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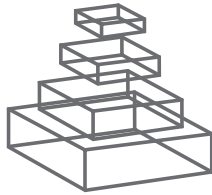
DNA, STATISTICS AND THE LAW: A CROSS-DISCIPLINARY APPROACH TO FORENSIC INFERENCE

Topic Editors

Alex Biedermann, Joelle Vuille and
Franco Taroni



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DNA, STATISTICS AND THE LAW: A CROSS-DISCIPLINARY APPROACH TO FORENSIC INFERENCE

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From ABO typing during the first half of the 20th century, to the use of enzymes and protein contained in blood serums and finally direct DNA typing, biology has been serving forensic purposes for many decades. Statistics, in turn, has been constantly underpinning the discussions of the probative value of results of biological analyses, in particular when defendants could not be considered as excluded as potential sources because of different genetic traits. The marriage between genetics and statistics has never been an easy one, though, as is illustrated by fierce arguments that peaked in the so-called “DNA wars” in some American courtrooms in the mid-1990s. This controversy has contributed to a lively production of research and publications on various interpretative topics, such as the collection of relevant data, foundations in population genetics as well as theoretical and practical considerations in probability and statistics.

Both DNA profiling as a technique and the associated statistical considerations are now widely accepted as robust, but this does not yet guarantee or imply a neat transition to their application in court. Indeed, statistical principles applied to results of forensic DNA profiling analyses are a necessary, yet not a sufficient preliminary requirement for the contextually meaningful use of DNA in the law. Ultimately, the appropriate use of DNA in the forensic context relies on inference, i.e. reasoning reasonably in the face of uncertainty. This is all the more challenging that such thought processes need to be adopted by stakeholders from various backgrounds and holding diverse interests.

Although several topics of the DNA controversy have been settled over time, some others are still debated (such as the question of how to deal with the probability of error), while yet others - purportedly settled topics - saw some recent revivals (e.g., the question of how to deal with database searches). In addition, new challenging topics have emerged over the last decade, such as the analysis and interpretation of traces containing only low quantities of DNA where artefacts of varying nature may affect results. Both technical and interpretative

research involving statistics thus represent areas where ongoing research is necessary, and where scholars from the natural sciences and the law should collaborate.

The articles in this Research Topic thus aim to investigate, from an interdisciplinary perspective, the current understanding of the strengths and limitations of DNA profiling results in legal applications. This Research Topic accepted contributions in all Frontiers article type categories and placed an emphasis on topics with a multidisciplinary perspective that explore (while not being limited to) statistical genetics for forensic scientists, case studies and reports, evaluation and interpretation of forensic findings, communication of expert findings to laypersons, quantitative legal reasoning and fact-finding using probability.

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DNA, statistics and the law: a cross-disciplinary approach to forensic inference

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Keywords: forensic DNA profiling, interpretation, probability theory, commercialization, DNA transfer, low-template DNA analysis, forensic molecular biology, bacterial DNA

The use of results of DNA analyses in the legal process is a highly ambivalent topic. On the one hand, scientists have never been in a better position to analyse biological matter of various natures, even in limited quantities and degraded conditions. On the other hand, the increasing amounts of scientific data that can be generated through modern analytical processes do not necessarily imply that evaluative questions that arise in the legal context are given more satisfactory answers. A fundamental question that has accompanied DNA analyses since the early days of their use in the legal process thus remains: how do we handle the challenges presented to us by the use of contemporary scientific and technological developments in the field of law? Under the general theme “DNA, statistics and the law,” the collection of articles in this Frontiers Research Topic pursues the goal of investigating this question from an interdisciplinary perspective, and with an emphasis on both current and future challenges.

As pointed out by Gunn et al. (2014) and Leake (2013), the forensic interest in DNA goes well beyond the standard approaches to DNA profiling that represent the current state-of-the-art in many contemporary legal systems, and this raises questions as to how new forms of data ought to be dealt with in an operational perspective (Milot et al., 2013). Although these frontiers topics clarify the extent to which there is room for exciting future research in this area, it should not distract us from the fact that even in the current state of forensic practice, there are hurdles and pressing topics that ask for efficient answers. Controversies over legal cases, such as the Perugia case (Vecchiotti and Zoppis, 2013), reveal that the field is still facing difficulties in setting the meaning of DNA profiling results appropriately into context (Champod, 2013; McKenna, 2013). One might be tempted to conclude that this is an issue that is confined to (and could thus be resolved within) the intersection between forensic science and the law. This perspective might, however, fall short of further dimensions, such as commercialization (Jackson, 2013). The publication of opinion pieces on this topic helps raise awareness on this topic and address some of this deficit.

On a methodological account, the field of statistics is often invoked as a remedy to deal with evaluative questions and many discussants tend to emphasize its traditional facet concerned with data processing. The case of statistics is more general, however, because it is a branch that involves an additional characterizing feature: reasoning coherently in the face of uncertainty (known in the context as *forensic inference*), using probability theory. Indeed,

existing literature abounds in rigorous and coherent approaches to cope with intricate evaluative questions (Biedermann, 2013; Juchli, 2013) of the kind that are also encountered in connection with forensic DNA. It is with some frustration, however, that we note that discussions surrounding evaluative questions, using probability, are still fraught with problems that have debates for a very long time. Prior probabilities are one example for this (Thompson et al., 2013).

In summary, the contributions in this Research Topic convince us that the extension of technical frontiers should also be accompanied by conceptual developments and understandings. Indeed, during personal discussions with the Topic Editors, one reviewer (Sheila Willis, Eolaíocht Fhóirínseach Éireann, Forensic Science Laboratory, Ireland) raised cultural understandings as a further relevant factor: “I think the problem is much deeper. The use of matching DNA as a heuristic for a definite link between person and place is embedded in the minds of scientists as well as jurors in spite of the scholarship to the contrary. The discriminating power of DNA has had a paradoxical effect in the development of forensic science. On one hand it prompted forensic science to be valued and used in a very widespread manner but on the other hand it promoted the commodification of forensic science with the belief that the test result is all-important and the context irrelevant. This latter view prompts the approach that the test can be produced anywhere and loses sight for the need of the very evaluation (...). It is vital that we address this. It is mixed with the commercialization issues but to focus too much on that aspect is to ignore the wider issues that also need to be addressed by: the publication of high profile cases where this approach has unfortunate consequences; increased education; critical mass of scientific opinion in favor of the approach argued for (...).”

We cannot but agree and hope that the collected papers in this Research Topic will be of interest to both scientists and other participants in the legal process. We thank all contributors and distinguished reviewers for their efforts to make this original collection timely and highly useful.

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DNA and the law in Italy: the experience of “the Perugia case”

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Today DNA analyses represent a method of exceptional importance for the resolution of judicial cases. On the one hand, they allow courts to secure criminal convictions, while on the other hand they can help exonerate innocent suspects. Unfortunately, DNA analyses are often considered an unbeatable and infallible method to discover the truth, with the consequence that judges feel forced either to “bow to science” or to totally refuse the genetic evidence when it is considered too complex. On the contrary, genetic investigations have limits that must always be considered and properly explained to the fact-finder by the forensic geneticist. Courts need to know what results were observed and how likely it is to observe such results under both the prosecution and defense hypotheses. This may be particularly challenging for low quantity, degraded or mixed genetic material, and is further complicated by the need to take into account the potential of (laboratory) error. Despite such circumstances, the evidence can still be informative although its probative value may be reduced.

The murder of British student Meredith Kercher in Perugia (Italy) in 2007 and the case that ensued have highlighted the limits of genetic analyses. Throughout Italy, this case has caused an intense scientific and (through the media) popular debate on the correct application of internationally recommended protocols and procedures as a preliminary quality and reliability guarantee for results presented in court. Particular attention has been drawn to the interpretation of genetic profiles derived from Low Template (LT) or Low Copy Number

(LCN) DNA and mixed samples. The two defendants, Amanda Knox and Raffaele Sollecito, were convicted after the first trial but then acquitted on appeal in 2011. The Italian Supreme Court overturned the acquittal in 2013, and a new trial will be held soon.

The Appellate Court experts (author Carla Vecchiotti was one of the two experts who reviewed the case for the Court of Appeal) were asked to repeat, if possible, the genetic analyses carried out during the initial investigation on certain items and whose results led to the conviction of the two defendants: a knife, considered by the prosecution to be the murder weapon, and a bra clasp belonging to the victim. If a repetition of the analyses was impossible due to insufficient biological material, the experts were asked to examine the technical report drawn up by the scientific police in the course of the first trial. According to this document, the scientists had observed DNA profiles corresponding to the victim on the knife blade, to the defendant Amanda Knox on the knife handle and to the defendant Raffaele Sollecito on the bra clasp. The report also concluded that the correspondences between the traces and the various people involved meant that these people were the source of the DNA in question.

As for the knife, collected from the inside of a drawer in Sollecito’s kitchen, the Appellate Court experts found neither traces of blood nor the presence of cellular material on the blade. The quantification analysis performed on the material collected from the blade provided a value of 5 pg/ μ l just in one sample, a result far below the value recommended

in the technical protocols of the new generation commercial kits for STR analysis (i.e., 0.25–0.5 ng of template DNA in the PCR reaction in a maximum input volume of 17.5 μ l for the PowerPlex® ESI 17 and ESX 17 System; 1 ng of template DNA in the PCR reaction in a maximum input volume of 10 μ l for the AmpFISTR® NGM Select™ PCR Amplification Kit). Since the amount of extracted DNA would not allow the required repetition of amplification, the Appellate Court experts decided not to proceed with the genetic analyses on the swabs taken from the knife (Butler and Hill, 2010). As for the bra clasp, it was recovered and collected from the crime scene floor 46 days after the murder. It could not be analyzed by the Appellate Court experts as it had been stored by the scientific police in a tube containing extraction buffer, which made it completely rusty. Consequently, the Court experts proceeded to examine the above-mentioned technical report in order to evaluate the results obtained from the analysis of the two items.

The knife was examined first. According to the technical report, the two samples of interest were sample A, taken from the handle, and sample B, taken from the blade. Regarding the nature of the recovered material, there was no scientifically conclusive evidence to support the possible blood nature of the sample taken from the blade (sample B) in that both the generic blood test and the human species test were negative. The conclusion that exfoliated cells were present on the sample taken from the handle (sample A) was equally lacking in scientific basis. No reliable method for quantifying the DNA

was employed, and the quantification performed with the Qubit Fluorimeter™ gave the result “too low” for sample B (knife blade), indicating a DNA amount below the sensitivity threshold of the Fluorimeter (200 pg/μl); therefore, presumably, a LT-DNA sample. In relation to the same sample B (knife blade), the electrophoretic graph showed peaks far below the 50 RFU threshold and allele imbalance ($H_b = \varphi_a/\varphi_b > 0.60$) for most of the alleles, thus indicating a LT-LCN sample. Yet, none of the recommendations issued by the international scientific community and aimed at obtaining scientifically reliable results when treating this challenging kind of samples were followed. Replicate analyses could have been performed at the time, although experts’ views on how to analyze LT-DNA have been evolving since then. The main issue with that type of samples is contamination: consequently, strict protocols must be applied during the inspection, collection, and sampling of such items at the crime scene (Giardina et al., 2011). The procedures recommended to reduce laboratory contamination are equally rigorous as it is well-known that contaminant DNA at low levels may derive from reagents and other laboratory consumables, from the technical staff and from cross-contamination from sample to sample. Indeed, in the context of the Kercher murder case, transfer of a suspect’s DNA into a crime scene sample was of particular importance: in fact, it appears that crime scene inspection procedures destined to minimize contamination were not carried out according to international protocols (Fischer, 2003; Laboratory Division of the Federal Bureau of Investigation, 2007; ICPO-Interpol, 2009). Furthermore, it seems that no attempts were made to discover such events.

As for the bra clasp, regarding the nature of the material recovered, there was no scientific evidence supporting the notion that flaking cells were present in the sample. The hypothesis formulated by the scientific police technical consultant about the nature of the material collected from the clasp is thus arbitrary, since it was not supported by any actual findings. After examining the electropherograms obtained from the autosomal STR analyses, the Appellate Court experts were able

to assert that, for the markers D8S1179, D21S11, D19S433, D5S818, allelic peaks were interpreted in a manner that did not conform to the recommendations made in current literature/practice. In particular, peaks were considered to be stutters whose heights were above 50 RFU (D19S433), exceeded the threshold of 15% of the major allele (D8S1179, D21S11, D5S818), or were not in a stutter position (D5S818), and thus should have been considered to be alleles (Gill et al., 2006). The DNA extracted from the bra clasp thus indicates the presence of several minor contributors, which was not disclosed by the scientific police. The electropherograms obtained from Y-STRs analysis also showed (besides the peaks designated as alleles in the technical report of the scientific police) the presence of additional peaks with heights exceeding the threshold of 50 RFU (Table 1). Despite not being in a stutter position, they were not taken into consideration. Instead, the report(ing) was limited to what was in agreement with the observations on the electropherograms of the autosomal STRs. The genetic profile thus derived from a mixture of unidentified biological substances, whose larger component corresponded to the profile of the victim and whose smaller components suggest the contribution of several male sources. Defendant Raffaele Sollecito showed a profile that was compatible with the profiling results for the trace found on the bra clasp. However, considering the particular circumstances under which the item was recovered and collected, it could not be ruled out that the results obtained from the analysis of the bra clasp derived from environmental contamination and/or contamination in some phase of the collection and/or handling of the item.

In conclusion, it is important to highlight some relevant issues concerning the interpretation of genetic profiles obtained from LT-LCN DNA and mixed samples. First of all, interpretation of a profile obtained for a particular item that is deciding which electrophoretic peaks are allelic and which are stutter or other artifact, must be done without reference to the suspect’s profile: it is the only way to minimize the risks of bias in the interpretation of the profile derived from the evidentiary samples. Interpreting a profile derived from a

sample with the suspect’s reference profile at hand conflicts with the principles of scientific integrity, balance, and coherence that should underlie the practice of forensic science (Budowle et al., 2009; Thompson, 2009). It is also clear that the weight of the evidence is a fundamental issue (Gill and Buckleton, 2010), as widespread public opinion holds that if DNA found on the crime scene matches the suspect, then he must be guilty of the crime. This logically wrong understanding unfortunately also extends to a considerable number of scientists, judges, and lawyers. In fact, there is a perception that failure to convict implies a failure of science. Such a view is extremely dangerous and it is therefore important to defend the idea that whether or not a suspect is convicted is an irrelevant question for the scientist, whose responsibility must only be to correctly explain the evidence in the context of the specific case. The question of how DNA corresponding to the suspect was transferred onto an item must therefore be assessed by the judge and not by the scientist, whose role is limited to presenting the various ways in which transfer can happen and the strength of support for each of the various scenarios (Gill and Buckleton, 2010).

In Italy, the Kercher case has defined a new way of conceiving of and addressing the scientific evidence in the context of a criminal trial (Montagna, 2012): the scientific and, subsequently, legal quality of the investigations performed at the crime scene depends on the compliance with internationally standardized procedures. There is now a better awareness of the importance to follow correct crime scene procedures in order to minimize the risk of contamination and, subsequently, the loss of reliability of any results obtained. Another element that has emerged during this debate is the increased awareness, in the international scientific community, of the need to develop structured reasoning models. These should assist in the evaluation of propositions according to which the suspect is or is not one of the persons who contributed to a particular mixed biological trace, in particular in the context of LT-LCN (including additional phenomena such as drop-in, drop-out, etc.). Finally, it is worth recalling a key principle of the

