



Correspondence

Population genetic analyses of the PowerPlex[®] Fusion kit in a cosmopolitan sample of Chubut Province (Patagonia Argentina)



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ABSTRACT

Allele frequencies and forensic parameters for 22 autosomal STR loci and DYS391 locus included in the PowerPlex[®] Fusion System kit were estimated in a sample of 770 unrelated individuals from Chubut Province, southern Patagonia. No significant deviations from Hardy–Weinberg equilibrium were observed after Bonferroni's correction. The combined power of discrimination and the combined probability of exclusion were >0.999999 and 0.999984, respectively. Comparisons with other worldwide populations were performed. The MDS obtained show a close biological relation between Chubut and Chile. The estimated interethnic admixture supports a high Native American contribution (46%) in the population sample of Chubut. These results enlarge the Argentine databases of autosomal STR and would provide a valuable contribution for identification tests and population genetic studies.

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Dear Editor,

Argentinian population is the result of the confluence of different geographical groups, mainly of Native Americans, Europeans and Sub-Saharan Africans. It has been demonstrated that the ancestral proportion of this admixture process in the current population is not homogeneously distributed across the country [1,2].

Chubut province is located in South Patagonia (Supplementary Fig. 1). It has only 509,108 inhabitants in an extension of 224,686 km² (population density 2.26 inhabitants/km²) [3]. The urban population is the result of the admixture between European people, who settled at the end of the nineteenth century, the local native communities (Mapuche and Tehuelche), and subsequent migrations from other provinces and bordering countries [4–6].

The PowerPlex[®] Fusion System (Promega Corp, USA) is a STR multiplex that includes 22 autosomal STRs, (D3S1358, D1S1656, D2S441, D10S1248, D13S317, Penta E, D16S539, D18S51, D2S1338, CSF1PO, Penta D, TH01, vWA, D21S11, D7S820, D5S818, TPOX, D8S1179, D12S391, D19S433, FGA, and D22S1045) plus amelogenin for gender determination and DYS391 male specific marker.

A population sample of 770 unrelated individuals from Chubut province was analyzed to provide useful information of population genetics for new markers included in the PowerPlex[®] Fusion System. Blood samples and buccal swabs were collected from routine paternity testing after informed consent. DNA was directly amplified using the PunchSolution[™] and SwabSolution[™] kits (Promega Corp, USA). Multiplex PCR amplification was performed using PowerPlex[®] Fusion System (Promega Corp., USA) following manufacturer instructions. The amplified products were genotyped with the ABI Prism 3130 DNA Genetic Analyzer along with Gene-Mapper ID 3.2.1 software (Applied Biosystems, Foster City, CA). Allele

frequencies, heterozygosities, exact test for Hardy–Weinberg (HW) equilibrium, and population differentiation tests were performed using the Arlequin version 3.5 [7]. The forensic statistical parameters were calculated using PowerStats v.1.2 software (Promega Corp.) [8]. An interethnic admixture analysis was performed for 13 STR loci (D3S1358, D13S317, D16S539, D18S51, CSF1PO, TH01, vWA, D21S11, D7S820, D5S818, TPOX, D8S1179, FGA), using ADMIX program based on the gene identity method of Chakraborty [9,10]. Parental populations were selected following the three-hybrid admixture model for Latin America: Amerindian of Patagonia (Mapuche and Tehuelche) [11]; European (Italy and Spain) [12,13], and Sub-Saharan (Angola and Mozambique) [14,15].

Additionally, frequency distribution for three loci included in the PowerPlex[®] Fusion System (D2S441, D10S1248, and D22S1045) not previously studied in Patagonia region, were compared based on pairwise tests with twelve worldwide populations [12,13,16–23]. Multidimensional scaling (MDS) plots were performed based on the Nei's distances matrix using the R software [24].

Allele frequencies and statistical parameters of forensic interest for the 22 autosomal STR loci and DYS391 locus are shown in Supplementary Table 1. No significant departures from HW equilibrium were observed after applying Bonferroni correction for the analyzed loci ($p > 0.05/22 = 0.0023$). The most informative system was Penta E, which showed the highest average power of discrimination (PD = 0.9846) and typical paternity index (TPI = 5.52). Meanwhile, D22S1045 marker showed the lowest values for all analyzed parameters (Supplementary Table 1). The combined power of discrimination and the combined probability of exclusion for the 22 autosomal STR loci were >0.999999 and 0.999984, respectively.

The admixture contributions estimated in the sample of Chubut were 0.5010 s.e ± 0.0099 European, 0.4590 s.e ± 0.0004 Amerindian

and 0.0390 s.e \pm 0.0099 Sub-Saharan. These results are consistent with previous studies for blood systems and mitochondrial lineages [4–6], autosomal STRs [11] and Ancestry Informative Markers (AIMs) [1], where Native American contribution is higher in the South Region of Argentina. This could be related to the late incorporation of the Patagonian region to the National State in the late nineteenth century, which allowed the aboriginal communities to preserve their autonomy for a longer period. The high Amerindian composition in Chubut could also be attributed to the migratory flow from bordering countries, mainly from Chile, which preserves a strong Native American root [3].

The population comparisons of markers D2S441, D10S1248 and D22S1045, showed significant differences between Chubut and ten populations from America, Europe, Asia and Africa (Supplementary Table 2). Meanwhile, no significant differences were observed between Chubut and Chile samples for these three markers ($p=0.168-0.873$), and no differences were observed for loci D2S441 ($p=0.051$), and D10S1248 ($p=0.085$) between Chubut and the Central Region of Argentina. The resulting MDS plot based on Nei's genetic distance matrix, showed Chubut and Chile in a single cluster, close to the Central Argentine Region (Supplementary Fig. 2). As well, an additional analysis, based on 15 STRs loci (D3S1358, D1S1656, D2S441, D10S1248, D16S539, D18S51, D2S1338, TH01, VWA, D21S11, D8S1179, D12S391, D19S433, FGA, D22S1045), showed a similar spatial distribution of the populations, where Chubut and Chile constitute the same biological affinity cluster (Supplementary Fig. 3).

In conclusion, this is the first study using the PowerPlex[®] Fusion System kit in Argentina. Our results add complementary information to that existing for the PowerPlex[®] 21 System [25], provide new valuable data of forensic interest for D2S441, D10S1248, and D22S1045 loci, and constitute a useful tool for population genetics studies. Finally, this new information, along with the admixture analysis, shows that the studies of the genetic constitution of urban populations in Argentina should not be addressed without accounting for regional particularities.

This paper follows the guidelines for publication of population data requested by the journal [26]. The quality of the genetic profiles was guaranteed through proficiency testing of the GHEP-ISFG WG (Spanish-Portuguese Speaking Working Group of International Society for Forensic Genetics) (<http://www.gep-isfg.org>) and the proficiency testing of the Argentine Society for Forensic Genetics (SAGF) (<http://www.sagf.org.ar>) which are regularly carried out.

Appendix A. Supplementary data

Supplementary data associated with this article can be found, in the online version, at <http://dx.doi.org/10.1016/j.fsigen.2015.07.020>.

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María Laura Parolin*

Luciano E. Real

Liza B. Martinazzo

Néstor G. Basso

Laboratorio de Identificación Genética (IDEGEN), Instituto de Diversidad y Evolución Austral (IDEAus-CONICET), Bvd. Brown, 2915 Puerto Madryn, Argentina

* Corresponding author.

E-mail address: parolin@cenpat-conicet.gob.ar (M. Parolin).

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