



Editorial

Update of the guidelines for the publication of genetic population data

Due to the massive number of submissions of varying quality with population genetic data we decided one year ago to raise the threshold regarding the acceptance of this type of publications to ensure a high standard of published data and, therefore, we updated the FSI: Genetics 2010 guidelines [1] to a new set of recommendations [2].

In the 2013 guidelines in addition to some new requirements in the procedure and regarding ethical standards, we significantly increased the number of markers and samples required for submission to the journal of papers presenting population data alone, with no additional information on new methods or other forensically relevant findings.

We have been working during last year with these new recommendations. During this period, we have received a number of critical comments from authors that we have considered and therefore we have decided to make some amendments in the requirements.

The first change refers to the minimal number of autosomal STRs. The 2013 guidelines indicate that for data comprising autosomal STR genotypes only, 17 different autosomal STR loci are required as a minimum. This was based on the average number of STRs that most laboratories are routinely using – in most cases forensic laboratories are using at least two PCR multiplexes (commercial kits or homemade) on their routine work for a minimal number of 17 autosomal STR markers. However, this number would exclude laboratories and national and international compilation efforts that are working with just one kit, for some of the most commonly used kits. Whilst we certainly recognize the need to restrict the number of small data sets submitted we think that the forensic community would benefit from the publication of a large and nationally important data set such as the ones that are being generated in some countries. For this reason we have decided to reinstate the 2010 recommendations requiring 15 STRs only.

Concerning X chromosome a minimum number of 12 STRs will be required and for the Y chromosome a minimum of 17 STRs will be required as well, taking into account that the core minimum haplotype (DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385) [3,4] must be provided if it has not been previously analyzed in the same population sample.

Collaborative efforts to produce large datasets are strongly encouraged and, therefore, the minimal number of markers should not be a limitation to the publication of large National or International collaborative databasing efforts when a significant number of laboratories and samples are involved.

There are also changes in the requirements for the minimum number of samples. We maintain the threshold of 500 samples for

autosomal and X-chromosomal STRs but we decrease the minimum required for Y-chromosomal and mtDNA data to 200 samples. With this reduction in sample number requirements for lineage markers, we desire to stimulate data generation for these types of genetic markers to improve understanding of potential population substructure through gathering relevant data from around the world.

Flexibility in the total sample number is still possible with small size populations of forensic interest (such as minorities and small indigenous population groups), with the editor assigned to the paper responsible to decide how exceptional the data provided is and to proceed accordingly. In this case, an effort should be made to include different type of markers for the same samples, namely, mtDNA sequences and autosomal, Y-chromosomal and/or X-chromosomal STRs, SNPs and/or InDel markers.

We would also like to stress that scientists are requested to provide minimum statistics, haplogroup assignments and comparison to “relevant” neighbors (already included in earlier guidelines) to provide the reader with information that is practical to a forensic case. With this additional information, these papers become more attractive for population and medical genetic researchers.

The requirements outlined here and in the previous recommendations are applied to those papers that are simply presenting population data – just a description of a database, and the calculation of diversity indices and population comparison analyses.

We strongly encourage authors to increase the quality of the data by adding other forensically relevant information (like mutation rates, recombination rates, case reports, etc...) or combining markers and populations together representing a serious population genetic approach that could make the study worthy of publication as a full paper in our journal.

References

- [1] A. Carracedo, J.M. Butler, L. Gusmão, W. Parson, L. Roewer, P.M. Schneider, Publication of population data for forensic purposes, *Forensic Sci. Int. Genet.* 4 (2010) 145–147.
- [2] A. Carracedo, J.M. Butler, L. Gusmão, A. Linacre, W. Parson, L. Roewer, P.M. Schneider, New guidelines for the publication of genetic population data, *Forensic Sci. Int. Genet.* 7 (2013) 217–220.
- [3] M. Kayser, A. Caglia, D. Corach, N. Fretwell, C. Gehrige, G. Graziosi, F. Heidorn, S. Herrmann, B. Herzog, M. Hidding, K. Honda, M. Jobling, M. Krawczak, K. Leim, S. Meuser, E. Meyer, W. Oesterreich, A. Pandya, W. Parson, G. Penacino, A. Perez Lezaun, A. Piccinini, M. Prinz, C. Schmitt, L. Roewer, Evaluation of Y-chromosomal STRs: a multicenter study, *Int. J. Legal Med.* 110 (1997) 125–133.
- [4] V.L. Pascali, M. Dobosz, B. Brinkmann, Coordinating Y-chromosomal STR research for the Courts, *Int. J. Legal Med.* 112 (1999) 1.

Angel Carracedo^{a,b,*}

^a*Institute of Forensic Science, Genomic Medicine Group,
University of Santiago de Compostela, Spain*

^b*Center of Excellence in Genomic Medicine Research,
King Abdulaziz University Jeddah, Saudi Arabia*

John M. Butler

*National Institute of Standards and Technology,
Gaithersburg, MD, USA*

Leonor Gusmão^{a,b}

^a*Institute of Pathology and Molecular Immunology of the
University of Porto (IPATIMUP), Portugal*

^b*DNA Diagnostic Laboratory (LDD),
State University of Rio de Janeiro (UERJ), Brazil*

Adrian Linacre

*School of Biological Sciences, Flinders University,
Adelaide, Australia*

Walther Parson^{a,b}

^a*Institute of Legal Medicine,
Innsbruck Medical University, Austria*

^b*Penn State Eberly College of Science,
University Park, PA, USA*

Lutz Roewer

*Institute of Legal Medicine, Charité-Universitätsmedizin,
Berlin, Germany*

Peter M. Schneider

*Institute of Legal Medicine, Medical Faculty,
University of Cologne, Germany*

*Corresponding author at: Institute of Forensic Science,
Genomic Medicine Group,
University of Santiago de Compostela,
Spain

E-mail address: angel.carracedo@usc.es (A. Carracedo).