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Editorial

Publication of population data for forensic purposes

In 2000 a new policy concerning the publication of population genetic data was set up in *Forensic Science International* [1] with the introduction of a new section entitled "Announcement of population data". The idea was to facilitate the publication of this type of data since the use of reliable allele or haplotype frequency estimates of the polymorphisms is a requirement in most countries, both in forensic and in paternity cases.

Announcements of population data consisted in short communications under a fixed format, avoiding the repetition of superfluous information (i.e., materials and methods) and concentrating the message on the key information needed for the use of genetic data for forensic and population genetics.

In our opinion, this type of paper completely fulfilled the aims of the editors and, even more importantly, has made an essential contribution to the dissemination of common standards all over the world. In addition they have motivated forensic practitioners (especially in countries with little development in forensic genetics) to introduce themselves in forensic research.

With the launch of the new journal, we decided to continue the same policy as a first step but keeping in mind that we have to move forward to increase the quality of the journal and to avoid having a journal exclusively devoted to announcements of population genetic data.

The number of population genetic papers from the very beginning has continuously increased, representing now more than 60% of the submissions to the journal. Therefore, it is time to raise the threshold regarding the acceptance of this type of publication but taking into account the importance of the dissemination of standards and the motivation that this type of research represents for some groups and countries.

For this reason, we have decided to move to a next step and to introduce a new section on Forensic Population Genetics in the journal.

Manuscripts with population genetic content can be submitted to this section at http://www.ees.elsevier.com/fsigen/ using three types of formats:

Forensic Population Genetics – Original papers: in this section full length papers on relevant population genetics issues of forensic interest will be considered for publication. The data should be original, the population genetic analysis must be of the highest quality and the data should have forensic relevance beyond the scope of simply reporting allele or haplotype frequencies.

Forensic Population Genetics – Short communications: understanding that both the quality of population data and the relevance of results are crucial, short communications will have the format of the former "Announcements of population data" with some changes (see below) in order to guarantee their quality.

Forensic Population Genetics – Letters to the Editor: if the relevance of the data is not sufficient for an original paper or a short communication, but still worthy of an announcement, the editors can invite authors to submit a letter to the editor. In this case the manuscript must be written in the form of a short letter to the editor summarizing the relevant information while the frequency data must be provided as an electronic supplement, e.g. a spreadsheet table, for online publication in the electronic repository of the journal.

The full-text of the original research papers in the forensic population genetics section will be published in print. Short communications and letters to the editor will be published in the electronic version of the journal only. For all forensic population genetics papers the actual population data are published in table format as electronic supplementary data only.

Population genetic data papers should always contain the following information:

1. Description of the population:

   With a variable length depending on the type of paper, a detailed description of the population is essential as well as a description of the interest of that population for population genetics and forensic purposes. Previous population genetic studies should be reported as well as the geographic location, ethnicity, method of sampling, and characteristics of the population.
2. Ethical requirements:

   Informed consent and/or specific approval of a recognized ethical committee are required and must be stated in the text. For STRs, the inclusion of the whole genotyping data will not be required due to ethical constraints for the publication of such types of data in some countries but the authors are requested to anonymize the data and to provide the anonymized data to interested researchers upon request if not prohibited by ethical constraints. The authors should state in the text that they understand and accept the requirements requested in this editorial.
3. Quality control:

   For STRs and SNPs the quality of the data must be guaranteed. The QC procedures followed by the authors must be specified. Certification of approval by proficiency testing programs is ideal and encouraged. Authors must state that they have strictly followed ISFG recommendations on the analysis of the DNA polymorphisms used [2], signifying the use of recommended nomenclature and guidelines regarding QC and statistical issues.

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4. mtDNA and Y chromosome polymorphisms:

MtDNA and Y chromosome data need special requirements. In this case the importance of high quality population DNA databases justifies a strict publication policy as follows:

4.1 mtDNA:

The executive board of the International Society for Forensic Genetics (ISFG) and the editors of Forensic Science International: genetics have invited EMPOP[1] to logistically organize and perform quality control (QC) of mtDNA sequences in the course of manuscript preparations for the journal *Forensic Science International: Genetics*. Before mtDNA papers are put forward to the editors for review, the authors are required to submit the data to EMPOP. After evaluation, the authors will be contacted by EMPOP, and the mtDNA sequences will be assigned EMPOP accession numbers that serve as indicators of successful QC for the editors and reviewers. The necessary steps for submission of mtDNA sequences to EMPOP are outlined below.

General comment: the presentation of partial control region sequences, such as those of the hypervariable segments (HVS) I and II only, is not any longer state of the art. Instead the authors should aim at the analysis of entire control region sequences [3], which increases the discrimination power of mtDNA testing in forensics and adds valuable information to the phylogenetic interpretation of mtDNA haplotypes. In compliance with earlier recommendations [4,5] the minimum requirement for acceptable data is full double-stranded sequence coverage.

- Step 1:

  Prepare your sequence data as shown in the EMPOP tutorial (see download section, www.empop.org). Additionally, an example can be downloaded there that can also be used as a template for the preparation of the data. In brief, the emp file is a "tab delimited" text file that can be created using a standard text editor or a spreadsheet software (then, save the file under .txt format and rename ".txt" to ".emp"). The first two lines specify individual details of the dataset and origin of the samples. Further lines list the individual mtDNA haplotypes headed by their sequence range(s). Note that a given sequence range is applied to all mtDNA haplotypes following this range until a new range is defined. Text lines, e.g. for individual comments, need to be identified using the "#" symbol. Please consult EMPOP for further details on data preparation.

- Step 2:

  Submit your file(s) to EMPOP using the e-mail address "data-submission@empop.org". The data will be quality-checked for format, plausibility, clerical errors, sequence range violation, reference errors, indels designation, and phantom mutations when using in-house softwares and NETWORK, which is also available through EMPOP. Note that tools for sequence data evaluation are continuously added to the EMPOP website to help the authors scrutinize their data before submission.

- Step 3:

  Communication may follow with respect to individual sequences. Once your data have passed QC you will receive the emp file of your data listing EMPOP accession numbers. Please provide these accession numbers together with your manuscript to the editor for initiating the review process.

- Step 4:

  Upon publication your data will be uploaded onto EMPOP.

  Important: the fact that sequence data quality is scrutinized by EMPOP should not relieve the authors from carefully inspecting their dataset. These data review should not be limited to the announced errors of the EMPOP QC but requests the authors to review the whole dataset. Additional errors in a posterior submissions will represent a serious drawback for the future acceptance of the paper. Quality control by EMPOP should act as a final check on the data. The EMPOP staff will be happy to provide help and guidance for the preparation of the population data.

4.2 YSTRs and YSNPs:

In the same way as for mtDNA submissions, the executive board of the ISFG and the editors of *Forensic Science International: Genetics* have invited YHRD[2] to logistically organize and perform quality control (QC) of YSTR/YSNP data in the course of manuscript preparations for the journal *Forensic Science International: Genetics*. Before YSTR/YSNP papers are put forward to the editors for review, the authors are required to submit the data to YHRD. After evaluation, the authors will be contacted by YHRD and the YSTR/YSNP data will be assigned to YHRD accession numbers that serve as the indication of successful QC for the editors and reviewers. The necessary steps for submission of YSTR/YSNP data to YHRD are outlined below.

- Step 1:

  Prepare your YSTR and YSNP data as explained at the website www.yhrd.org/contribute and in the YHRD manual (www.yhrd.org/downloads/manual.pdf). The YHRD input file is a standard spreadsheet file. The first two columns specify a sample identification number and the origin of the samples. For the latter we request a ternary identifier in the form "region, country [ethnic group]" – e.g. "Berlin, Germany [German]". The geographic background of the samples should be further detailed in an accompanying text or by maps. The other columns list the common YSTR loci and the panel of typed YSNPs, specified by "+" for the derived state and "−" for the ancestral state at a given locus. The last two columns contain the haplogroup designation according to the most updated nomenclature [6], and the final branch marker used for haplogroup assignment, e.g. Q1a3a and M3. Synonymous marker names are allowed. If the haplogroup is unknown, then use a "?" symbol. The YHRD software verifies the correct haplogroup assignment based on the "+/−" input files.

  The file should list the individual haplotypes with a single haplotype per line using unique identification numbers. Identical haplotypes should be listed separately. Please note the following format rules:
  - alleles at duplicated loci are separated by a comma (e.g. "11,14")
  - alleles containing incomplete repeat motifs are designated by a dot (e.g. 11.2)
  - confirmed "null" alleles are indicated by a "0"

  Note that allelic drop-outs at certain YSTR loci may occur due to either molecular mechanisms (e.g. chromosomal rearrangements or deletions, primer site mutations) or technical problems (e.g. low amounts of DNA template,

[1] European DNA Profiling (EDNAP) Group's mitochondrial DNA population database project; www.empop.org.

[2] Y Chromosome Haplotype Reference Database; www.yhrd.org.

degradation). As used here, the term "null allele" refers to allele loss due to molecular mechanisms. These should be reported.

- Step 2:

  The file should be sent as an e-mail attachment to the following addresses "lutz.roewer@charite.de" and "sascha.willuweit@charite.de". The text of the e-mail should contain the title of the study and an author name with an e-mail address for contact. The data will be quality-checked for format, clerical errors, allelic range violation using in-house software (e.g. NETWORK, AMOVA).
- Step 3:

  Communication may follow with respect to individual haplotypes/haplogroups. Once your data have passed QC you will receive the yhrd-file of your data listing YHRD accession numbers for all your samples. Please provide these accession numbers together with your manuscript to the editor of the journal.
- Step 4:

  Upon publication your data will be uploaded onto YHRD.

5. SNPs:

   Population data of SNPs will be considered but only for SNP sets previously validated for forensic purposes.
6. Table formats:

   Genotyping results should follow a standard spreadsheet table format and should be submitted as an electronic supplement file to be published only in the electronic repository of the journal.

   Figures and tables will be published only in the electronic repository of the journal in the case of letters to the editor, as well as in the case of short communications.
7. Authors must state in the paper that they have strictly followed the requirements of this guideline and the ISFG recommendations

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