



## Letter to the Editor

**Genetic variation of 15 autosomal STR loci in various populations from southern Africa**

Dear Editor,

We determined the allele frequencies for the 15 autosomal STR loci included in the AmpF/STR<sup>®</sup> Identifiler<sup>™</sup> kit in 324 unrelated individuals from southern African, representing San, Khoe, Coloured and Bantu-speaking populations. All individuals in the study originated from southern African countries including South Africa, Botswana, Namibia and Angola (Supplementary Table 1 and Supplementary Figure 1). The individuals represented different sub-groups of the San, Khoe, Coloured, southeastern Bantu-speaking and southwestern Bantu-speaking populations (Supplementary Table 1).

Informed consent was received from all participants and the study was approved by the Human Research Ethics Committee at the University of the Witwatersrand [Protocol numbers M050902 (CMS) and M980553 (HS)] as well as the Working Group of Indigenous Minorities in Southern Africa (WIMSA) and the South African San Council (SASI). DNA was extracted from 10 ml EDTA-blood using the salting-out method [1] and the PureGene<sup>®</sup> Genomic DNA Purification Kit (Gentra Systems) was used to extract DNA from buccal swabs according to the manufacturer's instructions. Extracted DNA was amplified using the AmpF/STR<sup>®</sup> Identifiler<sup>™</sup> kit (Applied Biosystems) according to manufacturer's instructions [2]. The amplified products were genotyped with the ABI 3730 XL<sup>®</sup> Genetic Analyzer (Applied Biosystems). GeneScan-600 LIZ was used as the internal lane standard and allele calling was performed using GeneMapper ID v4.0.

Relationships of pairs of individuals were inferred with Relpair v2.0.1 [3,4] and 16 related individuals were excluded from further analyses. Allele frequencies were calculated for the remaining 324 unrelated individuals. The observed and expected heterozygosity, a test of Hardy–Weinberg equilibrium and an exact test of population differentiation [5] were computed with PowerMarker v3.25 [6]. Power of discrimination and power of exclusion were calculated with PowerStats v12 [7]. ISFG recommendations on the analysis of DNA polymorphisms were followed [8] and recommended nomenclature and guidelines regarding QC and statistical issues were applied.

Allele frequencies for each of the population groups are shown in Supplementary Table 2. The *p*-values of the exact test of population differentiation between each pair of the five populations in our study and two other African populations are shown in Supplementary Table 3. The overall match probability for the 15 studied loci in the whole study group varied from 1 in  $7.3 \times 10^{11}$  in southwestern Bantu-speakers to 1 in  $6.8 \times 10^{17}$  in the Coloured group (Supplementary Table 2). The combined power of exclusion was 0.9999924 or higher for all populations in the sample group (Supplementary Table 2). Markers D13S317, D18S51 and D19S433

deviated from Hardy–Weinberg equilibrium in single populations at a significance level of 0.05. However, by applying Bonferroni correction [9], only marker D19S433 showed significant deviation in the San population (Supplementary Table 2). For several loci (between 1 and 10) and pairs of populations, the allele frequencies differed significantly ( $p < 0.05$ , Supplementary Table 3). Pairwise comparisons of allele frequencies between southeastern Bantu-speakers and other southern African groups showed many (between 7 and 10) significantly differentiated loci (except for the comparison with southwestern Bantu-speakers where only 3 loci had significantly different allele frequencies). The pairwise allele frequency comparison of Coloured and San populations also had 9 significantly differentiated loci. The mean number of differing loci across the pairwise comparisons among southern African populations was 4.7 loci. When comparing the allele frequencies of each of the southern African study populations to allele frequencies in either a Somali [10] or an Ugandan [11] population (Supplementary Table 3), the mean number of loci (across pairwise comparisons) that showed significant difference was greater for these comparisons (11.4 and 8.2 respectively) than for pairwise comparisons among the southern African populations. The southwestern Bantu-speakers have the smallest number of differing loci when compared with the other two African populations, however, the small sample size of the southwestern Bantu-speakers might affect the comparison. The observations and trends described above for the pairwise comparisons pertain when using the Bonferroni corrected significance level (Supplementary Table 3).

This paper follows the ISFG and EDNAP guidelines and practices [12–15].

**Conflict of interest**

None.

**Acknowledgements**

We are grateful to all subjects who participated in our research. CMS and HS were involved in sample collection and were supported during collection by grants awarded to HS from the South African Medical Research Council and the National Research Foundation; HS and CS from the National Health Laboratory Service Research Trust; and CS was supported by the National Research Foundation. STR typing was funded by a grant from the Magn Bergvall foundation to MJ.

**Appendix A. Supplementary data**

Supplementary data associated with this article can be found, in the online version, at [doi:10.1016/j.fsigen.2010.12.013](https://doi.org/10.1016/j.fsigen.2010.12.013).

## References

- [1] S.A. Miller, D.D. Dykes, H.F. Polesky, A simple salting out procedure for extracting DNA from human nucleated cells, *Nucleic Acids Res.* 16 (1988) 1215.
- [2] Applied Biosystems, AmpFISTR Identifier™ PCR Amplification Kit User's Manual, 2010.
- [3] M. Boehnke, N.J. Cox, Accurate inference of relationships in sib-pair linkage studies, *Am. J. Human Genet.* 61 (1997) 423–429.
- [4] M.P. Epstein, W.L. Duren, M. Boehnke, Improved inference of relationship for pairs of individuals, *Am. J. Human Genet.* 67 (2000) 1219–1231.
- [5] M. Raymond, F. Rousset, An exact test for population differentiation, *Evol. Int. J. Org. Evol.* 49 (1995) 1280–1283.
- [6] K. Liu, S.V. Muse, PowerMarker: an integrated analysis environment for genetic marker analysis, *Bioinformatics* 21 (2005) 2128–2129.
- [7] Promega, PowerStats version 12 Promega corporation website: <http://www.promega.com/geneticidtools/powerstats/>.
- [8] P.M. Schneider, Scientific standards for studies in forensic genetics, *Forensic Sci. Int. Genet.* 165 (2007) 238–243.
- [9] C.E. Bonferroni, Pubblicazioni del R Istituto Superiore di Scienze Economiche e Commerciali di Firenze, *Teoria statistica delle classi e calcolo delle probabilita* 8 (1936) 3–62.
- [10] A.O. Tillmar, G. Backstrom, K. Montelius, Genetic variation of 15 autosomal STR loci in a Somali population, *Forensic Sci. Int. Genet.* 4 (2009) e19–20.
- [11] V. Gomes, P. Sanchez-Diz, C. Alves, I. Gomes, A. Amorim, A. Carracedo, L. Gusmao, Population data defined by 15 autosomal STR loci in Karamoja population (Uganda) using AmpF/STR Identifier kit, *Forensic Sci. Int. Genet.* 3 (2009) e55–58.
- [12] A. Carracedo, J.M. Butler, L. Gusmao, W. Parson, L. Roewer, P.M. Schneider, Publication of population data for forensic purposes, *Forensic Sci. Int. Genet.* 4 (2010) 145–147.
- [13] W. Bar, B. Brinkmann, B. Budowle, A. Carracedo, P. Gill, P. Lincoln, W. Mayr, B. Olaisen, DNA recommendations. Further report of the DNA Commission of the ISFH regarding the use of short tandem repeat systems. International Society for Forensic Haemogenetics, *Int. J. Legal Med.* 110 (1997) 175–176.
- [14] P. Gill, B. Brinkmann, E. d'Aloja, J. Andersen, W. Bar, A. Carracedo, B. Dupuy, B. Eriksen, M. Jangblad, V. Johnsson, A.D. Kloosterman, P. Lincoln, N. Morling, S. Rand, M. Sabatier, R. Scheithauer, P. Schneider, M.C. Vide, Considerations from the European DNA profiling group (EDNAP) concerning STR nomenclature, *Forensic Sci. Int. Genet.* 87 (1997) 185–192.
- [15] B. Olaisen, W. Bar, B. Brinkmann, B. Budowle, A. Carracedo, P. Gill, P. Lincoln, W.R. Mayr, S. Rand, DNA recommendations 1997 of the International Society for Forensic Genetics, *Vox Sang* 74 (1998) 61–63.

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16 August 2010