Int J Legal Med (2012) 126:975–976
DOI 10.1007/s00414-012-0739-x

POPULATION DATA

Genetic profile characterization of ten X-STRs in a sample from Paraná, Brazil

Luciellen d`Avila Giacomel Kobachuk · Marcelo Malaghini · José Samuel da Silva · Leonor Gusmão · Maria da Graça Bicalho

Received: 23 January 2012 / Accepted: 28 June 2012 / Published online: 20 July 2012

Abstract This work reports the allele frequencies for ten X-STRs (DXS8378, DXS7132, DXS9898, DXS6809, DXS9902, DXS6789, DXS7133, DXS7423, GATA172D05, GATA31E08) in a sample of 800 individuals from Paraná, Brazil. No deviations from the Hardy–Weinberg equilibrium were observed. Linkage disequilibrium analysis did not reveal association between the X-STRs. High overall power of discrimination was obtained for female and male samples, and high probability of exclusion was observed in father/mother/daughter trios and father/daughter duos. Genetic comparisons revealed significant differences between Paraná and other Brazilian populations.

Keywords X chromosome · STRs · Human identification · Genetic population data · Paraná · Brazil

Several X-STRs multiplex PCR systems have recently been validated for forensic use [1–5], but few are common to different works. Recently, a decaplex system was developed and validated that was carried out by the Spanish and Portuguese-Speaking Working Group of the International Society For Forensic Genetics (GHEP-ISFG) working group [6]. The present study aimed at the genetic characterization of the ten X-STRs standardized by the GHEP-ISFG in Paraná, Brazil.

A total of 401 male and 399 female samples from unrelated subjects were selected from paternity cases performed in Laboratório de Genética Molecular Forense of Polícia Científica do Paraná. The samples were obtained by fingertip puncture, after signing of consent. This study was approved by the Research Ethics Committee of the Division of Health Sciences, Federal University of Paraná. DNA extraction, PCR, analysis on an ABI310 genetic analyzer, and statistical evaluation were performed as described before [6, 7]. The allele frequencies and the results of the forensic efficiency parameter calculations are given in Table S1 and Table S2. For a significance level of 0.005 (after the Bonferroni correction), no deviations from the Hardy–Weinberg equilibrium were observed for any of the analyzed loci. An allele not previously described was found in DXS6809 locus with a size corresponding to 34.1 repeats. In our study, GATA172D05 was the most informative marker and DXS7133 was the least informative. Overall, the power of discrimination was 0.99999999995 for females and 0.9999994 for males. The combined probability of exclusion was 0.999997 for trios and 0.99987 for duos. For a significance level of 0.001 (after the Bonferroni correction for 45 comparisons) in the linkage disequilibrium (LD) analysis, a significant p value was obtained only for GATA172D05-DXS7133 between alleles 13 and 8, respectively. Those loci

---

Electronic supplementary material The online version of this article (doi:10.1007/s00414-012-0739-x) contains supplementary material, which is available to authorized users.

L. d`Avila Giacomel Kobachuk (✉) · J. S. da Silva ·
M. G. Bicalho
Laboratório de Imunogenética e Histocompatibilidade,
Universidade Federal do Paraná,
Centro Politécnico, Avenida Cel. Francisco H. dos Santos,
81530-900 Curitiba, Paraná, Brazil
e-mail: lugiacomel@uol.com.br

L. d`Avila Giacomel Kobachuk · M. Malaghini
Laboratório de Genética Molecular Forense,
Instituto de Criminalística,
Avenida Visconde de Guarapuava, 2652,
80010-100 Curitiba, Paraná, Brazil

L. Gusmão
Institute of Molecular Pathology and Immunology
of the University of Porto (IPATIMUP),
4200-465 Porto, Portugal

occupied a physical distance of 4 Mb. However, LD does not depend exclusively on the physical distance between loci, but it may result from random genetic drift, founder effects, recent interethnic admixture, population stratification, and sampling effects [5, 8, 9]. Therefore, the linkage disequilibrium that we detected might be spurious or due to sampling effects, corroborating the suggestion by Martins et al. [8].

A pairwise population comparison between Paraná and other Brazilian populations with available data for the same markers studied in our sample [4–6, 8, 10] was performed at a single locus level (F_{ST} genetic distance analysis). The results (Supplementary Table S3) showed significant differences ($p<0.05$) for São Paulo (DXS8378, DXS6789 and DXS7133), Rio de Janeiro (DXS7133), Belo Horizonte (DXS7423 and DXS7133), Mato Grosso (GATA31E08), Santa Catarina (DXS9902 and GATA31E08), Rio Grande do Sul (DXS7423 and DXS9898), Belém (DXS9898, DXS7133, GATA31E08), and Amazonas (DXS7133 and GATA31E08) populations.

This study was pioneer in the analysis of X-STRs decaplex (DXS8378, DXS9902, DXS7132, DXS9898, DXS6809, DXS6789, DXS7133, GATA172D05, GATA31E08 e DXS7423) in 800 unrelated individuals among the population of the State of Paraná. The previously published studies for the same markers analyzed Brazilian populations originating from different geographical regions in Brazil; it is important to highlight that there are differences in the ethnic formation related to these different regions as well as the expressively lower sampling employed in these previous studies.

Acknowledgments The authors would like to acknowledge the Universidade Federal do Paraná, the Laboratório de Imunogenética e Histocompatibilidade and Polícia Científica do Paraná. This work was supported by the Fundação da Universidade Federal (FUNPAR).

References

1. Tabbada KA, Ungria MCA, Faustino LP, Athanasiadou D, Stradmann-Bellinghausen B, Schneider PM (2005) Development of a pentaplex X-chromosomal short tandem repeat typing system and population genetic studies. Forensic Sci Int 154:173–180
2. Gomes I, Alves C, Maxzud K, Pereira R, Prata MJ, Sánchez-Diz P, Carracedo A, Amorim A, Gusmão L (2007) Analysis of 10 X-STRs in three African populations. Forensic Sci Int Genet 1:208–211
3. Robino C, Giolitti A, Gino S, Torre C (2006) Development of two multiplex PCR systems for the analysis of 12 X-chromosomal STR loci in a northwestern Italian population sample. Int J Leg Med 120:315–318
4. Cainé LM, Zarrabeitia MT, Riancho JA, Pinheiro MF (2010) Genetic data of a Brazilian population sample (Santa Catarina) using an X-STR decaplex. J Forensic Leg Med 17:272–274
5. Ribeiro-Rodrigues EM, Palha TJBF, Bittencourt EA, Ribeiro-dos-Santos A, Santos S (2011) Extensive survey of 12 X-STRs reveals genetic heterogeneity among Brazilian populations. Int J Leg Med 125:445–452
6. Gusmão L, Sánchez-Diz P, Alves C, Gomes I, Zarrabeitia MT, Abovich M, Atmetlla I, Bobillo C, Bravo L, Builes J, Cainé L, Calvo R, Carvalho E, Carvalho M, Cicarelli R, Catelli L, Corach D, Espinoza M, García O, Malaghini M, Martins J, Pinheiro F, Porto MJ, Raimondi E, Riancho JÁ, Rodríguez A, Rodríguez A, Cardozo AR, Schneider V, Silva S, Tavares C, Toscanini U, Vullo C, Whittle M, Yurrebaso I, Carracedo A, Amorim A (2009) A GEP–ISFG collaborative study on the optimization of an X-STR decaplex: data on 15 Iberian and Latin American populations. Int J Leg Med 123:227–234
7. Lareu MV, Phillips CP, Carracedo A, Lincoln PJ, Syndercombe Court D, Thomson JA (1994) Investigation of the STR locus HUMTH01 using PCR and two electrophoresis formats: UK and Galician Caucasian population surveys and usefulness in paternity investigations. Forensic Sci Int 66:41–52
8. Martins JÁ, Costa JC, Paneto GG, Figueiredo RF, Gusmão L, Sánchez-Diz P, Carracedo A, Cicarelli RMB (2010) Genetic profile characterization of 10 X-STRs in four populations of the southeastern region of Brazil. Int J Leg Med 124(5):427–432
9. Chakravarti A (1999) Population genetics: making sense out of a sequence. Nat Genet 21:56–60
10. Ribeiro-Rodrigues EM, Leite FPN, Hutz MH, Palha TJBF, Santos AKCR, Santos SEB (2008) A multiplex PCR for 11 X chromosome STR markers and population data from a Brazilian Amazon region. Forensic Sci Int Genet 2:154–158