Analysis of paternal lineages in Brazilian and African populations

Mónica Carvalho¹, Pedro Brito¹, Virgínia Lopes¹, Lisa Andrade¹, Mª João Anjos¹, Francisco Corte Real²,³ and Leonor Gusmão⁴

¹Forensic Genetics Service, Centre Branch, National Institute of Legal Medicine, Coimbra, Portugal.
²National Institute of Legal Medicine, Coimbra, Portugal.
³Faculty of Medicine, University of Coimbra, Coimbra, Portugal.
⁴Institute of Pathology and Molecular Immunology, University of Porto, Porto, Portugal.

Abstract

The present-day Brazilian population is a consequence of the admixture of various peoples of very different origins, namely, Amerindians, Europeans and Africans. The proportion of each genetic contribution is known to be very heterogeneous throughout the country. The aim of the present study was to compare the male lineages present in two distinct Brazilian populations, as well as to evaluate the African contribution to their male genetic substrate. Thus, two Brazilian population samples from Manaus (State of Amazon) and Ribeirão Preto (State of São Paulo) and three African samples from Guinea Bissau, Angola and Mozambique were typed for a set of nine Y chromosome specific STRs. The data were compared with those from African, Amerindian and European populations. By using Y-STR haplotype information, low genetic distances were found between the Manaus and Ribeirão Preto populations, as well as between these and others from Iberia. Likewise, no significant distances were observed between any of the African samples from Angola, Mozambique and Guinea Bissau. Highly significant Rst values were found between both Brazilian samples and all the African and Amerindian populations. The absence of a significant Sub-Saharan African male component resulting from the slave trade, and the low frequency in Amerindian ancestry Y-lineages in the Manaus and Ribeirão Preto population samples are in accordance with the accentuated gender asymmetry in admixture processes that has been systematically reported in colonial South American populations.

Key words: chromosome Y, STRs, lineages, Brazil, Africa.

Received: August 19, 2009; Accepted: February 23, 2010.

Introduction

South America was already inhabited when the first European settlers arrived. The first of these, more specifically the Portuguese, landed in Brazil in 1500, in territory already occupied by Amerindians. The region was quickly colonized, with the intention to explore its natural resources. Throughout the history different people from all over the world arrived at this territory, especially in the colonial period, with a great affluence from Africa due to the slave trade, thereby initiating the admixture of Amerindians, Africans and Europeans. In the 16th century, on the occasion of the arrival of the Portuguese, the native population was around 2.5 million. Between the 16th and 19th centuries, about 4 million Sub-Saharan African slaves arrived in Brazil (IBGE, 2000). Initially, admixture was mainly between native females and Portuguese male navigators, due to the insignificant immigration of European women (Carvalho-Silva et al., 2001). The subsequent migration contributed to a constant influx of both Africans and Europeans, thereby giving rise to the present very heterogeneous population, due to the different proportions of local admixture throughout the country (Ribeiro, 1995).

In this work, we attempted to study this historic influence on the genetic background of present-day populations, by analyzing two Brazilian populations, one from Manaus (State of Amazon) and the other from Ribeirão Preto (State of São Paulo). Three African populations, from Guinea Bissau, Angola and Mozambique were also included in the study, since they represent the ancestral populations of most Africans that arrived in Brazil during the period of the slave trade. The five populations were genetically characterized for Y chromosome specific STR loci by typing 9 markers (DYS19, DYS389 I, DYS389 II, DYS390, DYS391, DYS392, DYS393 and DYS385, the latter including 2 loci), whereas the Brazilian samples were typed for five SNP markers (M2, M3, M35, M213 and SRY10831). In order to evaluate the possible male contributions to our samples, a comparison was made between...
our data and those already available for Brazilian, Amerindian, African and European populations.

Materials and Methods

DNA samples

The Y-chromosomal minimal haplotype was defined by 9 Y-STRs (DYS19, DYS389 I, DYS389 II, DYS390, DYS391, DYS392, DYS393, DYS385) in unrelated individuals from Manaus (N = 42), Ribeirão Preto (N = 65), Guinea Bissau (N = 32), Angola (N = 48) and Mozambique (N = 36). The samples from Brazil, Guinea Bissau and Mozambique were collected in district hospitals. The Brazilian samples were composed of individuals from the cities Manaus and Ribeirão Preto. The African samples from Guinea Bissau and Mozambique consisted of individuals living in the regions of Bissau and Maputo, respectively. The samples from Angola were collected in the northern region of the country, and included individuals from the villages of N’Dalatando and Lucala, in Kwanza province, and from the province of Uíge. Blood samples were obtained with written informed consent.

Marker typing

For samples from Manaus and Ribeirão Preto, the minimal haplotype was typed using a PowerPlexY PCR Amplification Kit (Promega), with primers and amplification conditions according to manufacturer’s instructions. As regards the others from Guinea Bissau, Angola and Mozambique, the STRs DYS19, DYS389 I, DYS389 II, DYS390 and DYS393 were amplified as described by Gusmão et al. (1999). DYS385 amplification conditions complied with the methodology, as described by Schneider et al. (1998), whereas multiplex amplification of DYS391, DYS392, DYS393 was according to Kloosterman et al. (1998). Alleles were designated according to the International Society for Forensic Genetics (ISFG) guidelines for forensic analysis using Y-STRs (Gusmão et al., 2006). For defining male haplogroups in population samples from Manaus and Ribeirão Preto, five Y-chromosome SNP markers (M2, M3, M35, M213 and SRY10831) were genotyped using methods as described by Silva et al. (2006). Haplogroup nomenclature was according to Karafet et al. (2006).

Statistical analysis

Both haplotype diversity, according to Nei (1987), and pairwise Rst genetic distances were calculated using Arlequin v. 3.0 software Excoffier et al. (2005), without considering DYS385. Rst genetic distances were visualized in two-dimensional space by using the Multi Dimensional Scaling (MDS) method included in the StatSoft, Inc. (2007) STATISTICA data analysis software system, version 8.0.

Results and Discussion

Y haplotype diversity

The haplotype results obtained for the STRs (DYS19, DYS389 I, DYS389 II, DYS390, DYS391, DYS392, DYS393 and DYS385) are provided as Supplementary Material (Tables S1 to S5). Haplotype diversity was estimated in all studied populations (Table 1). A comparison of haplotype diversity revealed high levels in Manaus and Ribeirão Preto, comparable to that observed in the African samples. Although higher diversity could be expected in African samples, the presence of male lineages of different origins in the Brazilian populations might have contributed to incrementing diversity.

In the sample from Manaus, all individuals presented different haplotypes (42 unique ones) with overall haplotype diversity of 1.000 ± 0.0052. In Ribeirão Preto, 58 different haplotypes were observed, 54 of which unique. In Ribeirão Preto, the most common haplotypes represented in more than one individual (RP1, RP5, RP6 and RP19 - Table S2) correspond to or are just a few steps apart from, the most frequent haplotype in all Iberian populations (Gusmão et al., 2003), which represents the core haplotype within the R1b1b2-M269 haplogroup. In Manaus, this core haplotype was encountered in only one individual (M3), whereas one or two step neighbors were found in 10 individuals (M1, M4, M10, M21, M22, M30, M37, M38, M39 and M42). Therefore, by analyzing this set of Y-STRs, it is possible to infer an important male-mediated European genetic influx in both of the Brazilian populations studied.

29 different haplotypes were observed in the Guinea Bissau population, 27 of which unique. In Angola, there were 42 different haplotypes, 36 unique. Finally, in Mo-

| Population        | N  | Number of different haplotypes | Number of unique haplotypes | Haplotype diversity |
|-------------------|----|--------------------------------|-----------------------------|---------------------|
| Manaus            | 42 | 42                             | 42                          | 1.0000 ± 0.0052     |
| Ribeirão Preto    | 65 | 58                             | 54                          | 0.9947 ± 0.0044     |
| Guinea Bissau     | 32 | 29                             | 27                          | 0.9919 ± 0.0110     |
| Angola            | 48 | 42                             | 36                          | 0.9947 ± 0.0054     |
| Mozambique        | 36 | 34                             | 32                          | 0.9968 ± 0.0075     |
zambia and 32 unique haplotypes were observed from a total of 34 different ones. Four different haplotypes are shared between Angola and Mozambique, and one between Angola and Guinea Bissau. Three of these shared haplotypes match the Bantu core described by Thomas et al. (2000), besides one differing by only one mutation step. When searching for shared haplotypes between African and Brazilian samples, a single hit was found between Angola and Manaus (M25 = A20), this also matching the Bantu modal. Apart from this haplotype found in Manaus, a search in both Brazilian samples did not reveal the presence of any other haplotype corresponding to the Bantu core. Only one haplotype was found in Ribeirão Preto that differed by a single step from the Bantu modal. Therefore, based on Y-STR results, a significant male-mediated African genetic influx could not be expected in both of these Brazilian populations.

Population comparison

The population samples from Manaus, Ribeirão Preto, Guinea Bissau, Mozambique and Angola were compared with different populations (listed in Table 2) through pairwise RST genetic distance analysis. The results obtained showed no significant differences between Manaus and Ribeirão Preto (RST = 0.0002, p = 0.4030). The same was observed when comparing these two populations with other urban and/or admixed South American (codes 3, 4, 5, 6, 7 and 12 in Table 2) or Iberian (13, 14, 15 and 16 - Table 2) populations, with RST values below 0.0011 (p > 0.0059). Among other American populations, a significant differentiation was found between Manaus or Ribeirão Preto, and African descendents from Rio de Janeiro (RST > 0.0817, p = 0.0002) and South Amerindians (populations 9, 10 and 11 in Table 2), RST > 0.0997 (p = 0.0015).

Within the African group, although p-values were not significant (p > 0.01), higher RST values were observed between the Guinea Bissau, Angola and Mozambique samples (0.00998 < RST < 0.05381) than those observed between the Brazilian and European samples. Highly significant RST values were found when comparing the Manaus and Ribeirão Preto with all the African populations (RST > 0.1311, p = 0.0000).

In multidimensional scaling (MDS) plot of pairwise RST genetic distances, based on Y-STR data (Figure 1), it is possible to note the formation of different clusters, these including a European and an African group, as well as two

Table 2 - List of South American, African and European populations used in population comparison analysis.

| Code | Population | N   | Reference                      |
|------|------------|-----|--------------------------------|
| 1    | Manaus - Brazil, State of Amazon | 42  | this study                     |
| 2    | Ribeirão Preto - Brazil, State of São Paulo | 65  | this study                     |
| 3    | Santa Catarina - Brazil, State of Santa Catarina | 109 | Caíné et al., 2005             |
| 4    | Belém - Brazil, State of Pará | 200 | Palha et al., 2007             |
| 5    | Rio de Janeiro - Brazil, State of Rio de Janeiro | 245 | Goes et al., 2005              |
| 6    | São Paulo - Brazil, State of São Paulo | 200 | Gois et al., 2007              |
| 7    | Rio Grande Sul - Brazil, State of Rio Grande do Sul | 203 | Leite et al., 2008             |
| 8    | Rio de Janeiro - Brazil, State of Rio de Janeiro (African descendents) | 135 | Domingues et al., 2007        |
| 9    | Rio Grande do Sul - Brazil, Guarani and Kaingang (Amerindians) | 42  | Leite et al., 2008             |
| 10   | Argentina, northern region, Colla - (Amerindians) | 48  | Toscanini et al., 2008         |
| 11   | Argentina, northwestern region, Toba (Amerindians) | 49  | Toscanini et al., 2008         |
| 12   | Buenos Aires - Argentina | 100 | Sanchez-Díaz et al., 2008      |
| 13   | Portugal, northern region | 244 | Sanchez-Díaz et al., 2008      |
| 14   | Portugal, central region | 100 | Bento et al., 2009             |
| 15   | Portugal, southern region | 100 | Sanchez-Díaz et al., 2008      |
| 16   | Spain | 148 | Martin et al., 2004            |
| 17   | Guinea Bissau | 32  | this study                     |
| 18   | Angola | 48  | this study                     |
| 19   | Mozambique | 36  | this study                     |
| 20   | Guinea Equatorial | 101 | Arroyo Pardo et al., 2005      |
| 21   | Cabinda - Angola | 208 | Beleza, 2005                   |
| 22   | Maputo - Mozambique, | 112 | Alves et al., 2003             |
| 23   | São Tome and Príncipe | 103 | Trovoada et al., 2001          |
| 24   | Cape Verde | 47  | Corte-Real et al., 2000        |
other clusters formed by European/Amerindian and Euro-
pean/African mixed populations. As expected, populations
from the Guinea Bissau, Angola and Mozambique groups
are on a line with other populations from continental Af-
rica. Manaus and Ribeirão Preto clearly group with Euro-
pean populations from Iberia, together with other urban
population samples from South America. These two popu-
lations stand well apart from the remaining clusters of Afri-
can, Amerindian or admixed ancestry.

Y-SNP haplogroups in the Brazilian populations

Five SNPs were typed in the Manaus and Ribeirão
Preto samples, in order to trace the origin of the Y-chromo-
somes in the current population. In each population sample,
a single chromosome belonged to the most frequent Sub-
Saharan African haplogroups by carrying the M2 mutation
(Tables S1 and S2). In Ribeirão Preto, a second African lin-
eage could be found, which lacks the M213 and SRY10831
mutations, therefore being classified in paragroup A*. One
out of 65 samples from Ribeirão Preto and 4 out of 42 from
Manaus carried both the M213 and M3 mutations that char-
acterize Amerindian haplogroups. The remaining samples,
were classified in F* (except Q3), E1b1b1 - M35 or Y*
(xA, E1b1a-b1, F*).

Based on the SNP results, we concluded that Europe
is the main source of paternal lineages existing in the pres-
ent-day population of Ribeirão Preto (95.4%), with African
and Amerindian lineages only representing 3.1% and 1.5%
of the chromosomes, respectively. In Manaus, the origin of
most chromosomes can also be traced to Europe (88.1%),
although a higher Amerindian component was found
(9.5%). Only a single African-ancestry chromosome
(2.4%) was detected in the sample from Manaus.

Conclusions

Many studies have been carried out to characterize
the genetic diversity of Brazilian populations aiming to
better understand colonization processes and the demo-
ographic history of its native populations (e.g. Bortolini et al.
2003; Abe-Sandes et. al. 2004; Silva et al. 2006). These
studies systematically revealed a particularly sub-struc-
tured country, with populations from distinct regions dif-
fering in their proportion of African, Amerindian and Euro-
pean ancestries. Studies of mtDNA and Y-chromosome
markers also revealed much higher genetic differentiation
at the maternal gene-pool level than at the paternal counter-
part (Marrero et al. 2005). Indeed, as regards Y chromo-
some lineage, a high European contribution was observed
in most Brazilian samples. This was also evident in the
present study, where Brazilian samples, as well as all other
general population samples countrywide, presented much
lower genetic distances when compared with Europeans
than with Africans or Amerindians. A European contribu-
tion was also evident in the South Amerindian sample stud-
yed by Leite et al. (2008) which presented a similar distance
(RST = 0.18) to the Toba Amerindian sample and any of the
Iberian samples. Nevertheless, from the SNP results it can
be inferred that, although genetic distance analysis based
on STR profiles allowed to identify the main European con-
tribution to the Brazilian samples, it was not able to detect
minor contributions. In fact, the almost 10% Amerindian
contribution to the Manaus sample was insufficient to pro-
duce significant genetic distance values between Manaus
and both Ribeirão Preto or Iberian populations.

The absence of a significant Sub-Saharan African
male component resulting from the slave trade or Amerin-
dian ancestry Y-lineages, in the Manaus and Ribeirão Preto
population samples, is in accordance with pronounced gen-
der asymmetry in admixture processes that has been sys-
tematically reported in colonial South American popula-
tions.

Acknowledgments

We acknowledge funding by Fundação para a Ciência
e a Tecnologia, POCI 2010.

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Internet Resources

IBGE - Instituto Brasileiro de Geografia Estatistica. http://www.ibge.gov.br (March 10, 2009).
STATISTICA - Data Analysis Software System. 
http://www.statsoft.com (July 18, 2009).

Supplementary Material

The following online material is available for this article:
Table S1 - Y chromosome haplotype distribution in the Manaus population sample (N = 42).
Table S2 - Y chromosome haplotype distribution in the Ribeirão Preto population sample (N = 65).
Table S3 - Y chromosome haplotype distribution in the Guinea Bissau population sample (N = 32).
Table S4 - Y chromosome haplotype distribution in the Angola population sample (N = 48).
Table S5 - Y chromosome haplotype distribution in the Mozambique population sample (N = 36).
This material is available as part of the online article from http://www.scielo.br/gmb.

Associate Editor: Francisco Mauro Salzano

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