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Short communication

Formulation and communication of evaluative forensic science expert opinion—A GHEP-ISFG contribution to the establishment of standards

António Amorim[a,b,c], Manuel Crespillo[d], Juan A. Luque[d], Lourdes Prieto[e], Oscar Garcia[f], Leonor Gusmão[a,b,g], Mercedes Aler[h], Pedro A. Barrio[i], Victor G. Saragoni[j], Nadia Pinto[a,b,k,*]

[a] Instituto de Patologia e Imunologia Molecular da Universidade do Porto (IPATIMUP), Porto, Portugal
[b] Instituto de Investigação e Inovação em Saúde (i3s), Universidade do Porto, Porto, Portugal
[c] Faculdade de Ciências, Universidade do Porto, Porto, Portugal
[d] Instituto Nacional de Toxicología y Ciencias Forenses, Departamento de Barcelona, Servicio de Biología, Ministerio de Justicia, Barcelona, Spain
[e] Instituto de Ciencias Forenses, Grupo de Medicina Xenómica, Universidade de Santiago de Compostela, Spain
[f] Forensic Science Unit, Forensic Genetics Section, Basque Country Police-Ertzaintza, Erandio, Bizkaia, Spain
[g] DNA Diagnostic Laboratory, Institute of Biology, State University of Rio de Janeiro, Rio de Janeiro, Brazil
[h] Sección de Genetica Forense y Criminalística, Instituto de Medicina Legal de Valencia, Spain
[i] Servicio de Biología, Departamento de Madrid, Instituto Nacional de Toxicología y Ciencias Forenses (INTCF), Spain
[j] Unidad de Genética Forense, Servicio Médico Legal (SML) de Santiago, Santiago, Chile
[k] CMUP Centro de Matemática da Universidade do Porto, Porto, Portugal

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ABSTRACT

Communicating and interpreting genetic evidence in the administration of justice is currently a matter of great concern, due to the theoretical and technical complexity of the evaluative reporting and large difference in expertise between forensic experts and law professionals. A large number of initiatives have been taken trying to bridge this gap, contributing to the education of both parties. Results however have not been very encouraging, as most of these initiatives try to cope globally with the problem, addressing simultaneously theoretical and technical approaches which are in a quite heterogeneous state of development and validation. In consequence, the extension and complexity of the resulting documents disheartens their study by professionals (both jurists and geneticists) and makes a consensus very hard to reach even among the genetic experts' community. Here we propose a 'back-to-basics', example-driven approach, in which a model report for the two most common situations faced by forensic laboratories is presented. We do hope that this strategy will provide a solid basis for a stepwise generalisation.

1. Introduction

The contribution of science to justice requires adequate communication between experts and the courts. This basic requirement is particularly demanding in forensic genetics, given the theoretical background required to understand the technically sophisticated results which are now currently delivered [13]. A pessimistic attitude on both sides – genetic experts and the judicial system players – has unfortunately grown in the last years, and as much as the understanding of likelihood ratios by jurists was quite poor, experts were not exempt from frequent mistakes. Indeed, almost a half of experts and jurists alike considered that adequate communication between experts and the courts is not feasible, the dialogue between them being characterized by gaps and misunderstandings, due to the large difference in professional expertise and frame of reference [10,14,25,21].

With this work we intend to counter this pessimism [3] and to present a reporting model for genetic results for the two most common situations faced by the genetic expertise: (a) are two samples from the same donor?; and (b) are two samples from a pair of individuals related as, say, father and offspring?, when the alternative is that the samples belong to unrelated individuals. These cases, the most common in both forensic and 'paternity' labs epitomize fairly well the issue at stake and provide an opportunity for future discussion and generalizations. Moreover, the explicitness of the technical bases and assumptions as well as the theoretical, statistical and probabilistic framework is mandatory, a

* Correspondence to: Nádia Pinto – IPATIMUP/I3s, Rua Alfredo Allen, 208, 4200-135 Porto, Portugal.
E-mail addresses: npinto@ipatimup.pt, nmgapinto@gmail.com (N. Pinto).

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task that we also undertake. The demand for this simple approach has been recently demonstrated by the results of the GHEP-ISFG 2015 quality control exercise in which just 18% of the participants were found to report correctly a standard paternity case [4]. We would like also to reinforce that, in both cases, the alternative hypothesis is that the samples belong to unrelated individuals and therefore the situations in which this cannot be assumed are outside of the scope of the main part of this work. For example, cases where the real donor of the sample (or the real father) can be brother, father or son of the tested man are not here considered, as well as the possibly incestuous situations where the alleged father is father, brother or son, of the mother of the child. In this way we avoid the theoretical complications related with pedigrees belonging to the same kinship class [17,18], restricting the analyses to the simplest cases among those of paternity and identity. The statistical framework is in accordance with the recommendations of the ISFG on biostatistics for paternity testing [11], assuming a 'classical' formulation of likelihood ratio in which two conditional probabilities are compared [8].

While this work was in progress under the stimulus of the GHEP-ISFG, the results of a similar initiative were reported by ENFSI ("ENFSI Guideline for Evaluative Reporting in Forensic Science", 2015. http://enfsi.eu/sites/default/files/documents/external_publications/m1_guideline.pdf, accessed 5th January 2016). Although the aims of both partially overlap («to standardise and improve evaluative reporting»), the means to achieve this goal are distinct. Indeed, we present here a model report for two categories of typical routine cases in any forensic lab, believing that the strategy for reaching a consensus focused on simple examples would be more fruitful to the community instead of trying to issue general standards or guidelines. Other previous efforts (e.g., [5]) also proved to be either too broad and/or being far from acceptable by a good number of experts [15,16,22,24].

With this simple example driven strategy, we avoided the still controversial issues at both technical and mathematical levels (e.g., low level DNA, mixtures), paving the way to build up a basis for future generalisation to more complex situations. Again for simplicity sake, evidence from non-recombining uniparentally transmitted haplotypes will not be included [2,7]. Moreover, since X-chromosome markers treatment is of much higher complexity, due to linkage and association [26], only unlinked, unassociated, autosomal loci will be considered. To keep the simplicity of the examples but also due to the state of art, all the individuals are assumed to belong to the same population. Considerations on the statistical treatment of mutations and silent alleles will not be made since there is no agreement among experts on how to deal with such complexities and its inclusion would compromise the 'basic' formulation we intended for this work.

This work assumedly does not address the grand question of communicating and interpreting statistical evidence in the administration of justice; this task of building up a system of responsible producers (experts) along with discerning consumers (judges, lawyers, prosecutors) is societal, but also an individual duty for every professional participant (for two excellent examples of publicly available results on this, Aitken et al. [1], Reference Manual on Scientific Evidence [23]).

A draft version of these model reports were previously distributed to the members of the GHEP-ISFG where it deserved general agreement and has benefited from various comments and suggestions.

A supplementary extension of this work is also presented, approaching kinship cases where, after a first report of identity/paternity was issued, an alternative hypothesis of kinship emerges. It is considered as motivating example a case where a paternity report primarily issued (and favoring paternity) is returned with the new information that the tested putative father refuses paternity, claiming that the real father is in fact his meanwhile deceased full brother – see Appendix 2 in Supplementary file.

2. Technical bases and assumptions

Samples are considered to have been collected under protocols of identification and chain of custody, leaving no doubts on their genuine origin, absence of contamination and mislabeling, assuming no human errors in handling, from collection to results output.

Genetic analyses on all involved samples are the same, both in terms of techniques employed and data analyses (or validated comparability) and results provide profiles with no evidence of mixture and/or drop-out/drop-in events.

3. Theoretical, statistical and probabilistic framework

In this context, all the markers used are autosomal and unlinked, and adequacy to the formal genetic model has been tested in a reasonably sized sample of meiotic transmissions. Conformity to Hardy-Weinberg expectations was also tested, as well as the absence of association between markers. Reliable estimates of allele frequencies (including silent alleles) and mutation rates are available.

The calculations involved in the report are as follows:

1 The probability of the observations (genetic profiles) under a specific hypothesis
2 The probability of the same observations (genetic profiles) under an alternative, mutually exclusive hypothesis [6]
3 A likelihood ratio between the probabilities 1. and 2.

In this framework, when we say 'unrelated' it means that the individuals are drawn at random from the population, so that the probability of being genetically closely related (i.e., linked by a close kinship relationship) is practically nil.

It is also assumed that a proper authority formally requests the evaluating report and that a hypothesis, even if in a somewhat implicit form, is put forward. In any case, the explicit and clear formulation of the alternative propositions used for the case by the expert is mandatory (see below).

4. Model report conclusions

4.1. Identity

In this case, it is further supposed that two genetic profiles match exactly for all markers examined. Under the assumptions above, the general alternative propositions corresponding to the conditional probabilities 3.1. and 3.2. above take the form:

a) The two profiles correspond to the same donor;
b) The two profiles correspond to different, genetically unrelated, donors;

and the likelihood ratio can be verbalized as:

Observing the same genetic profile in both samples is x times more likely assuming they came from the same donor than resulting from a pair of unrelated donors.

4.2. Paternity

Here, two types of situation are possible: a putative father/child duo or a trio (the mother of the child is included in the analyses).

4.2.1. Motherless case

Profiles for the putative father and offspring are obtained and the alternative propositions corresponding to the general conditional probabilities 3.1. and 3.2. take the form:

a) The profiles correspond to a pair of individuals genetically related as father/child;
b) The profiles correspond to a pair of genetically unrelated individuals;

And the likelihood ratio can be phrased as:

The observed genetic profiles are x times more likely assuming that the individual is the biological father than considering he is genetically unrelated to the child.

4.2.2. Trio case

Profiles for the putative father, undoubted mother and offspring are obtained and the alternative propositions corresponding to the general conditional probabilities 3.1. and 3.2. take the form:

a) The profiles correspond to individuals related as mother/father/child;
b) The profiles correspond to a trio where the alleged father is an individual genetically unrelated with the unquestioned duo mother/child;

And the likelihood ratio can be described as:

The observed genetic profiles are x times more likely assuming that the individual is the biological father than considering he is genetically unrelated to the mother/child pair.

5. Model report example

In order to allow the global assessment evaluation and criticism of the proposed model, a mock report for the case putative father/child duo is shown in the Appendix 1 in Supplementary file, which should be adapted to local legislation and specific procedures of each laboratory.

6. Discussion and conclusions

We hope to have provided at least an objective basis for an open discussion on this key issue of forensic practice – the communication with the courts and laypersons in general. This need has been widely recognized in particular in quality controls and collaborative exercises [9,19,20], but few initiatives have been taken to ensure a substantive progress in the production of a standardized reporting framework, even for routine, simple and typified situations like those here presented, and recommendations issued by various scientific and professional bodies are too general to prevent the current unclear situation.

By providing an example driven approach we hope to contribute for an objective basis to establish a standard of communication for the most simple and frequent types of situation where an evaluative report on genetic evidence is requested. We trust that the establishment of such a standard will allow and encourage further developments to more complex cases and situations. Moreover, the attainment of a consensus on admittedly simple cases (by far the most frequent) will counteract a recent wave of mistrust on DNA evidence caused by unfortunate mishaps, highly impacted in the media (e.g. [12,27]), relaunching confidence among and between practitioners and laymen.

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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.2016.09.003.

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