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POPULATION DATA

Analysis of 15 autosomal STR loci from Mar del Plata and Bahia Blanca (Central Region of Argentina)

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Abstract Allele frequencies for the 15 short tandem repeats (STRs) loci included in the AmpFlSTR® Identifiler kit were estimated in a sample of unrelated individuals from Mar del Plata (MDQ; $N=180$) and Bahia Blanca (BB; $N=85$) (Buenos Aires, Argentina). Biological samples were obtained from voluntary donors and forensic cases. Both populations were in Hardy–Weinberg equilibrium after Bonferroni correction, except for locus vWA in MDQ and D2S1338 in BB. FGA was the most informative locus, and the least discriminating locus was TPOX in both samples. The combined power of discrimination (PDc) and the combined probability of exclusion (PEc) were similar in MDQ and BB samples ($0.999999998<PDc<0.999999999$ and $0.999999979<PEc<0.999999989$). The multidimentional scaling plot from Rst genetic distance matrix and the interethnic admixture estimation supported a higher European contribution in populations of the central region compared with populations from other regions of Argentina with higher Amerindian composition. These results enlarge the Argentine databases of autosomal STR loci, revealed as an excellent tool for human identification tests and population genetic analysis.

Keywords Autosomal STRs · Admixed population · Population genetic structure

The Argentina population is the product of the confluence of different geographical groups, basically from Native American and European ancestry. In the last two centuries, massive migratory waves from Europe, mainly Spain and Italy, settled in the Buenos Aires Province [1]. Since the ancestral proportion of the current cosmopolitan Argentinean population is not homogeneously distributed across the country, an updated picture on the population structure and genetic diversity is of fundamental interest to medical, forensic, and anthropological research. In this sense, the study of Mar del Plata (MDQ) and Bahia Blanca (BB) populations will provide new information about the genetic composition of the central area and will help to understand the biological relationships established among different regions of the country. Besides, the analysis of 15 autosomal short tandem repeat (STR) data will contribute to develop the Argentine databases of autosomal STRs.

A total of 265 blood samples of unrelated individuals from Mar del Plata and Bahia Blanca were analyzed (Supplemental Figure 1). The samples were collected with personal informed consent from volunteer donors who attended the regional hospital and the local private Hemocentro of MDQ ($N=90$) and from forensic cases (MDQ, $N=90$; BB, $N=85$) analyzed in the genetic laboratory of the Ministerio de Seguridad de Buenos Aires.

DNA was obtained using the standard phenol–chloroform method for the samples from the medical centers and by using FTA classic cards, as described by the manufacturer, for the

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forensic samples (http://www.whatman.com). Multiplex PCR amplification of 15 autosomal STRs (D8S1179, D21S11, D7S820, CSF1PO, D3S1358, TH01, D13S317, D16S539, D21338, D19S443, vWA, TPOX D18S51, D5S818, and FGA) was performed using AmpF*l*STR® Identifiler PCR Amplification kit (Applied Biosystems, Foster City, CA, USA) according to the manufacturer's instructions. For genetic typing, an ABI Prism 3130 DNA Genetic Analyzer along with GeneMapper ID 3.2.1 software (Applied Biosystems, Foster City, CA) was used.

The quality of the genetic profiles was guaranteed through control testing of the Argentine Society of Forensic Genetics (http://www.sagf.org.ar), and the study followed the guidelines and recommendations suggested by Carracedo et al. [2], Poetsch et al. [3], and the International Society of Forensic Genetics (http//www.isfg.org).

Allele frequencies, heterozygozities, and exact test for Hardy–Weinberg (HW) equilibrium were performed by Arlequin v.3.5 (http://cmpg.unibe.ch/software/arlequin35/) for the 15 autosomal STR markers in both MDQ (Supplemental Table 1) and BB (Supplemental Table 2) populations. Since allele frequencies of medical centers' and forensic samples from Mar del Plata did not differ, they were together considered as an only one set of samples. STR markers were in Hardy–Weinberg equilibrium after Bonferroni correction in both samples except vWA locus in MDQ and D2S1338 in BB.

The forensic statistical parameters were obtained using PowerStats v.1.2 software (Promega Corp.) (Supplemental Table 1 and Supplemental Table 2). The most informative system in MDQ and BB samples was FGA, with the highest average power of discrimination (PD=0.9690 and PD=0.9581, respectively) and typical paternity index (TPI=3.980 and TPI=3.770, respectively). On the other hand, TPOX marker showed the lowest values for all parameters also analyzed in both samples. These results were according to other studies in cosmopolitan populations of Argentina (Supplemental Table 3). The combined power of discrimination (PDc) and the combined probability of exclusion (PEc) for the 15 STR loci were 0.999999999 and 0.999999979, respectively, for MDQ and 0.999999998 and 0.999999989 for BB.

In order to analyze the population structure, these samples were compared with those published by other authors for admixed Argentinean populations and for parental populations (Supplemental Figure 1 and Supplemental Table 3). Genotype datasets were reconstructed using R software (http://www.r-project.org). Test of population differentiation, analysis of molecular variance (AMOVA), and pairwise Rst values were performed by Arlequin v.3.5 (http://cmpg.unibe.ch/software/arlequin35/). AMOVA results presented a low degree of genetic differentiation among admixed Argentinean populations (Fst=0.0121), appropriate for forensic markers [4]. Multidimensional scaling (MDS) plot was performed based on the Rst distances matrix using the R software (Supplemental Figure 2). Populations from central region and northeast of Argentina were not significantly different and represented a homogeneous cluster, contrasting with northwestern and southern Patagonian populations closer to Amerindian parental population. Interethnic admixture was calculated using ADMIX 95 software (www.genetica.fmed.edu.uy/software.htm). The estimated contributions in MDQ sample were 0.771 s.e.±0.0009 European, 0.014 s.e.±0.0007 African, and 0.215 s.e.±0.0004 Amerindian. And for BB sample, the proportions were 0.678 s.e.±0.0006, 0.042 s.e.±0.0005, and 0.278 s.e±0.0003, respectively. These results are consistent with previous studies for blood systems [5, 6] and ancestry informative markers (AIMs) [7], where European contribution is higher in central region of Argentina and decreases toward north and south at the same time that the Amerindian contribution increases.

In conclusion, the analyses of the 15 autosomal STR markers from AmpFlSTR® Identifiler PCR Amplification kit indicated a mainly European contribution to Buenos Aires Province populations of Mar del Plata and Bahia Blanca, in contrast with other regions of Argentina. These results enlarged the Argentine databases of autosomal STR loci and aimed to constitute these markers as an excellent tool for human identification tests and population genetic analysis of admixed Argentinean populations.

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