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# Relatedness and DNA: are we taking it seriously enough?

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**Abstract**

In forensic DNA testimony most DNA laboratories report the match probability for an unrelated person from some relevant population. These laboratories typically make available the match probability for relatives when requested. This practice has served well for many years. However, as the discrimination power of our multiplexes has increased the estimated match probabilities for both related and unrelated people have become markedly smaller. Associated with this general reduction in match probabilities have been the observations that the relative balance between the match probabilities of the many unrelated people and the few relatives of a suspect has changed. We suggest that we should now report routinely the match probability for a sibling whenever the suspect has a non-excluded sibling.


*Keywords:* DNA interpretation; Relatedness; Match probability

## 1. Introduction

Typically in forensic DNA testimony a sample of biological material associated with a crime is compared to a sample from a suspect. The genotyping results from the scene and the suspect are compared. If the suspect can be excluded as a contributor to the material at the scene then that is usually the end of the matter. However, if he cannot be excluded it is customary to provide some assessment of the strength of the DNA evidence. Historically this has been undertaken by assessing the probability of this evidence if the suspect is, indeed, not the donor of the stain. If the suspect is not the donor of the stain then we need to explain the evidence. Possible explanations include:

- a laboratory error,
- a relative was the donor, or
- an unrelated person was the donor.

In this paper, we consider the relative impact of the latter two explanations. Evaluation of these options is not original. We, rather, call for a re-examination of reporting practice in view of the evolution of modern multiplexes to a larger number of loci.

The first DNA profiles were termed ‘‘multilocus profiles’’ [1–3]. These were based on minisatellite repeat sequences. After digestion of the DNA radiolabelled probes were applied at low stringency that bound to the fragments from a large number of loci. The resultant autoradiographs looked somewhat like a bar code and this was an analogy that was commonly used. At the time (and subsequently) it was not know how many loci were involved or which bands were allelic or linked [1,2,4,5].

The next stage of DNA evidence involved the same restriction fragment length technology (RFLP) but the probes were applied at high stringency and were designed

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to visualise a single locus. These loci were still minisatellites. As expected, individuals typically showed only one or two alleles per locus. Laboratories originally implemented between three and nine loci for such tests.

The next evolution was the advent of PCR, coupled with the visualisation of STR loci. This is the method of choice today. Early multiplexes visualised three or four loci progressing through six loci and now many forensic laboratories implement multiplexes that visualise 9, 10, 13 or 15 STR loci and paternity testing laboratories may have more. These developments are expensive and involve high implementation costs, but are highly desirable from the point of view of discrimination.

At this point it is important to state that we should not get into a ''locus counting'' frame of mind. In court we have often experienced questions suggesting the more loci the better. Hence, why did we not use 10, 11, 12, or more loci? There are many factors that affect the utility of a multiplex and the number of loci is only one of them. Each locus may have differing polymorphisms and there may be other factors suggesting the inclusion of a particular locus in a multiplex system. These other factors may include such things as the availability of suitable primers, mutation rates, heterozygote balance, and performance under the compromise conditions of the multiplex.

Most DNA evidence is reported with a measure of the strength of the evidence. This measure is most often an estimated match probability or a likelihood ratio. These match probabilities or likelihood ratios are typically calculated for an ''unrelated'' person. This may be done using either the product rule or modifications that take account of subpopulation effects [6]. Sampling uncertainty may, or may not, be assessed for each case [7–12].

As more loci are added to multiplexes the various assumptions underlying the estimation of match probabilities become more difficult to test. Of note is that the number of assumptions of approximate linkage equilibrium within the population (when using the product rule) or subpopulation (when using Balding and Nichols' equations [6]) increases as a function of the number of loci. However, in general, as more loci have been added the discriminating power of the multiplexes is believed to have increased.

Occasionally the match probability for a relative may be requested by the defence and is duly reported. Sometimes close relatives are eliminated by genotyping. It is certainly not a novel suggestion that the effect of relatives should be assessed [6,10,13–19]. The formulae for match probabilities for siblings are quoted in Appendix A.

The question discussed here is whether or not the time has come to routinely report the match probability for a sibling as well as that for an unrelated person in all DNA casework. If decided upon, this change will involve some effort since the technology to implement such a suggestion for mixed samples exists but is not yet developed into user-friendly systems [20].

## 2. Results

It is necessary to consider by what criterion we may decide whether or not to include the estimated match probability for a sibling in addition to that for an unrelated person. Clearly, including an additional number makes an already complex evidential statement more so. There would need to be a good reason to include an additional number in a statement. It would seem reasonable to include this number if it was ''important'' for the decision making process.

How could we know whether or not the match probability for a sibling was important? David Balding has pointed the way clearly in his papers [14–16]. We take up his argument. As many multiplexes contain the amelogenin locus that allows sex determination we assume that the stain at the scene is from a known sex, say male. Hence, we confine ourselves to the consideration of male suspects. Consider a very simplistic view of the population of the US. Since we are restricting ourselves to males we model this population as containing the suspect, the single brother of this suspect, and 125 million unrelated males. Clearly, this model is simplistic but it allows us to make the necessary point most straightforwardly. Adding further relatives of the suspect, such as additional siblings, to the population simply strengthens the point.

Suppose that some non-genetic evidence has been produced or will be produced that suggests that the suspect is the donor of the stain DNA. The defence may also produce some evidence that suggests that he is not the donor. Such evidence affects the jurors' assessment of the probability that the suspect is the true donor of the material. Since this evidence is separate to the genetic evidence the probability based upon it is termed the ''prior probability'' even though it may not necessarily precede the genetic evidence. Typically this prior probability is unknown to the scientists. It is most likely to be a non-numerical subjective evaluation in the minds of the jurors.

There may also be non-genetic evidence for or against the other possible donors. These alternative donors are the single brother and the 125 million unrelated males in our simple population. There may be evidence that has eliminated some of these. They may have been genotyped or eliminated by other means. In more rare instances there may be evidence suggesting a specific alternative donor or donors. In all cases there will be information such as opportunity, ability, or access to the crime scene that may affect the probability that certain persons are donors. This information may be of such simple form as that small children or invalids are unlikely to have committed a criminal act involving physical strength.

It is important to consider any non-genetic evidence that points towards or away from the single brother of the suspect. For the purposes of this argument we assume that there is no evidence of this sort. If this assumption