present study |
|                   | 378-33,534 Bi-parental loci<sup>d</sup> | 23 77 0 | Salzano and Bortolini (2002) |
| South              | 226 mtDNA<sup>b</sup> | 4 84 12 | Bortolini et al. (1997); Hünemeier et al. (2007); present study |
|                   | 55 Y-chromosome<sup>c</sup> | 56 38 6 | Hünemeier et al. (2007); present study |
|                   | 53-3,236 Bi-parental loci<sup>d</sup> | 36 51 13 | Salzano and Bortolini (2002) |

<sup>a</sup>ND = No data available; <sup>b</sup>HVS-I sequences and SNPs in the mtDNA codifying region; <sup>c</sup>SNPs in the non-recombining region of the Y-chromosome; <sup>d</sup>Classic blood group plus protein polymorphisms.
contrast, groups or populations identified as white in the Northeast and Southeast had very similar proportions of Amerindian, European and African ancestries in all of the genetic systems investigated. Since European colonization started almost simultaneously in the Northeast and Southeast, this similarity suggests that the dynamics of admixture were similar in both regions.

These striking differences between and within the skin color groups in Brazilian regions can be due to several factors, and some can be the same as those presented for the Porto Alegre case. The type of classification employed to classify the samples in the different studies is also important. Both self and interviewer classifications are full of subjectivities (Salzano and Bortolini, 2002). Distinct admixture dynamics due to historical events and social practices should also be considered. Overall, our results corroborate the idea that, despite some general trends, there is a notable heterogeneity in the pattern of admixture within and among populations/groups in Brazil (Bortolini et al., 1999; Alves-Silva et al., 2000; Marrerro et al., 2005). Our findings also highlight the intricacies of past and present patterns of mating in a complex society with a relatively recent multiethnic origin, and the relative instability of phenotypic classifications within this society.

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