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DNA commission of the International society for forensic genetics: Assessing the value of forensic biological evidence - Guidelines highlighting the importance of propositions
Part I: evaluation of DNA profiling comparisons given (sub-) source propositions

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A B S T R A C T

The interpretation of evidence continues to be one of the biggest challenges facing the forensic community. This is the first of two papers intended to provide advice on difficult aspects of evaluation and in particular on the formulation of propositions. The scientist has a dual role: investigator (crime-focused), where often there is no suspect available and a database search may be required; evaluator (suspect-focused), where the strength of evidence is assessed in the context of the case. In investigative mode, generally the aim is to produce leads regarding the source of the DNA. Sub-source level propositions will be adequate to help identify potential suspects who can be further investigated by the authorities. Once in evaluative mode, given the defence version of events of the person of interest, it may become necessary to consider alternatives that go beyond the source of the DNA (i.e., to consider activity level propositions). In the evaluation phase, it is crucial that formulation of propositions allows the assessment of all the results that will help with the issue at hand. Propositions should therefore be precise (indication of the number of contributors, information on the relevant population etc.), be about causes, not effects (e.g. a 'matching' DNA profile) and to avoid bias, must not be findings-led. This means that ideally, propositions should be decided based on the case information and before the results of the comparisons are known. This paper primarily reflects upon what has been coined as "sub-source level propositions". These are restricted to the evaluation of the DNA profiles themselves, and help answer the issue regarding the source of the DNA. It is to be emphasised that likelihood ratios given sub-source level propositions cannot be carried over to a different level – for example, activity level propositions, where the DNA evidence is put into the context of the alleged activities. This would be highly misleading and could give rise to miscarriages of justice; this will be discussed in a second paper.

The value of forensic results depends not only on propositions, but also on the type of results (e.g. allelic designations, peak heights, replicates) and upon the

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model used: it is therefore important to discuss these aspects. Finally, since communication is key to help understanding by courts, we will explore how to convey the value of the results and explain the importance of avoiding the practice of transposing the conditional.

1. Introduction

The ISFG DNA commission has previously published recommendations on how to assign value to a comparison between DNA profiles [1,2]. With the advent of more sensitive methods of DNA detection, it is possible to analyse minute quantities of trace-material. This has brought new challenges to the evaluation of DNA results (i.e. mixtures) and of biological results in general (i.e. biological origin of the DNA, transfer and persistence phenomena). The formulation of propositions[2] in such cases can be less straightforward, than it would be for single DNA profiles derived for example from a large amount of blood. As outlined in several publications [4–9], formulation of propositions is essential, because the value of the results will depend on propositions utilized. A further aspect relates to the meaning of DNA in the context of alleged activities. Whereas there has been considerable uptake of methods used to analyse the strength of evidence of a DNA profile, there is a paucity of advice related to the meaning of the evidence in relation to the alleged 'activities'. Consequently, the DNA commission will produce advice to assist forensic geneticists to evaluate DNA and biological results whose value is impacted by phenomena such as secondary (or tertiary) transfer, contamination or 'fortuitous' presence of DNA in the environment.

The guidelines are divided into two main papers: first we describe the recommendations that have been made regarding the value of compared profiles given (sub-) source level propositions and recall main principles. We provide *recommendations* - proposals as to the best course of action- along with *considerations*- suggestions that require careful thought in relation to the specifics of a given case (it may not be so easy to generalise a best course of action as with a recommendation). The advice is updated to take account of recent developments, particularly with the introduction of probabilistic models and the evaluation of mixtures. We discuss the importance to distinguish the comparison of profiles and the value of this comparison in the 'investigation' phase (when there is no person of interest yet) and in the context of 'evaluation' (when a person of interest has been identified, who may or may not dispute the results). We present guidelines regarding the formulation of propositions, taking into consideration the literature and the England and Wales forensic science regulator's provisional guidance on DNA mixture interpretation [10]. Indeed, this document provides essential advice on formulation of propositions in the context of mixtures. We will discuss the formulation of propositions given 'sub-source' level and the importance of not blending results with propositions.

As the value of results depends not only on propositions (level in the hierarchy, population considered, relatives, number of contributors), but also on the type of results considered (presence/absence of allelic peaks, peak heights, use of replicates) and the assumptions of the model and data used, we also discuss these aspects. We conclude with a section to describe how the value of DNA results can be (mis)understood, by emphasising the fact that the probability of the results given propositions is not the same thing as the probability of the propositions given the results. We exemplify this difference with the so-called 'database problem'.

In the second paper: "DNA Commission of the International Society for Forensic Genetics: Assessing the value of forensic biological evidence - guidelines highlighting the importance of propositions" (in preparation), we will provide considerations and recommendations in the field of evaluation of forensic biological results in the context of the activities alleged in the case. We will give examples and present Bayesian networks as a means to deal with the complexity of real casework more comprehensively and in a transparent way. Methods of analysis are a well-researched and documented area, however, the *evaluation* of the results obtained from using these methods is often less well formalised. We prefer an experimental approach to enable assignment of probabilities in the context of transfer evidence. There may be a perception that such probabilities are less data-supported than probabilities derived for assessing the value of the comparison of DNA profiles. But, this does not mean that experts can solely rely on experience acquired in casework. Consequently, it is important to ensure that methods of evaluation are as robust as methods of analysis.

2. Evaluation of DNA profiling comparisons: context

In this paper, we will outline an existing framework (i.e. the likelihood ratio approach) that allows the assessment of forensic results whatever the type of case (single trace, mixtures, low-template, paternity cases, missing persons etc.). To assess the value of a DNA profile, the first aspect to consider is whether the profile has sufficient information to be used in casework. Depending on the complexity, a decision will be made whether or not to compare the profile to the person(s) of interest (POI). In situations where there is no suspect, the scientist will act as investigator. If there is a person of interest, one will usually act as evaluator [11]. Depending upon the scientist's role, he/she will be asked different questions and will therefore give different answers, it is thus important to identify the issue and determine how forensic analysis can help the formulation of propositions and the choice of analytical methods.

2.1. Investigative versus evaluative reporting

As mentioned above, forensic DNA scientists have a dual role: they are asked both to provide investigation leads and to provide the value of a comparison in the context of a case. It is crucial to distinguish between these two roles [11], in particular with reference to the propositions used.

One can use likelihood ratios in both the investigative and the evaluative phase, the main difference is that in the evaluation phase, there will be a suspect/defendant. In this situation it will be necessary to account for the defence's view of events. The scientist operates in 'investigator mode' in the initial stages of a case. A typical example is where a database search is carried out, because there is no suspect associated with the crime-scene. Here, what is of interest is to provide investigation leads, by giving information about who could be the source of the DNA. Therefore, what we call sub-source propositions (i.e. propositions regarding the source of the DNA) are fit for purpose. In a database with N individuals, each individual $X_{i=1..N}$ is compared with the crime-stain in turn. *Before* the comparison is carried out, all individuals in the database may be considered to be possible candidates. The laboratory may also be given possible candidates following a research into a national DNA database using CODIS, for example.

In a case of a single stain where there is no person of interest yet, sub-source level propositions could be:

- The DNA is from the candidate X_i
- The DNA is from some unknown individual

[2] From the Oxford English Dictionary: A proposition is a statement that expresses a concept that can be true or false. Some people use the term hypothesis. Here, as described by [[3]] we will use the term proposition as this helps reduce the risk of confusion between evaluation and hypothesis testing which may be associated with the term 'hypothesis'.

Either candidate X_i gives a high LR (LR > > 1)[3] so that he/she will be forwarded for further investigation, else a low LR (LR < 1) is achieved in which case he/she may be eliminated from the investigation and is no longer considered a possible candidate. If the LR is very high (in the order of 10^9 for a large database of several million[4]), typically, either one person remains after the search and comparison, or none is discovered. Alternatively, in the case of partial DNA profiles which give intermediate LRs > 1 there may be several candidates found.

To reiterate, at this stage, there is no defendant, and the scientist is working as an investigator. The prosecuting authorities will be notified about the potential candidates and they decide if he/she subsequently becomes a POI. Further investigation will follow (interviews, witness information, searches of premises etc.) where non-DNA evidence will be gathered and where the POI's account of the facts will be asked. Once a suspect is identified the scientist switches to evaluative mode. If there is sufficient combined evidence, then it may be decided to prosecute him, and the person consequently becomes a defendant.

Consideration 1

The scientist works in an investigative mode if there is no person of interest in the case. If a suspect is identified, then generally the scientist switches to evaluative mode with respect to this suspect and needs to assign the value of their results in the context of the case. If there is new information (in particular from the POI), the scientist will need to re-evaluate the results. It is thus important that reports contain a caveat relating to this aspect.

2.2. When is evaluative reporting appropriate?

Once the POI has been identified, prosecuting authorities investigate the case further: non-DNA evidence will be considered and information regarding the circumstances of the case (in particular the POI's account of events) will be gathered. The scientist then operates in 'evaluative' mode and the principles of interpretation apply [12]. As a model, we refer the reader to the ENFSI guidelines for evaluative reporting [13] where the conditions under which evaluative reporting must take place are described as follows:

> *"Evaluative reports for use in court should be produced when two conditions are met:*
> *1. The forensic practitioner has been asked by a mandating authority or party to examine and/or compare material (typically recovered trace material with reference material from known potential sources).*
> *2. The forensic practitioner seeks to evaluate results with respect to particular competing propositions set by the specific case circumstances or as indicated by the mandating authority."*

In the adversarial system of justice the court acts as an impartial referee between prosecution and defence. The scientist can only act in an evaluative role - the investigator role is purely pre-trial. The inquisitorial system differs in that the court takes an active part in the investigation of an offence [14]. Consequently, the scientist may operate in both investigative and evaluative modes depending upon the questions put by the judge. In the adversarial system the judge's role is passive (neutral) whereas in the inquisitorial system the judge controls the search for evidence and questions the witnesses, playing a central role to determine the truth. He/she is not impartial, and can be regarded as the mandating authority described above. In all cases, the over-riding duty of the scientist is to be neutral and impartial.

3. A framework for evaluation of biological results

Several previous ISFG commissions recommended the use of the likelihood ratio (LR) as the preferred metric to assess the value of DNA results [1,2]. There is also a vast amount of forensic literature that advocates the use of LRs to evaluate findings, and explains the shortcomings of alternative methods [4,15–17]. More generally, the evaluation of forensic results should be based on three principles [12]. The first principle (or to be more precise: "precept") says that the value of the results should be considered given at least one alternative proposition. The assignment of a likelihood ratio therefore requires a pair of mutually exclusive propositions that reflect two competing positions, for example: that of the prosecution and the defence [13]. These do not need to be exhaustive, but should reflect the positions of both parties. The second precept is that the value of scientific results is dependent on the information used by the scientist. This information encapsulates the relevant case circumstances, the data used, the scientist's assumptions and the model chosen. The relevant case circumstances include only the case information that is needed for the formulation of the propositions and for assigning the probabilities of the results. An example of relevant case circumstances would be: 'The events took place in the United Kingdom'. This information will allow the scientist to select the most relevant population genetic database. An example of forensically irrelevant case circumstances (that is not needed) would be: 'The witness recognised Mr Jones as the offender'. It is not the domain of the expert to combine this 'prior' information with the DNA typing results (we refer the reader to Section 7), which leads us to the third precept, which is that the scientist is concerned only with the probability of the results given the propositions, and not with the probability of the propositions themselves.

These three precepts are essential. They show that the value of the results depends on propositions, case circumstances (for example the number of persons possibly involved), assumptions and knowledge. Depending on this information and the results we want to assess, different models will be adopted, hence different values obtained. It is therefore important to outline the results that we assess (e.g., allelic peak presence and peak heights), propositions, assumptions, and case information.

Consideration 2

As described by Evett et al. [18], there are no true likelihood ratios, just like there are no true models [19]. Depending on our assumptions, our knowledge and the results we want to assess, different models will be adopted, hence different values for the LR will be obtained. It is therefore important to outline in our statements what factors impact evaluation (propositions, information, assumptions, data, and choice of model).

4. Propositions

To assign the value of the results, based on the available information, case tailored mutually exclusive propositions are needed. Jackson et al. [11] summarised that there is a requirement for:

1) A prosecution and an alternative defence proposition must be proposed.
2) "These should be formulated from the framework of circumstances of the case and through dialogue between parties in the criminal justice system".

Where the 'framework of circumstances' is a detailed consideration of all the relevant aspects of the case, that includes the alternative views of the prosecution and defence.

The concept of hierarchy of propositions [5] is very useful to

[3] How high this LR needs to be in order to identify potential candidates for further investigation, will be a matter of laboratory policy. In practice, the candidates will be ranked from high to low LR. Each candidate will be investigated with respect to the background information and either forwarded for further investigation, or will be eliminated. There is a cost implication with large investigations, hence the policy will depend upon size of database, the relevant population, the type of crime and its seriousness.

[4] If a subset of the database is analysed e.g. because the perpetrator is known to come from a city of a few million then a much lower LR may be considered.

Table 1
Relationship of various levels in the hierarchy of propositions relative to the purpose, issues, results and factors to consider. Within source variation refers to the variability of the results (e.g. presence/absence of peaks, peak heights, DNA quantities) given that the DNA came from the POI. The table is adapted from the SEFE online course (http://www.formation-continue-unil-epfl.ch/en/formation/statistics-evaluation-forensic-evidence-cas/).

Level in the hierarchy of propositions	Purpose	Issue	Results	Factors
Sub-source	Investigation	Who could be the source of the DNA?	DNA profile	Occurrence in the relevant population
	Evaluation	Is the DNA from Mr S?		Within source variation
Source	Investigation	Who could be the source of the biological fluid?	DNA profile Presumptive tests	Occurrence in the relevant population
	Evaluation	Is the biological fluid from Mr S?		Within source variation Test false pos./neg. Cross-reactivity etc.
Activity	Evaluation Did Mr S perform the given activity?		DNA profile Presumptive tests Extrinsic characteristics (relative quantity of DNA, where it was recovered) Multiple traces	Occurrence in the relevant population Within source variation Transfer, persistence, and recovery (TPR) DNA present for unknown reasons (background)

identify how forensic science can help the court discriminate propositions of interest. There are now five levels in the hierarchy: sub-sub-source, sub-source, source, activity and offence. It is worth noting that it is always for the court to give an opinion on propositions, whatever the level. Depending on the results and the factors that forensic scientists take into account in their evaluation, they will need propositions to be set at different levels. The key point is that forensic scientists need to add value (thus have specialised knowledge) when considering propositions that are higher in the hierarchy. Table 1 can be used as a guide to choose the appropriate level. We have not included the offence and sub-sub-source levels. Indeed, it would be rare for DNA scientists to add value by considering their results given offence level propositions though it is possible (e.g., for combining evidence resulting from different activities or when multiple offenders are involved [20]). And, it would be rare for the DNA scientist not to add value when considering results given sub-source level propositions instead of sub-sub-source propositions (where one only considers part of the DNA profile, for example a major contributor to a mixture of two or more individuals).

In court, the expert may be asked to help address various levels in the hierarchy of propositions. Each level, however, requires a separate evaluation (i.e. a different LR). It is important to specify that an LR calculated given propositions that are at a lower level cannot automatically be carried over to propositions at a higher level. There are exceptions to this. For example, if there is a complete DNA profile recovered from a pool of blood from a stabbing-victim then the LRs given sub-source and source level propositions can be the same. Conversely, if quantities of DNA are low so that the LR given sub-source propositions is also low, and a detected body fluid is also low-level, then the association of the DNA with a given body fluid may be disputed by the defence. Under this circumstance, the LR assigned given sub-source level propositions can still be reported, but the LR given source level propositions (e.g. "the DNA from Mr X originated from blood vs the DNA from Mr X originated from skin cells") is a separate evaluation. Nevertheless, a calculation given source level propositions can still be made by the scientist [21] if the origin of the DNA is not agreed on.

To summarise, the purpose of the scientist is to evaluate the evidence given the question of the issue that is before the court which forensic science can help with. It is entirely dependent on the case circumstances, the background information and the court itself, whether both the source of the DNA and/or the activities are contested.

In order to make this clear, the scientist will generally include a caveat to the statement, such as: "*Assuming it is accepted* by all parties that the origin of the DNA is Mr X, the probability of recovering DNA on this item, with the observed relative quantity/quality, given that the person performed the alleged activity is…".[5]

Source level propositions are generally most appropriate when the type of tissue or biological fluid source of the DNA itself (e.g. semen, blood, saliva etc.) is not contested or is not relevant.

4.1. Propositions, value of results and likelihood ratio formulae

As mentioned earlier, the value of the results will depend upon propositions. Below we show examples of LR formulae with different pairs of propositions set at the sub-source level as an example (their choice will depend on case information).

4.1.1. Single DNA profile

Abbreviations are used to signify the prosecution and defence propositions as H_p and H_d respectively and the likelihood ratio (LR) is the ratio of two conditional probabilities written as short-hand in formulae. We follow the notation of Evett and Weir [4]

$$\mathrm{LR} = \frac{\Pr(E|H_p, I)}{\Pr(E|H_d, I)} \quad (1)$$

Where E denotes the evidence, or more specifically the results that are to be assessed (e.g., the DNA profiles of the references and the crime-stain) and Pr is probability; the vertical line is called the conditioning bar, since all probabilities must be conditioned on various assumptions and information. The elements behind the conditioning bar are taken as a given. The formulation in equation (1) can be verbally described as:

$\Pr(E|H_p,I)$: The probability of the DNA profiles given the prosecution proposition is true, and given conditioning information I versus

$\Pr(E|H_d,I)$: The probability of the DNA profiles given the defence proposition is true, and given conditioning information I

4.1.2. Mixtures

All likelihood ratio formulae follow this structure, but they can be complex. For mixtures, the evidence of the crime stain profile is denoted E_C. The main considerations are the number of contributors, and whether there is conditioning on a known person (e.g. a victim, or the

[5] It is instructive to read a UK appeal court decision: R. v. Weller, Neutral Citation Number: [2010] EWCA Crim 1085. The judgement states: "It therefore was common ground at the trial and on this appeal that the DNA had come from Emma." As the source of the DNA was not contested, the court moved on to discuss the next level in the hierarchy of propositions – in this case the activity level.

owner of the item). Under a typical set of propositions, the prosecution's view would be that the suspect contributed to the DNA mixture. The defence's proposition would be that an unknown person is the source.

Depending on the situation and the object analysed, the presence of DNA from one of the known persons may not be contested. For example, it may also be appropriate to include the victim as a contributor in both propositions. A typical case would be where we analyse a vaginal swab that contains semen. Indeed, there would generally be no dispute that the item came from the victim, thus there is a prior expectation of the presence of this person's DNA under both propositions. Let us suppose that the issue here is whether the DNA is from Mr S or some other person(s) and that there is no assumed known contributor (such as the person to whom the objects belongs). From the case information and from the observation of the crime stain profile, we can infer that the DNA mixture is from two persons. For this case, where the issue regards whether the DNA is from Mr S or not, the competing propositions could be:

H_p: The DNA mixture is from Mr S and an unknown person unrelated[6] to S

H_d: The DNA mixture is from two unknown persons, unrelated to each other or to Mr S

If we denote the results by:

E_C: DNA mixture profile derived from the crime scene

G_S: DNA profile of Mr S

Then, the likelihood ratio formula[7] is expressed as:

$$LR = \frac{\Pr(E_C|G_S, H_p, I)}{\Pr(E_C|G_S, H_d, I)} \quad (2)$$

With

$$\Pr\left(E_C \middle| G_S, H_d, I\right)$$

$$= \sum_i \sum_j \Pr(E_C|G_i, G_j, G_S, H_d, I)\Pr(G_i|G_j, G_S, H_d, I)\Pr(G_j|G_S, H_d, I)$$

where, G_i and G_j are the possible genotypes of the unknown persons.

If we now consider a three-person mixed crime-scene DNA profile, in a situation where an individual's DNA is assumed to be present under both views[8] competing propositions could be:

H_p: The crime stain contains DNA from Mr S, the victim and an unknown person

H_d: The crime stain contains DNA from the victim and two unknown persons, unrelated to Mr S and the victim

G_V: DNA profile of the victim

$$LR = \frac{Pr(E_C|G_S,\ G_V,\ H_p,\ I)}{Pr(E_C|G_S,\ G_V,\ H_d,\ I)} \quad (3)$$

We see here the importance of understanding the issue in the case (e.g., whose DNA presence is contested) in order to formulate useful propositions. Depending on these propositions, the likelihood ratio formulae will differ. They will also differ, depending on the assumptions made: hence these should always be mentioned as indicated above. The likelihood ratio approach is very flexible. It may be expanded, for example, to accommodate multiple contributors. The numbers of contributors do not need to be equivalent under both propositions [2], even if they generally are.

One advantage of the likelihood ratio over methods such as the combined probability of inclusion (CPI) and allied methods is that it can be used to express the strength of evidence to support the defence proposition as well as the prosecution proposition. A LR $>$ 1 supports the prosecution proposition rather than the defence's, whereas a LR $<$ 1 supports the defence proposition rather than the prosecution proposition. A LR of one is neutral, the results do not support one proposition more than the other.

Recommendation 1:

The value of DNA and biological results is given by assigning a likelihood ratio. This implies the formulation of at least two mutually exclusive propositions. Assumptions regarding the model and the background information (i.e., case information and data) used should be disclosed.

4.2. Formulation of propositions at sub-source level: common pitfalls

Ideally, in order to prevent bias, propositions should be set *before* knowing the results of the comparison between the contested DNA and a possible contributor. This is part of case pre-assessment described by [13,23]. However, the probability of the results given both propositions may be unexpected and may prompt the forensic scientist to go back into investigative mode [8]. For example, suppose individual X is accused of an assault and the results support the proposition that X is not the source, but an unknown person is. This leads to further investigation (e.g., a database search may suggest a new candidate) and a new set of propositions is therefore formed.

4.2.1. Distinguishing results from propositions

Hicks et al. [24] explain that observations should not be interwoven with propositions. An example of this error with sub-source level propositions would be:

H_p: The matching DNA comes from candidate S

H_d: The matching DNA comes from an unknown person

A DNA profile is said to 'match' if for all shared markers, the allele designations in the crime stain profile are the same as the alleles with which it is being compared to. This should not be confused with the 'identity' of the donor. For a discussion on 'match' versus 'identity' see [25].

Consequently, with this example, the results (the 'match') clearly appear in the proposition. Consider now how to evaluate the matching profiles given these propositions. Under prosecution's proposition the probability of seeing matching profiles given that you have matching profiles from candidate S is 1. Indeed, if the scene of crime DNA profile is $E_C = \{a,b\}$ and the candidate S has genotype $G_S = \{a,b\}$, then the probability of observing a 'match' is one, i.e. $\Pr(E_C = \{a,b\}|G_S = \{a,b\},H_p) = 1$). Similarly, the probability of observing matching profiles, given that you have a matching profile that comes from an unknown person is 1, i.e. $\Pr(E_C = \{a,b\}|G_U = \{a,b\},H_d) = 1$. Consequently, if the unknown person has a matching profile, he/she must have genotype ($G_U = \{a,b\}$).

Therefore:

$\Pr(E|H_p,I) = 1$ (matching DNA comes from S)

$\Pr(E|H_d,I) = 1$ (matching DNA comes from an unknown person)

Hence LR = 1 and we are no further forward because the evidence value is neutral[9]. The court is left with assessing the 'match' without

[6] It is standard to apply the 'unrelated' caveat. But of course it should be pointed out that if there is the possibility from case circumstances that a relative such as a brother was the perpetrator then the propositions ought to reflect this. It is also possible to carry out a single calculation that includes grouping potential relatives e.g., brothers, cousins – see section 3.5.3 of [22].

[7] For convenience, some notations in the literature may include the unknown genotype G_U after the conditioning bar, but the formal/standard way is to condition only upon information that we have. We do not have information about the unknown individuals, so it should not appear in the conditional (i.e., behind the conditional bar).

[8] Therefore, not contested by either party.

[9] A further general example, involving paint fragments in an accident, could be: what is the probability of a match (blue = blue) given that the matching (blue = blue) paint comes from the person's car and given that the matching (blue = blue) paint comes from an unknown car. If the paint matches, then the car must be blue; hence the probability of observing this match given there is a match is one. One cannot have the same words to describe the results and the propositions.

any help from the DNA scientists.

4.2.2. Avoiding the inclusion of results in propositions

To reiterate, in order to formulate propositions that are not biased against a given person, it is important to formulate them *before* the comparison process involving a person whose DNA presence may be contested. However, it is admissible to formulate propositions based on the trace itself (for example, in order to determine if the trace is a mixture). This is also valid for the expected presence of DNA of a person given both points of views. This would typically be the case for the DNA profiles of the persons who own the objects or for intimate swabs. Continuing with the previous examples, findings-led propositions are avoided by dropping the word 'match':

H_p: The DNA comes from Mr S

H_d: The DNA comes from an unknown person

Now there is no mention of the results (i.e. the 'matching' DNA). It is clear that the propositions are independent of any results.

Recommendation 2:

Results should clearly be distinguished from propositions [9], as DNA specialists assess the former and decision makers the latter. Avoid terms like:"the matching DNA comes from X".

4.3. Examples of possible findings-led propositions

This example is provided as it has often been observed in casework and is routinely raised by participants on training courses. It also exemplifies the importance of distinguishing between our roles as investigators and evaluators. The 'two suspect' problem [9,26,27] was initially identified from a casework example where two individuals, Mr. Smith and Mr. Doe were accused of a violent assault and a three-person mixture was retrieved from a crime stain, a skin swab taken from the victim. The LR was assigned considering both suspects as contributors under the prosecution proposition, and neither being contributors under the defence proposition. Although a high LR was obtained supporting the prosecution proposition, because the mixture was partial and unbalanced, using non-contributor analysis, for investigation purposes (described in section 4.3.1.), it was shown that the proposition that suspect (Mr. Doe) was a contributor of DNA to the mixture could not be supported. Consequently, there was danger of misrepresenting the evidence by applying a single likelihood ratio to both of the two defendants together in the case. In the UK Regulator's Forensic Science Guidance consultation on DNA Mixture Interpretation [10], they draw attention to this element as well: *"if the questioned profile is partial and unbalanced then it would seem wrong to assign the same evidential weight to both POIs, particularly if the genotype of one has alleles corresponding to large peaks, whereas the other has alleles that appear as peaks close to the analytical threshold"*. Clearly, the problem becomes bigger with higher order mixtures, such as four or five persons.

4.3.1. The two suspect problem

Suppose that the police arrested Mr Smith and Doe and accused them of both being complicit in a murder. A sample was taken from the victim's body, from an area of importance, but which did not appear to have any body fluids present - the evidence was assumed to comprise epithelial cells and free DNA that reflected a so-called 'trace' DNA sample (the terminology of 'trace' versus 'touch' DNA will be discussed in the second paper). Two persons were potentially implicated. However, the defendants claimed to have been elsewhere at the time and presented alibi evidence, saying that they were in each other's company. The police requested the scientist to evaluate the evidence in order to help establish whether or not DNA was present from the two persons of interest.

The scientist then analyses the trace and it is evaluated as a mixture of three people. Upon inspection, the genotype of the victim is well represented as expected.

4.3.1.1. Our role as investigator. Observing the profile, the scientist notes many corresponding elements between Mr Smith and the trace. The trace and Mr Doe's DNA profiles also have some allelic designations that are the same, but there are a number of differences, which is not unexpected given the quantity of DNA. Indeed given the first proposition, dropout could have occurred. In such a case, as mentioned, it might be of interest to the police investigation to know if it is possible that the mixture originated from all three given persons and the scientist may be asked - for investigation purposes - to consider the DNA results given the following preliminary sub-source propositions:

- The DNA mixture originates from Mr Smith, Mr Doe, and the victim
- The DNA mixture originates from the victim and two unknown individuals

However, with these kinds of propositions it is important to be aware that the results are taken as a package. Considered apart, with two sets of propositions and two separate likelihood ratio calculations, the value of the comparisons will be different, but considered together there is only one value that applies to both POIs. In other words, taking the proposition that the mixture is from victim and both Smith and Doe, the calculation does not give any information regarding the individuals as they are taken as a whole - i.e. the strength of the evidence is used to infer the presence of both individuals, and not one in isolation of the other. Consequently, it is entirely possible that a large likelihood ratio can be achieved when one (Mr Smith) is the ground truth donor and the other (Mr Doe) is not. It is therefore desirable to explore whether this is the case and there are two methods to do this: individual LR calculations (for evaluative and investigative purposes) and non-contributor tests (that are particularly appropriate in investigative mode, when there are no suspects in order to decide whether one should search the mixture in a national database).

Suppose a likelihood ratio of x is calculated for a crime stain. Non-contributor (*nc*) tests are carried out by replacing each suspect (S) in turn with a large number of random ($R_{1..N}$) profiles [27] to generate a new LR_{nc} per random profile. For simple proposition pairs (Table 2), how often a $LR_{nc} = x$ or more is accommodated by $Pr(LR_{nc} > x|Hd) < = 1/x$, so we expect less than N/x 'matches', where there are N comparisons [28] and the mean $LR_{avg} = 1$ under Turing's expectation [26]. For complex proposition pairs shown in Table 2, how often $LR_{nc} > x$ occurs is accommodated by $Pr(LR_{nc} > x|Hd) < = LR_{Sa}/x$, where LR_{Sa} is the LR produced using propositions where the known contributor in the numerator is S_a. For the example in Table 2, LR_{Sa} is

Table 2

Non contributor (*nc*) and Turing's expectations for simple propositions (e.g. *Hp*:S + U; *Hd*:U + U) where suspect S_a (S_1 = Mr Smith; S_2 = Mr Doe) is replaced by random profiles (R) in *nc* tests to generate a series of $LR_{nc} = x$ (V = victim; U = unknown). For complex propositions (e.g. *Hp*:V + S_1 + S_2; *Hd*:V + U + U) either S_1 or S_2 are replaced in turn with R to carry out *nc* tests.

Proposition type	H_p	H_p (*nc* test)	H_d	Expectation (*nc*)	LR_{avg} (Turing's expectation)
Simple proposition pairs	S + U	R + U	U + U	$Pr(LR_{nc} > x\|Hd) \leq 1/x$	$LR_{avg} = 1$
	V + S	V + R	V + U	$Pr(LR_{nc} > x\|Hd) \leq 1/x$	$LR_{avg} = 1$
Complex proposition pairs	V + S_1 + S_2	V + S_1 + R	V + U + U	$Pr(LR_{nc} > x\|Hd) \leq LR_{S1}/x$	LR_{avg} scaled to LR_{S1}
	V + S_1 + S_2	V + R + S_2	V + U + U	$Pr(LR_{nc} > x\|Hd) \leq LR_{S2}/x$	LR_{avg} scaled to LR_{S2}

produced using propositions:

H_{p1}: The DNA originates from the victim, Mr Smith (S_1) and an unknown person

H_{p2}: The DNA originates from the victim, Mr Doe (S_2) and an unknown person

H_d: The DNA originates from the victim and two unknown persons

For complex proposition pairs, the average $LR_{avg} = LR_{Sa}$ for Turing's expectation, hence calculations to test $LR_{avg} = 1$ must be scaled with respect to LR_{Sa}.

If a series of randomly generated profiles give LRs that are of the same order of magnitude as the one achieved in the case, then we can conclude that the model is not discriminating with regard to that particular individual i.e. the results are not informative. Under this circumstance, the non-contributor test may also be reported in investigative mode. An example is:

4.3.1.2. Investigative reporting example[10]. "This figure can be qualified with an investigative test known as a 'non-contributor test'. To do this we replace Mr Smith with a random unrelated individual and we repeat the measurement of the likelihood ratio. We do this a total of 1000 times, with a different random individual each time. When this was carried out with Mr. Smith, the maximum likelihood ratio observed was of the order of 0.01. However, when Mr Doe was substituted with 1000 random samples, the maximum LR observed was 10 million. This shows that although the trace can be explained as a mixture of the three persons, the possible contribution of Mr Doe needs to be viewed with great caution and one should assess the value of the profiles (from Mr Doe and Mr Smith) separately. Indeed, the likelihood ratio given the proposition where both POIs are contributors may be large, but the information provided by the minor contributor (Mr Doe) is too small to help discriminate him from a random person."

4.3.1.3. Our role as evaluators. In the above example, if the issue is whether or not Mr Smith or Mr Doe contributed to the mixture, then, as recommended by [10], the way forward would be to evaluate the evidence of each POI separately, and assign individual LRs. To do this, two different constructs are required, where a LR is assigned separately for each of the POIs.

- The DNA originates from Mr Smith, the victim and an unknown person
- The DNA originates from the victim and two unknown persons

and

- The DNA originates from Mr Doe, the victim and an unknown person
- The DNA originates from the victim and two unknown persons

This results in two separate likelihood ratios, for Mr Smith and Mr Doe respectively.

Alternatively, if it is accepted by the defence that the other person's DNA is present in the crime stain [8,9], then the accepted propositions may be (for Mr Smith):

- The DNA originates from Mr Smith, Mr Doe and the victim
- The DNA originates from Mr Doe, the victim and an unknown person

or for Mr Doe

- The DNA originates from Mr Smith, Mr Doe and the victim
- The DNA originates from Mr Smith, the victim and an unknown person

4.3.1.4. Evaluative reporting example. "The DNA profile of the trace has been compared with the profile of Mr Smith, Mr Doe and the victim. Based on the information available, the presence of the DNA from the victim is not contested, but the DNA of the two other persons are. The value of results from Mr Smith and Doe has therefore been assigned separately.

For Mr Smith, the results of the comparison were assessed given the proposition (a) that the DNA originates from Mr Smith, the victim and an unknown person and the proposition (b) that the DNA originates from the victim and two unknown persons. The DNA results are in the order of 10 million times more probable if the first proposition (a) is true than if the alternative (b) is true.

For Mr Doe, the results of the comparison were assessed given the proposition (a) that the DNA originates from Mr Doe, the victim and an unknown person and the proposition (b) that the DNA originates from the victim and two unknown persons. The DNA results are in the order of 100 times more probable if the second proposition (b) is true, compared to the alternative (a)[11].

If any of the given case information is incorrect or if further information is made available (in particular with regards to propositions), it will be necessary to reconsider the evaluation of the results."

These different schemes help address the individual contribution in different ways. The 'two suspect effect' occurs only when the issue regards whether the person(s) of interest contributed to the trace or if unknown person(s) did instead (there are two or more 'known' individuals appearing in the numerator that are *both* replaced by 'unknowns' in the denominator). If the presence of a person's DNA is not contested by either party, then this obviously has no effect (e.g. in a simple case only one known individual is replaced as unknown: DNA is from suspect and victim vs. DNA is from unknown and victim, so that there is no effect if a victim's DNA presence is assumed in both numerator and denominator).

The two-suspect problem is an example of a wider situation encountered in forensic biology, whereby there are multiple references for comparison to the evidence[12]. For example, police may have submitted items from a drug house along with the references from 10 people who are suspected of working there. Given the ability of multiplexes to generate DNA profiles from very little material, it is likely that multiple profiles will be obtained in the case, and there is little case circumstance to guide which (and in what combination) references should be conditioned upon. In these cases, scientists work more as investigators. As outlined in [8] it can be useful to indicate which combinations are compatible with the mixture and whether additional contributors are needed to explain the mixture. One can provide LRs for each individual provided that the propositions are based on some case information. If propositions are constructed in this way and LRs support multiple individuals as contributors, then some indication of whether they can jointly be contributors is often -as noted above- an important additional consideration from an investigator's point of view. Provided that known individuals are included under both prosecution and defence propositions, they can be accommodated without restriction (illustrated by the *victim* in the two-suspect example discussed above). However, an important caveat is the implicit assumption that the defence are in

[10] Suppose that the LR calculated for Hp: victim, Mr. Doe and Mr. Smith vs. Hd: victim, unknown, unknown is of the order 10m. For the non-contributor test we carry out the analysis as described in Table 2, substituting Mr Doe and Mr Smith respectively in each series of tests. A $LR < 1$ favours the defence hypothesis.

[11] Here a $LR > 1$ favours the defence proposition as the propositions are inversed: $LR = \Pr(E|H_d)/\Pr(E|H_p)$

[12] As noted in [8] these complex situations with multiple contributors and POI's might be better suited for the investigation phase than the evaluation phase.

agreement that a known individual(s) can be included under the alternative in the way suggested above.

Recommendation 3

Propositions should be formulated in order to help answer the issue at hand and be based on the case information (not on the results of the comparison). They should be formulated without knowledge of the results of the comparison made between the trace and the person whose DNA presence is contested (e.g., the suspect's).

When the issue regards the possible presence of DNA from several persons of interest, effort should be made to evaluate the profiles separately, and not as a whole. This is especially important if the information available from one part of the profile (e.g. major) is different from the other (minor, partial). For evaluation, this can be achieved by considering the result of the comparison between the given person and the trace and calculating individual LRs for each person. The report should be fully transparent on what propositions have been considered and on what basis.

For *investigative* purpose, it might be useful to explore whether the results support the proposition that the two persons together are (or not) the source of the DNA. In such a case, one can assign one LR. A non-contributor test can be helpful, also for investigative purposes.

4.3.2. Assigning the number of contributors

The number of contributors may affect the value of the results (i.e. LR) significantly. A commonly used method to assign the number of contributors, called the maximum allele count (MAC), is based upon summing the maximum number of unique alleles (L_{max}) observed at a locus in a crime-stain, including the set of known contributors in a proposition and dividing by two [27][13]. The number of contributors is portrayed as equal or smaller than $L_{max}/2$. The assessment of number of contributors may be corroborated by using the total allele count (TAC) as described by [29]. However, this scheme does not take into account peak height, which can be an important factor, and is often used by analysts in the determination of the number of contributors to a DNA profile. Utilisation of peak heights requires a basic understanding of DNA profile behaviour (i.e. stutter, peak height variability and additivity of masked peaks) and there has been ample literature on these behaviours [30–33].

The higher the number of contributors, the more likely it is that the number is underestimated because of 'allele masking' where alleles are shared between different individuals [34]. If this is a concern, one can add a contributor to the propositions. Quantitative models are usually insensitive to an excess number of contributors being postulated (i.e. the LR is changed very little), especially if there is little dropout of the alleles shared between the trace and the POI. This is because quantitative models assign very low mixture proportions to excess contributors [35] hence their contribution to the LR is minimal. A probabilistic solution to the problem of assigning the number of contributors (*nc*) has been given [36] where it is suggested to use priors for a range of values of *nc* (which may differ between H_p and H_d) in single calculation of the LR.. This is an ideal solution that requires input of prior probabilities regarding the number of contributors[14].

Where additional contributors have a demonstrable effect on the value of the results, primarily for qualitative models, if the addition of contributors was unrestrained, and equivalent under both propositions then the LR value will tend to decrease as more contributors are hypothesised. It would be clearly unreasonable to hypothesise ten contributors to a three-person mixture, for example.

The solution was proposed by the ISFG DNA commission [2], recommendation 5 and this remains the best advice to deal with uncertain numbers of contributors, updated here:

To conclude Evett et al. [37] have shown that:

> *Provided the scientist has followed the guidelines and addressed propositions that are based on the number of contributors that best explains the questioned profile, then it is not to the advantage of the defendant to change the defence proposition to address a greater number of contributors.*

Similarly, Taylor et al. [38] carrying out the same process probabilistically conclude that:

> *It should also be noted that due to the slight favouring of simpler (lower contributor) models, there is still no advantage in artificially increasing the number of contributors to one or both of the hypotheses as this will tend to drive the LR's support away from the proposition with the greater number of contributors.*

and Budowle et al. [39] state:

> *While the formal logic for calculating the LR is provided elsewhere…we stress that every effort should be made to provide the best estimate of the number of contributors. It is not in the best interest of the defence to suggest (an) unreasonable number of contributors; usually this will increase the LR favouring the prosecution's position.*

There are always exceptions however, which is why it is useful to explore and report the effect of the number of contributors on the calculations, provided that the propositions are reasonable in the context of the case [40–43].

Recommendation 4 (adaptation from [10])

The scientist should assign a value (or several) to the number of contributors to the trace. This will be based on case information, the observation of the DNA profiles of the trace and of the persons whose DNA presence is not contested (e.g a victim in an intimate swab). The reasoning to support this decision should be indicated.

4.4. Sub-sub-source level propositions: major contributor

If the motivation for using only part of the mixture (e.g., major components) is based on the trace only and not on the result of the comparison, then it is permissible to consider propositions such as :

- The POI is the origin of the DNA from the major component
- An unknown person is the origin of the DNA from the major component

These propositions have been named sub-sub-source level propositions [44].

However, one should note that this simplification works only if it is possible to unambiguously distinguish a major component of the DNA profile at all loci, and is based on defined and comprehensible criteria

[13] This process may require a pre-screen to remove stutters. If there is uncertainty about whether potential stutters should be removed then they may be included as alleles in the assessment. Note that profiles from known individuals included in propositions must be included in the assessment of numbers of contributors. Suppose we have a crime stain with alleles *a,b* (an apparent single contributor) and a suspect reference with alleles *c,d*. Under the prosecution proposition the *c,d* alleles have dropped out. There are four unique alleles in the set, hence a minimum two contributors under *prosecution proposition* but only one under *defence*. Adopting $N+1$ contributors might introduce prosecution bias. When faced with these kinds of situations it is recommended that it may be appropriate to explore several different models with different numbers of contributors. If there is a strong difference between use of N vs. $N+1$ contributors then this must be disclosed in reports, along with reasoned explanations.

[14] When deciding the numbers of contributors (*nc*) we apply a Pr = 0 or 1. The proposal of [36] is to apply priors to a range of *nc* values so that the Pr takes a value between zero and one; all the probabilities must sum to one.

e.g. [45].

Sub-sub-source level propositions are not appropriate if any of the following circumstances are fulfilled:

a)
   a) If both minor and major components have been compared to the POI
   b) The components cannot be clearly classified into major/minor
   c) The probabilistic genotyping method takes into account peak height, or assigns different rates of drop-out to different contributors

Then the whole mixture should be considered using standard sub-source propositions such as:

- The POI and an unknown person are the origin of the DNA mixture
- Two unknown persons are the origin of the DNA mixture

Recommendation 5

In straightforward cases, without reference to the profile of the person of interest, one can adopt propositions considering only part of the mixture (i.e. a clearly defined major contributor). But, in general, it is best to consider the mixture as a whole.

4.5. Source level propositions

The ENFSI guideline for evaluative reporting [13] gives examples when source level propositions are appropriate. A typical example for forensic genetics would occur when a large pool of blood is found from which a single good quality DNA profile is derived. In this case, the origin of the biological fluid will generally not be in dispute. Therefore, it is not problematic to have the word 'blood' in both propositions

Examples of source level propositions could be:

- Mr S is the origin of the blood
- An unknown person is the origin of the blood

If the source of the biological fluid(s) is disputed (for example if there is a small quantity of body fluid or in the case of a mixture where it is not clear which contributor provided the detected body fluid), then it will generally not be appropriate to report given source level propositions [21,46]. More value will be added by considering activity level propositions.

5. Value of results: type of results and models

Different models may be used to assess the results of the same case, for example the prosecution and defence may carry out separate analyses using separate software. These models may be based on different results (e.g. peak heights or not) and different modelling assumptions. It would be expected that different answers will be obtained but in general, models based upon similar principles should provide LRs that are broadly similar. To place into perspective, according to [47] an order of magnitude difference is considered negligible. If two LRs are widely divergent, e.g. there are several orders of magnitude difference or if one provides support for one proposition, and the other gives support for the alternative, then the reasons for discrepancies need to be explored, ideally before the court report is issued[15].

Consideration 3

It is to be expected that different (validated) models will be used to analyse the same DNA profile for court, for example if defence wishes to check an analysis forwarded by the prosecution. Cross-checking results using validated models based on different principles are to be encouraged for quality reasons. Although different models, prepared by different providers, should provide'similar'results if the input data are the same, they will nevertheless differ if modelling assumptions vary. Differences of an order of magnitude are considered negligible. If two different models give widely divergent results, then investigation is required to discover the reasons.

5.1. Mixtures and low level STR DNA profiles

There have been two previous ISFG DNA commissions that have reported on STR DNA mixtures and recommended methods that take account of drop-out and drop-in [1,2]. Over recent years a number of new software solutions have been introduced to interpret more complex DNA samples (multi-contributor mixtures with dropout and drop-in); for further details see review article [48]. With the introduction of new multiplexes, the sensitivity of detection has simultaneously increased so that it is now routine to analyse a very few number of cells [49] – see a review of over 24,000 samples that were analysed by six laboratories [50,51]. Consequently, it is more likely that low-level mixtures from multiple contributors are encountered. This in itself raises new interpretation challenges. Early methods to interpret complex, challenging DNA profiles used the consensus method [52] that required replicate analysis of a sample extract; only those alleles confirmed to be present twice were reported. Variants on the consensus method have been reported [53]. It should be noted that the consensus methods were always meant as approximations of what was referred to at the time as the 'statistical' method (which we would now call a probabilistic method). They came to achieve popularity as they did not require custom written programs to apply.

There are limitations to the consensus method, especially when there are high dropout rates. Consequently, the method is being superseded, partly as a result of the second ISFG DNA commission [1] which advised on the use of 'drop-out' and 'drop-in' models. Since publication, there has been a plethora of models introduced, some of them qualitative [54–57], (alternatively described discrete or semi-continuous), taking into account alleles without direct reference to the peak height and other characteristics, and others that are quantitative [58–61], taking into account allele peak height, stutter, degradation, and other attributes of a DNA profile (see reviews by [47,48]). There is now a general direction within the community towards adopting these kinds of models to interpret complex DNA profiles, supported in Europe by training programmes, sponsored by ISFG, EUROFORGEN-NOE and CEPOL. The consensus methods are therefore quickly becoming redundant. The likelihood ratio framework is always used as the method to express the strength of the evidence of complex DNA profiles (i.e. mixtures that may have drop-in/drop-out), because 'match probabilities' and allied methods do not make sense to use within the mixture interpretation framework.

Consideration 4

The value of DNA results will depend on the data used and on the model adopted. We make no recommendation on which probabilistic models should be used, other than they should have been validated and their application and limitations understood within the guidance provided by a recent ISFG DNA commission [62].

There is no accepted definition about what constitutes low-template (or low copy number) DNA. The increased sensitivity of modern multiplexes and introduction of new analytical platforms such as the AB 3500xl (Thermo Fisher Scientific) means that all laboratories have a capability to analyse very small amounts of DNA. However, there is no clear-cut delineation between conventional and low-template DNA, and it is not helpful to think in these terms [63]. It is often the case that in mixed samples, some contributors are low-template while others in the mixture would be considered as high template. Certainly, with lower

[15] This would best be progressed by expert discussion to discover the common ground rather than an argument in a court-room.

levels of DNA, issues of contamination, background and secondary transfer become more important, but this does not mean that these issues are avoided with so-called 'conventional DNA'[16]. Indeed, a belief that conventional DNA profiling is somehow free of these effects gives a false sense of security. However, the methods discussed in this paper can be universally applied so that any distinction between conventional and low template DNA is essentially redundant. There is of course a limit of generating useful information when the stochastic variation resulting from the amplification of very low amounts of DNA overrides any meaningful peak information. Consequently the overall profile quality must be assessed and considered to be acceptable before any attempts to compare and interpret such STR typing results are made.

Recommendation6

For complex cases (e.g. mixtures), the assignment of a likelihood ratio using validated software is prerequisite to determine the value of evidence. A number of different software that can accommodate drop-in, drop-out, peak height variability or relationships are available to assist with the approach. Probabilistic models are now preferred to the consensus approach.

6. Value of results: relevant population and data

6.1. Relevant population

When a suspect has been identified, it is often assumed that the alternative proposition is that the DNA is from some unknown unrelated person (or random man). The question regarding the population of origin of this unknown individual [64] depends on the case information. Usually the relevant population is considered to be anyone in the local region (unless there is specific information that suggests the true offender belongs to a specific bio-geographic population). One must note, that if the suspect is considered as a possible offender, then this means that the possible offender may also come from the same subpopulation. Because, the relevant population will often comprise more than one bio-geographic group, to address this one database is compiled per group. The evidential strength will be different for each population that is considered to be relevant for an alternate source of DNA. There are methods for stratifying the multiple evidential weights that have been obtained using the separate ethnic databases into a single value [65]. This is a very elegant solution. When analysing multiple loci the questions regarding relatives becomes more and more pre-dominant. If it is appropriate (e.g., because of the case circumstances), one should consider close relatives, not available for testing, as shown by [17,22] and consider alternatives like:

- The DNA is from Mr X
- The DNA is from a brother of Mr X

One can also consider siblings, parents, cousins and unknown individuals by using what has been coined the unifying formula [17], pp.139-146.

- The DNA is from Mr X
- The DNA is from an unknown person related or not to Mr X

Recommendation 7

The reference population used in an analysis, and the genetic relationship between contributors, must be explained in a statement. One should ensure that the alternative is appropriate and sufficiently specific. If the case circumstances suggest that a relative might be an alternative source, one should consider relatives of the person of interest.

[16] 'Conventional' in the sense that the total amount of DNA in the PCR assay exceeds a stochastic threshold, dependent on methodology, typically more than 200 pg for a single trace.

6.2. Data used to assign the probability of a DNA profile

6.2.1. Population databases

Calculations used to assess strength of evidence are dependent upon an assessment of allele probabilities at loci of interest from a relevant population. To facilitate, it is standard practice to collect samples from known individuals originating from the same population. Before inclusion into a frequency database, profiles are checked to ensure that there are no duplicates, that samples originate from unrelated individuals and there are no allele designation errors [66]. It is usual for scientists to sample local populations before undertaking routine genetic analysis for any given system hence a large number of geographic populations are available on line. The ENFSI DNA Working Group has collaborated to collect data for European population databases to make them publicly available (STRidER) http://strider.online/ [67]; the NIST database also has worldwide populations at STRBase; http://strbase.nist.gov/populationdata.htm.

6.2.2. Sub-structure of populations

In the evaluation of evidence, the optimal position is to neither understate nor overstate the strength of the evidence. Many jurisdictions apply adjustments to calculations in order to err on the side of conservativeness, in order to ensure that the evidence is not overstated. Adjustments may be made to compensate for bias related to small sample sizes of the population database (to correct for low incidence of alleles that may not be properly represented in the sample); Fst corrections may be simultaneously applied to correct for sub population effects, taking account of shared ancestry between the defendant and possible offender. A number of different size bias corrections have been proposed [68,69] whereas the formula to correct for sub-population effect has been defined by Balding and Nichols [70] and this is generally adopted in criminal matters. For the value of the Fst, one can refer, for example, to the Table 3 of the study by Buckleton et al. [71].

Consideration5

Methods used to assign the strength of evidence for DNA profiles include the use of adjustments–size bias corrections and Fst (theta) in order to take account of the effect of population substructuring. The aim is to be fair and in any case not to overstate strength of evidence.

7. Value of results: communication

In the press, at trial, in statements, at conferences, it is common to encounter the fallacy that is known as the transposed conditional [72] or the prosecutor's fallacy. It has been known about for more than 30 years. The fallacy consists of thinking that the probability of the results *given the propositions* is similar to the probability of the propositions *given the results* (consequently transposing the conditional). An example of a transposed conditional would be [73]:

> "(…) using the Australian Caucasian population sample data it was 800 billion times more likely that the sample originated from the accused than from an unknown person taken at random."

This statement is based on the propositions and not on the results, it is incorrect reasoning.

If we use the notation presented earlier and write the statement:

H_p: The sample originated from the accused

H_d: The sample originated from an unknown person

E_C: DNA profile of the crime scene stain

G_S: DNA profile of the suspect

I : is the conditioning information

With $E_C = G_S$

$$\frac{\Pr(H_p|E_c,\ G_s,\ I)}{\Pr(H_d|E_c,\ G_s,\ I)} = 800\ \text{billion}. \quad (4)$$

The propositions above are conditioned upon the results, therefore these are posterior odds (not a likelihood ratio). If the statement is changed to "…the results are 800 billion times more likely *if* the sample originated from…" then the results are now correctly conditioned upon the proposition.

Then, the likelihood ratio formula is correctly expressed as:

$$\frac{\Pr(E_C|G_S,\ H_p,\ I)}{\Pr(E_C|G_S,\ H_d,\ I)} = 800\ \textit{billion} \quad (5)$$

7.1. Avoiding the transposed conditional

There are a couple of ways to avoid the transposed conditional [15]: for example in the scientist's statement there should always be an 'if' or a 'given', and the statement ought to be on the results not on the propositions. An example of a correct statement of the value of DNA profiles would be:

> "(…) using the Caucasian population sample data the DNA results are a billion times more probable ***if*** the sample originated from the accused than if it originated from an unknown person taken at random."

When the statement contains the word 'that', then this should raise a red flag.

7.2. An example of impact of prior odds: database searches

The 'database controversy' arose from a recommendation of the National Research Council (NRCII) in 1996 [74] where they proposed that the strength of the evidence from a database search be reduced by dividing the likelihood ratio by the number of people in the database. This idea was refuted by various authors [75,76] who concluded that the evidential value of a DNA match was actually increased when a database size *N* was searched, since non-matching individuals were eliminated as the donor. The controversy hinged upon the choice of propositions to consider. Stockmarr [77], who supported the NRC recommendation proposed the correct propositions were:

- The donor of the crime stain is in the database
- The donor of the crime stain is not in the database

However, opponents [76,78] pointed out that this does not address the relevant court-going question which is:

- Mr X is the donor to the crime stain
- An unknown person is the donor to the crime stain

There is a consensus that the second set of propositions is the appropriate version for court; the database search has little effect on the likelihood ratio. But, we can argue that the first set is appropriate when we act as investigators, hence this distinction allows us to acknowledge what is our role and what is the issue we are helping with.

Regardless of consensus, a continuing debate periodically re-emerges in the literature on the effect of the database search on the strength of the evidence. However, it is not the value of the results that is mainly impacted, but the value of the propositions. To understand the reasons for this we now need to consider two types of crime described by situations (a) and (b):

a) A suspect is identified by non-DNA evidence, such as eye witness identification, which enables his/her apprehension without recourse to a database search.
b) There is no initial non-DNA evidence to identify a suspect. A database search is carried out and an individual is identified.

To begin, before the evidence is known, we assume that the perpetrator is from a suspect population of size *N*. Next, assuming that the DNA evidence has a very high value, but before considering the non-DNA evidence, the likelihood ratio is slightly greater in the situation (b) because *N*-1 individuals are eliminated from the inquiry. There has been no database search with (a) so no individuals are eliminated from the population – but the effect of the non-DNA evidence has not been considered yet.

Nevertheless individuals could be wrongly implicated by a database search and the case of Raymond Easton[17] is a classic example. In order to explore how this can happen, given that the likelihood ratio appears highly probative, it is necessary to consider that in situation (a) where there is no database search and before the DNA evidence is known, based on the other information there should be a high probability for the court to believe that the suspect is the perpetrator compared to anyone else. In situation (b), where there is a database search and before the DNA evidence is known, in the absence of other information, there is an equal probability for the court to believe that the suspect is the perpetrator compared to anyone else, but when there are many people to consider, their prior probability for any single individual (such as the suspect) being the perpetrator, is very low. Note that there is no *a priori* reason to suppose the perpetrator to be on the database, hence the comparison must always be with the population of individuals who could have committed the crime.

A priori belief regarding the propositions is expressed mathematically by prior odds. When there is a defendant, prior odds and resultant posterior odds are the remit of the court[18] and not that of the scientist. It is expressed as:

$$\begin{aligned}&\text{Posterior odds (the DNA comes from Mr T. vs. the DNA comes from an unknown person)}\\ &\quad = \text{prior odds x likelihood ratio}\end{aligned}$$

Consequently, in the examples above, prior odds for the situation (a) are high whereas the prior odds for the situation (b) are very low. In the case of Raymond Easton, there was no other evidence to suppose that he was implicated compared to 30 million other men in the UK. For illustration purposes, based on a population size $N+1$, this can form a prior probability $1/(N+1)$ and prior odds of 1 to *N*. Then, with these prior odds, posterior odds are 37:30 and the posterior probability that Easton is the source is approximately 0.5. This is shown in the calculation below[19]:

$$\begin{aligned}&\textit{Posterior odds} = \textit{prior odds} \times LR\\ &\textit{Posterior odds} = \frac{1}{30\times 10^6} \times 37 \times 10^6\\ &\textit{Posterior odds} = \frac{37}{30}\\ &\textit{Posterior probability} = \frac{37}{(37+30)}\\ &\textit{Posterior probability} \approx 0.5\end{aligned} \quad (6)$$

Provided other evidence is found in the case then prior odds can always be updated with the new information provided e.g. in the case of a burglary, following a database search:

[17] The evidence was given as *sic* [37-million-to-one]. This individual was charged with burglary, even though he lived 175 miles away from the crime scene, he was in the advanced stages of Parkinson's disease and was unable to walk more than 10m unaided https://www.theguardian.com/uk/2003/apr/27/ukcrime7

[18] In the inquisitorial system it may be appropriate for the scientist to actively engage with a discussion on the prior odds under direction of a judge.

[19] To carry out the conversion, if the odds are a:b (eg., 37:30), then the probability is a/(a+b) (e.g., 37/67). With prior odds of 100:1, the probability of the proposition will be 100/100+1.

1) Stolen items may be found at the premises of the suspect
2) Associates may be found who give evidence against the suspect

On the other hand, prior odds can be assigned as zero, for example where there is evidence that the person cannot be the source of the DNA, so posterior odds are also zero. The database search may impact prior odds indirectly by revealing investigative leads that provide information which further informs the prior odds (in this example points 1 and 2 above result in a substantial increase in the court's prior odds of guilt for the suspect). However, in some cases there is no other evidence other than the DNA profile and the correspondence following a database search. In such cases, prior odds are generally small.

In R v. Tsekiri [79], the defendant was found guilty of burglary. The only evidence was presence of DNA (mixture) on a door handle, and the court acknowledged: *"the finding of DNA attributable to a defendant at the scene of a crime was the sole evidence against a defendant*". The jury was provided with a *sic* [match probability 1:1 billion]. To show the difference between the LR and posterior odds, we can take as an example one over the UK population of males as prior odds. We see that here the posterior probability would be 0.97. The probability that he is not the source would be 3%, and not 1 in a billion.

$$\begin{aligned} &\textit{Posterior odds} = \textit{prior odds} \times LR \\ &\textit{Posterior odds} = \frac{1}{30 \times 10^6} \times 10^9 \\ &\textit{Posterior odds} = 100/3 \\ &\textit{Posterior probability} = 100/(100+3) \\ &\textit{Posterior probability} \approx 0.97 \end{aligned} \quad (7)$$

When a jury is provided with a figure of 1 billion, of course the evidence may appear to be compelling, supporting the prosecution case. It is well possible that in this example, the jury is being led down the path of findings biased interpretation because the word 'match' implies 'identity'. There appears to have been no advice in the judgement what to do with this figure of 1 billion. The word 'match' invites the prosecutor's fallacy as lay-persons can easily believe that a 'match' is synonymous to 'identification' [80]. There is the implicit risk of *confirmation bias* where evidence inconvenient to the prosecution is ignored or underweighted [81] and there is the associated danger that apparently compelling 'evidence' may be used to infer the *activity* (i.e. the act of touching the door handle). This will be described in detail in part II of our work. For a single full DNA profile where the assigned LR can be larger than a billion, prior odds would have to be extremely low to have any practical effect (ignoring relatedness effects and the probability of error) on the decision taken by the jury/court.

Debates on database searches periodically emerge in the literature. In a recent exchange, Nordgaard [82] asks: "*why does this debate keep re-emerging*?" and provides an answer: *"..the risk behind the fear is that the court would not use prior odds for the individual to be the source of the recovered DNA. If that is the case there would be no differences between a database hit case and a probable cause case with respect to the decision about guilt, if the DNA match is the only evidence presented. In other words a conviction would be solely built on the DNA match."*

Recommendation 9

It is crucial to outline that scientists do not give their opinion on who is the source of the DNA. There is a difference between the probability of the results given that the DNA is from an unknown person and the probability that the DNA is from an unknown person given the result. To equate one with the other is known as the transposed conditional, the prosecutor's fallacy, or the source probability error. It is thus important to explain what a likelihood ratio is and what it is not. This can be done by training or by providing a table with different odds, the LR and resulting posterior odds [75]. Because of the dangers of misrepresentation, it is essential to convey that scientists do not give opinions on the probability of propositions [25] and this is reinforced here.

7.3. Thoughts on prior odds

a) Before a case goes to court, the prosecuting authorities must decide if they believe that there is sufficient evidence to realistically support a prosecution. Consequently, the scientist could show the impact of prior odds to investigators in order to ensure that there has been a realistic consideration of the evidence as a separate exercise, before the case goes further. The scientist may aid investigators in the formulation of prior odds so they can pre-assess their case. However, once the case goes to court, the weighting of each kind of evidence and the combination of the various parts of the evidence to come to a decision of the guilt/innocence is the province of the court and not of the scientist. The scientist may advise on relevant considerations if so requested by the court.
b) One cannot overstate the importance of distinguishing the probability of the DNA results given that the DNA came from an unknown person and the probability that the person is not the source of the DNA. When there is no other evidence in the case, if the court does not consider the other evidence (or the absence of it) in the case then even with very powerful results, there is a high risk of miscarriages of justice. Another danger to avoid is that the strength of the DNA results given source propositions is not equal to its value given activity level propositions. The scientist should help explain the issue to the court by including a caveat in their statements.
c) The method of forming prior odds is the province of the court. Nevertheless, it may be useful to explain the impact the likelihood ratio on posterior odds (i.e., how to combine the information of the case and the information given by the DNA). This could be accomplished by furnishing a court with a table of several prior odds, the LR in the case and its impact on posterior odds for the case in question as described by [75]. The court could use the table to update its posterior odds based upon chosen prior odds. This would require careful explanation from the scientist – such a discussion would be invaluable when there was no other evidence in a case, other than DNA. However, the DNA commission notes that such a procedure has never been used in practice, other than for paternity cases. Because some cases are wrongful arrests that never proceed to trial it would be a useful exercise for investigators to be aware of the fallacy of the transposed conditional and be cognisant of prior odds and their effect.
d) Some courts (e.g. UK) have recommended prior odds to be formed on the basis of a population size (e.g. population of a city) [83]. Also see [84] for examples of statements in relation to Y-chromosome analysis. However, there is debate on how to decide which population to use and its size. The DNA Commission does not see a requirement to make a recommendation on how/whether to advise a court to assign prior odds as this is regarded to be outside the province of the scientist.
e) However, the DNA commission thinks it is important for *investigators* to be aware of the transposed conditional and this can be shown by the potential impact of prior odds on posterior odds. This way of thinking can also be useful when deciding whether there is sufficient evidence to prosecute a case – this process is separate to formal court proceedings (except for the inquisitorial court under direction of the judge). This is especially true for suspects identified solely as a result of database search, or where the probability of error has to be considered [85].
f) One should note that usually, the probability of a contamination is not taken into account by scientists when assessing their results. The assumption that results are error free must be disclosed both to investigators and to the Court.

Consideration 6

If DNA is the sole evidence in a case, then a suspect may be identified from a database search. If the investigation does not yield any other evidence, then investigators should be all the more

aware of the fallacy of the transposed conditional. To assign the probability of a proposition, they should take into account*prior odds and the DNA results*, in order to establish if there is sufficient evidence to prosecute a case. In court, it is not the remit of the scientist to assign prior odds. However, the scientist should explain to the court that they do not give an opinion on propositions.

8. Concluding remarks

This paper has focused upon the evaluation of results given subsource and source propositions. It has been assumed that the DNA process was error free, which we know is untrue. This aspect is not ideal, and makes it all the more important to underline that we only report on the value of the results given our assumptions. In the second part of these recommendations, we move up the hierarchy of propositions framework to the next level, providing considerations and recommendations in the field of evaluation of forensic biological results in the context of the *activities* alleged in the case. Activity level propositions are challenging, but are crucially important to consider because of transfer phenomena. We will give examples and present Bayesian networks as a means to deal with the complexity of real casework more comprehensively and in a transparent way. Methods of analysis are a well-researched and documented area, however, the *evaluation* of the results obtained from using these methods is often less well formalised. We advocate an experimental approach to enable assignment of probabilities in the context of transfer evidence. There may be a perception that such probabilities are less data-supported than probabilities derived for assessing the value of the comparison of DNA profiles. But, this does not mean that experts can solely rely on experience acquired in casework. Consequently, it is important to ensure that methods of evaluation are as robust as methods of analysis.

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