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Opening the DNA black box: demythologizing forensic genetics

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Social impact is intrinsic to any applied branch of genetics. Since the goal of forensic genetics is to provide expertise in legal disputes, the expert should make her- or himself understood. The urgent need for measures to deepen the communication between experts and non-experts requires debunking of some myths surrounding forensic genetics, and to (re)center the discussion on the solid ground of formal genetics. “Classical” forensic sciences rely on the assumption of discernible uniqueness, while forensic genetics deals with types of observations. It computes expected frequency values for the observations using empirical estimates within a theoretical framework, allowing the evaluation of the probabilities of the same observation under alternative, mutually exclusive and exhaustive hypotheses. Consequently, the interpretation of DNA evidence entails fewer risks of error than classical forensic evidence. However, clear regulations and a total separation between the institutions performing criminal investigation and those acting as expert witnesses are required.

Keywords: forensics; genetics; DNA

Introduction

The legal and criminological implications of forensic and police uses of DNA profiling have been under increasing focus within the last two decades. DNA-based evidence has gained an unprecedented degree of trust, but at the same time given rise to a proportionally strong sense of fear (Duster 2006). This paradoxical situation results from two characteristics of DNA technology: an allegedly unlimited evidential power, along with an also allegedly high level of sophistication and complexity which is commonly thought to be inaccessible to non-experts. DNA has become a “black box” in the double meaning of the phrase: it has assumed the role of the ultimate and indestructible recorder of our “secret” genetic code, and the role as an impenetrable and incomprehensible truth machine (Lynch et al. 2009).

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Accordingly, the purpose of this paper is also twofold: to demonstrate that (1) the results of forensic DNA testing are limited in both theoretical and practical terms; and (2) it is indeed both possible and necessary for non-experts to obtain a basic – albeit sufficient – understanding of its principles and methods. Obviously, such an understanding is a prerequisite for the adequate use of the DNA expertise in court, but it is arguably also very pertinent to the work of scholars and researchers looking at the societal dimensions of forensic DNA technologies. The unprecedented speed of the development of the field is not a sufficient excuse for the lack of available authoritative reference material dedicated to the public understanding of this applied science. Moreover, a status of undisputed power and exceptionality has been granted to DNA, precluding the promulgation of an analysis of its limitations.

For such an analysis it will be necessary to debunk some of the myths surrounding forensic DNA testing through a brief analysis of its foundations in formal genetics. The profound differences between “classical” forensic sciences and forensic genetics will be discussed as well as the probabilistic framework involved in the transmission of information contained in the results of a genetic profiling.

Genetics and forensics
An excellent definition of forensic genetics can be found in the official journal of one of the leading international scientific societies devoted to this field (Anon 2007, p. 1), Forensic Science International: Genetics: “The application of genetics to human and non-human material (in the sense of a science with the purpose of studying inherited characteristics for the analysis of inter- and intra-specific variations in populations) for the resolution of legal conflicts.” It is self-evident that the results from any forensic expertise have no meaning outside theoretical and technological frameworks; therefore a brief digression into these foundations is mandatory. For a start the essential theoretical genetic bases will be outlined before attempting to analyze the forensic applications and current technologies.

Genetic theory
When the genetic theory was advanced almost 150 years ago (Mendel 1866), it was so revolutionary and against common sense that it was (partially) rediscovered only in 1900 and still today defies our intuitions. The theory’s essentials can be formulated in a simple set of sentences:

- Genetic information is discrete.
- Each individual possesses two copies of the central unit of genetic information.
- Out of these two copies, just one is randomly transmitted to each offspring.

Modern developments of the theory are quite complex, particularly at the population and evolutionary levels. In the context of this paper, only their main consequences for the relevant forensic applications will be considered.
First, the fact that genetic information is of a digital kind results directly from the chemical nature of its repository, the deoxyribonucleic acid, or DNA, composed by a variable sequence of units, symbolized by the letters A, T, G and C. It implies that genetic differences between individuals, if properly analyzed, are also digital. In other words, the genetic differences at this level are of kind and not of degree; hence terms like phenotype or genotype. This concept implies that at a particular location the genetic sequence of an individual will be defined by a pair of occupation states out of a very finite set of possible choices. In the simplest case, the sequence would be either A/A, A/T, A/G, and so forth, each of these pairs symbolizing a genotype. In this context, there are therefore no unique individuals: everyone belongs to a class shared by many others – at least potentially, depending on the frequencies of A, T, G and C. Thus, individuals cannot be distinguished from each other at this level of analysis. The same reasoning applies to other characteristics exhibited by each of us, and that is why we are said to belong to blood group A or B and those classified into the same group behave in the same way in respect of transfusions, since they share the same phenotype. If we accept this fashion of analyzing genetic and biological properties it is not difficult to extrapolate that they do not provide individualization: they just allocate each of us to classes. To achieve a satisfactory probability of distinguishing any pair of individuals, a good number of positions in the genetic material must be typed in order to generate a profile, for which the population diversity has been previously found to be reasonably high. The problems of finding and typing such genetic markers will be discussed in the following section, where the application of the genetic theory to forensics will be analyzed.

**Forensic genetics**

Well before the advent of genetics, many sciences have contributed to forensics (James et al. 2009). However, forensic genetics enjoys an essentially distinct epistemological status among all other sister disciplines (Saks and Koehler 2005). Indeed, and quoting these authors, all traditional forensic sciences rest on a central assumption: that two indistinguishable marks must have been produced by a single object. [...] According to this assumption [of discernible uniqueness], markings produced by different people or objects are observably different. Thus, when a pair of markings is not observably different, [...] the marks were made by the same person or object. (Saks and Koehler 2005, p. 892)

As explained above, genetics proceeds in a way that is quite the reverse: it does not rely on the uniqueness assumption; in contrast, it deals with more or less frequently found types of observations and is able to calculate expected values using empirical estimations within a theoretical framework.

Let us try to substantiate these assertions (for a somewhat different perspective, see Edmond 2011). When comparing two classical fingerprints – or bullets, photos, etc. – a wide range of techniques is used, delivering a series of measurements...
obtained in both. These results, along with visual inspection of the evidence, are then contrasted in a rather poorly standardized fashion, and the expert, also based in his/her empirical experience issues an opinion – very seldom quantified – on their possible identity. In consequence it is not surprising to sometimes encounter opposite expert opinions on the same evidence, as well as to observe a frighteningly high level of error rates, not significantly different from those observed among untrained eyewitnesses’ testimonies (Saks and Koehler 2005).

Conversely, forensic genetics does not seek individually distinctive features in a genetic profile, as shown in the previous section; the topic will now be developed in more detail materializing a few technical aspects.

Although forensic genetics has been around since the discovery of ABO blood groups (Landsteiner 1900) a new impetus was gained with the mastering of technologies able to directly disclose differences in DNA sequences (for a review accessible to non-geneticists, see Jobling and Gill 2004). All validated routine forensic genetic analyses currently employ a preparative technique: polymerase chain reaction (PCR), which specifically detects and amplifies short regions of DNA from our genome. The regions that are currently used in this field – and constitute the information deposited in DNA databases for forensic purposes – have been selected according to two criteria: (1) to correspond to non-coding tracts – i.e. there is, to our knowledge, no information contained there and so no physical or psychological characteristics of the individual can be inferred from its analysis; and (2) to be polymorphic – that is, the DNA sequence is quite variable between individuals. They are named short tandem repeats (STRs) and contain a variable number of tandemly repeated motifs (like GATA). Using as an example one of these regions, D3S1358, included in the FBI Combined DNA Index System (CODIS), an individual can be typed as 16–19. This would simply mean that s/he has inherited from one of his/her parents a stretch of DNA containing 16 repeats in that particular region, and from the other person a stretch containing 19 repeats in the same locus. Since a large series of validation studies preceded the introduction of this system in routine analyses, estimates of the corresponding frequencies are already at hand. Suppose for instance that the 19 repeats variant (which our fictitious person inherited from parent A) has a frequency of 1% (1/100); genetic theory predicts that genotype 19–19 is expected to be found once for each 10,000 individuals (assuming random mating, 1/100 × 1/100 = 1/10,000); the probability of finding such a genotype in a pair of randomly chosen individuals drops to 1 in 100,000,000 (1/10,000 × 1/10,000). Both predictions can be tested against real data (assuming the database is large enough).

How far are we from the “discernible uniqueness” assumption required for the other forensic disciplines? Not only is uniqueness not required, but indeed the opposite property is used – genetic profiling is always a classification process, the assignment of an individual to a group. Of course, if just a few of these STRs were typed in a given individual, then the specific genotype constellation turns would be, in practical terms, almost unique, because of the statistical
improbability of finding different (i.e. unrelated) people who have the exact same genetic variants at the same loci. For instance, the combination of all 13 CODIS STRs provides a matching probability – i.e. the chance of finding two random individuals with the same profile – of less than one in a trillion (Butler 2005).

It must be said that all these suppositions were made assuming no technical difficulties in the classification (phenotyping) process itself. This assumption is only partially valid: from the simple situation outlined above, described as using a very robust genetic marker on non-problematic samples, to more challenging scenarios of degraded and/or vestigial traces\(^1\) and less validated markers or techniques, a huge variation in the confidence of the results exists in forensic practice. To illustrate this, I quote from a published analysis of the results of a collaborative exercise undertaken by a large number of forensic laboratories to improve the quality of DNA typing:

A plethora of different interpretations [of the mitochondrial DNA evidence in the simulated forensic cases emulating the work carried out by the laboratories in real forensic situations] were used in this part of the exercise. This is to some extent expected and reflects the lack of standards and consensus among forensic geneticists. (Prieto et al. 2008, p. 129)

Forensics implies dispute, conflict, a difference of opinion, which formally translates into the existence of (at least) two alternative explanations for the same fact, i.e. for the concordance of two DNA profiles. Recall, as just shown above that this “fact” (supposedly established beyond doubt) is indeed a scientific construction, with all the inherent complexities and limitations. Nonetheless let us assume that both defense and prosecution agree on the admissibility of the evidence, i.e., no doubts are raised on the validity of the genetic profiles generated from both the crime scene sample and the suspect. Classically, in legal proceedings, we say that the admitted evidence is explained to the court as (1) originating from the suspect (the prosecution hypothesis); or, alternatively, according to the defense (2) resulting from the action of someone else. How can the genetic expertise contribute to reaching a decision? In order to understand this process, a short digression into the probabilistic evaluation of genetic evidence must be undertaken.

Probabilities and quantification of hypotheses’ likelihoods

All readers must have seen staggering figures, like those mentioned above, labeled as “probability of identity” or “probability of paternity.” In my opinion this wording does not do justice to what is indeed the result of the genetic expertise and in fact entails serious misunderstandings.

The (re)definition of some notions pertaining to the world of mathematics, yet also used in common language, is required. The first is the concept of probability itself. The probability of a specific event is commonly and simply defined as the frequency of that event. In more formal terms, probability of an event is the ratio of the number of cases favorable to it, to the number of all cases possible.
It is a convenient way to summarize quantitatively our previous experience on a specific case and allows us to forecast the likelihood of its future occurrence. This is obviously not the issue when we move into the forensic scenario – the event has occurred (both litigants agree upon that) but there is disagreement on the causes behind it. So it seems that the same event can have different probabilities pertaining to its causation. To make it clear, let us suppose that I promise to draw an ace from a deck of cards and I succeed in doing so. The reader can look upon this outcome under two alternative and mutually exclusive explanatory hypotheses: (H1) I cheated; or (H2) pure luck. This is because my drawing an ace from a deck of cards upon my first try is a statistical improbability. Can we calculate the probabilities of the event assuming each of these hypotheses? Indeed we can: the probability of the observation assuming I was cheating (P|H1) is one, while the probability of the observation assuming random chance, or luck, (P|H2) is 4/52. If we agree upon this reasoning thus far, it is then possible to compare the two probabilities, building up a likelihood ratio, $L = P|H1/P|H1 = 1/(4/52) = 52/4 = 13$ and state that it is 13 times more likely to observe that outcome if I was cheating rather than achieving it by sheer chance.

The quantification of the genetic evidence in forensics is carried out in the same way; this method has gained unanimous acceptance among experts around the world (Gill et al. 2006, Gjertson et al. 2007; see also: Amorim 2008, Pinto et al. 2010). Let us suppose that a biological sample, such as a hair, organic fluid, etc., which does not originate from the victim, is found at a homicide scene. When typed for the D3S1358 marker mentioned above, this sample shows the genotype “19”; this is also the case for a suspect, the provider of a “reference sample.” It has been shown that the probability of finding such a genotype by chance is 1/10,000. Therefore, under the prosecutor’s hypothesis (the crime scene sample was left by the suspect), the probability of this type of observation (P|H1) is 1/1000, while assuming the defense explanation (the crime scene sample was left by someone else) the probability of same observation (P|H2) would be 1/10,000 × 1/10,000. Therefore, the likelihood ratio takes the value of 10,000, which means that the occurrence of this observation (obtaining this evidence) is 10,000 times more likely if both samples have originated from the same individual rather than resulting from two distinct persons, provided that the suspect does not have an identical twin.

Unfortunately this likelihood ratio is often referred as “probability of identity.” I hope to have made clear that this final value is not a probability in the strict sense, but a ratio of probabilities. The same happens when we state that a certain land area is 10 times larger than a second one: in neither case are we specifying the acres of each one, nor does the value 10 have units; it is dimensionless. Therefore, the likelihood ratio, as a comparison between probabilities is not a property of the examined individual and just translates the relative likelihoods of an event – the evidence – when explained by two alternative, exhaustive, mutually exclusive hypotheses. It is important to note that these conditions are not met in
the case where the suspect has an identical twin, since they are different persons albeit genetically indistinguishable.

Another caveat is related to the over-interpretation of results of genetic analyses. If in a specific case the genetic expert obtains a very high likelihood ratio, this result cannot be squarely translated into a seal of conviction. In fact, the samples might well have been produced by the same individual but the crime scene sample could have been left there under circumstances having nothing to do with the crime. Quoting Lynch et al. (2009, p. 191), “DNA evidence is meaningful only when it is embedded in stories that mention other evidence, possible suspects, and how the evidence itself was handled and interpreted.”

This issue is indeed of major importance, as the interpretation and the quantification of the evidential value of the results are sources of disagreement between genetic experts themselves. These problems are quite well described in the following quotation, from a review of the results of a series of proficiency testing programs undertaken for quality control in forensic labs: “The different theoretical tests proposed in [the] 2007 exercise generated the largest discussion. […] Results dispersion makes [it] difficult to establish a consensus […] value and evidenced different statistical procedures” (García-Hirschfeld et al. 2008, p. 676).

To put it bluntly, even if we assume that forensic DNA analysis was entirely free from technical difficulties or errors, the question of how to correctly interpret and evaluate non-trivial situations is a contested issue among experts. The result is that in a great number of cases, no consensus can be reached. The reasons for this are twofold: first, the high rate of technical vs. interpretation errors, and second, the experts’ own perceptions of their weaknesses and educational deficits. For the first, the experience from quality improvement programs has shown that while the rate of non-conformity of typing results is, on average, below 1% depending on the markers employed, the proportion of reports involving deviations from the correct statistical value reaches the worrying figure of 20% (García-Hirschfeld et al. 2008). The same source of information, corroborated by specific enquiries consistently reveals that “statistics” is by far the most required continuous education topic by forensic genetics practitioners.

**Investigative genetics**

Until this point just one of the facets of the interplay between genetics and justice has been the focus: geneticists were supposed to enter the judicial system only as expert witnesses, implying that the evidence was produced and carried to the court by different players. There is, however, another role, played many times by the same actors: criminal investigation. This side of the forensic application has gained increased importance due to the informative power of genetic analyses, which have been shown to be capable of identifying suspects in the absence of any other evidence. Such capability has been the basis for the public’s acceptance of DNA databases, when demonstrated in particularly shocking crimes.
The ambitious scope of current and future applications of genetics is well portrayed by a recently launched journal, *Investigative Genetics*, which aims at the development and application of molecular genetics in a wide range of science disciplines with societal relevance (Kayser et al. 2010).

In sum, even if we cannot predict how the field will develop in the future, we can say that the overlap between the two roles – crime investigation and expert witness – carries with it the seeds of contention: this overlap makes it difficult to comply with the deontological requirement of the independence of the expert, an especially serious danger in non-adversarial legal systems. Individual and institutional concerns (Jamieson 2010) have been mounting and have surely contributed to a dramatic revision – i.e. the abolition – of the status of the UK Forensic Science Service (Budowle et al. 2011). I will conclude this section with the presentation of one other source of dissent: the claimed capability of predicting ancestry or physical characteristics of the donor on the basis of his or her DNA (phenotypic profiling). Even if the prediction were to be accurate (and some experts dispute the validity of any claims to accuracy, remarking that many external characteristics can easily be modified, despite the genetic background by the use of simple cosmetics) the possibility raises many legal and ethical problems. Indeed, it is explicitly forbidden, in many jurisdictions, in accordance with European Council Resolution 2001/C 187/01, of 25 June 2001, and is usually abhorred by ethical committees. Moreover it contradicts the initial assumptions on permissible markers in DNA databases and risks a serious setback in the public opinion and political acceptance of forensic genetics (Enserink 2011a, 2011b). In summary, phenotypic profiling entails the disclosure of two types of information inferred from DNA analysis alone: the reconstruction of genetic profiles of the donor’s relatives and physical characteristics associated with ethnicity. Concerning the inference of genetic profiles of relatives from the profile of someone already included in a database (so-called familial searching, or genetic proximity testing), besides theoretical and technical problems a major ethical and legal issue arises: genetic information is obtained for persons who do not meet the legal inclusion criteria in the database and have not been consulted for consent. The inference of physical characteristics poses no less serious problems: on the one hand the correlations are weak – i.e. the inference is far from perfect and not transferable across human populations, as has been proved for skin color in Europeans and Asians (Norton et al. 2007), and easily modified (surgically or by simple cosmetics), but moreover, those traits are ethnically associated, risking the creation of bias in crime investigation towards minorities.

**Concluding discussion**

A recent study has shown that understanding of likelihood ratios by jurists was “quite poor,” but also that forensic experts “made many mistakes themselves” (de Keijser and Elffers 2012, p. 205). Moreover, the same study (in a Dutch
context, but I have no reason to believe the situation is restricted only to the Netherlands) has revealed a very pessimistic attitude among all types of participants in the analysis: 57% of defense lawyers, about one-third of the judges and 42% of experts think that adequate communication between experts and the courts is not feasible, the dialogue between them being characterized by gaps and misunderstandings, due to the large difference in professional expertise and frame of reference.

One of the purposes of this paper was to contribute to bridging these gaps. I hope to have shown that DNA cannot be treated as a “black box,” just the opposite: (1) genetic evidence evaluation can be made comprehensible to non-experts; (2) genetic experts must present their results in such a form; and (3) genetic experts should be ready to discuss and explain their results to non-experts. Both parties should make an effort to achieve these goals: geneticists must leave the comfortable opacity which precludes open debate and criticism, and non-experts are also responsible for first, avoiding the analysis of, and making the decisions upon, information they do not understand, and secondly, obtaining the necessary understanding when such analysis and decision-making are required. A concerted action on this last point is strongly recommended, with professional associations and universities being key players. The non-compromising stance of (some) experts, claiming that they can only “respond to the questions put to them by the lawyers” and thus “if the wrong questions are asked, the situation is difficult to rectify” (Gill 2009, p. 34) is unacceptable: it is not the role of the expert to judge the questions as “wrong” (at most ill-posed or scientifically unanswerable), nor there is anything to “rectify.” The temptation to usurp the judicial power and replace it by authoritarian “science” still lurks: “the controversy of DNA profiling is rooted not in the science, but mainly in the restrictions of the adversarial system” (Gill 2009, p. 34).

It is obvious that the diversity of judicial systems also implies very heterogeneous interplays between experts and courts. In particular adversarial systems involving jurors with diverse backgrounds and literacy levels pose serious problems to the materialization of the informed understanding of the genetic evidence. The context of the adversarial system is indeed the one where a steady effort to improve the understanding of expert witnesses’ reporting on the part of non-experts seems to be more important. However, inquisitorial systems are also not immune to the problems stemming from difficulties in the understanding of expert witnesses’ reports, as judges, prosecutors, and lawyers are also non-experts in genetics.

The urgency of measures to improve such understandings, and to strengthen and deepen the communication between geneticists and other actors working within the criminal justice system, is underlined by the widening of societal implications resulting from recent advances in forensics. For example, the developments in the field of investigative genetics are leading to an increasing number of cases where all the evidence presented to court is exclusively DNA based: first the suspect is identified solely through genetics (as through a DNA database familial searching) and then the only evidence available connecting him/her to the crime rests upon DNA profiles (for examples, see Gershaw et al. 2011). It must be
remembered that admissibility of DNA as sole source of evidence is (still) explicitly ruled out in some jurisdictions; some legal systems, however, have recently been modified to accommodate some of the new developments in forensic genetics and genomics (Enserink 2011a, 2011b).

It will be difficult to make up for the time lost meanwhile, but as I hope to have demonstrated, “classical” genetic profiles (involving non-coding DNA) are a less “risky” kind of evidence than classical forensic evidence, such as photographs or dermal prints. In fact, standard DNA profiles are much less invasive of individual privacy, being at the same time much more reliable and – still more important – in virtually all jurisdictions legally regulated, externally supervised, and controlled.

There is a great variety of legal frameworks among countries; when looking in the field of regulation pertaining to DNA databases alone, it becomes clear that a wide range of solutions has been implemented (as discussed in Hindmarsh and Prainsack 2010). In addition, practices more than often diverge from the initial legal frameworks and constraints (Dahl and Sætnan 2009, Machado and Silva 2009). In any case, insufficient attention has been paid to the conflict of interests potentially raised by the twofold role of most genetic experts and their institutions: acting as analysts of investigative crime scene samples and as expert witnesses. The fact that they can simultaneously perform crime investigation – including the selection of a suspect, and act as expert witnesses – issuing an opinion on the suspect’s claimed innocence, is an obvious source of conflict. These problems are further aggravated in countries where just one institution is legally entitled and allowed to perform both roles. I suggest an investigation of the implications of this double role in the administration of justice is necessitated by the points raised in this paper, and strongly recommend the adoption of legislation preventing this duplication. In brief, those (persons and institutions) in charge of investigation (producing evidence, guiding the search, suggesting a suspect to the prosecution) should be separated from those who, acting as expert witnesses, independently evaluate the evidence.

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Note

1. Genetic profiling of evidence containing very low amounts of DNA (the so-called LCN, low copy number DNA) has been particularly vulnerable to criticisms and hot debate among the experts’ community (see, for instance, Gilbert 2010).
References


