God’s signature: DNA profiling, the new gold standard in forensic science

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In the mid-1980s, when the first DNA profiling techniques were developed, the name DNA ‘fingerprinting’ was widely used. At the time, fingerprinting was a well-established forensic method, and it was rarely questioned in the courts. Fingerprint examiners were permitted to describe matching prints as evidence of individual identity, and they were not required to give probability estimates. Despite its nominal association with the older technique, DNA ‘fingerprinting’ went through a period of controversy, especially in the US courts. The association with fingerprinting was questioned, and experts were required to qualify their testimony with probability figures. Heated debate occurred in scientific publications and law courts about the statistical and population genetic assumptions that went into the probability calculations presented in court cases. However, by the late 1990s DNA profiling was so widely accepted that it became a basis for invidious comparison with all other forms of forensic evidence, including fingerprinting. In the past three years, the admissibility of fingerprint evidence has been challenged in several US federal and state courts. This article discusses the socio-legal and socio-technical issues that led to the inversion of credibility that characterized the intertwined history of the two techniques.

That’s God’s signature. God’s signature is never a forgery [1].

Therefore the results must be flawed until someone proves to me otherwise.

Dr Edward Blake of Cellmark Diagnostics, the company hired by Mr Godschalk’s defense to perform the tests, expressed no such doubts about the scientific evidence. He likened the DNA profile result to a signature, which ‘is expected to occur in no more than a few human beings who have ever lived’. Another expert gave an even stronger account to the press, saying that the matching profiles developed from the crime evidence could only come from one person in the world. Still another expert contacted by defense lawyer Peter Neufeld stated that the chances of laboratory error in this case were ‘nonexistent’.

Who (or what) do we trust?

Among several issues raised by this case is that of trust: who, or what, should a court trust when making a decision of great consequence for the fate of an individual and the safety of the community? Although they have been hesitant and somewhat inconsistent, courts in the UK and US, as well as in many other nations, have invested increasing trust in science, specifically, in the forensic science of DNA profiling. In spite of protests from police and prosecutors such as Mr Castor, when faced with contradiction between the results of DNA profiling and other forms of evidence, judges and juries tend to place greater trust in the ‘scientific’ evidence.

Trust and certainty are bound up together [3]. The acceptance of DNA profiling as a certain, error-free method of personal identification has profoundly influenced the degree of trust invested in it, compared with other forms of criminological evidence. Eyewitness evidence has long been regarded as fallible, but courts in the UK and US entrusted jurors to make wise judgements about the credibility of eyewitness testimony in the circumstances of particular cases. And, although defendants frequently recant their confessions and claim that they are coerced into confessing, until the advent of DNA profiling, courts frequently took confessions as tantamount to guilt. Older forms of forensic evidence — handwriting analysis, lie detector tests, fiber analysis, ballistics, blood-spatter analysis, bite-mark analysis — which rely upon expert judgement and have limited connection to established science, are now called into question in comparison with the new ‘gold standard’ of DNA profiling. Even the old gold standard — friction-ridge identification, commonly known as fingerprinting — has been challenged. Scotland Yard
abandoned as unscientific the 16-point standard for declaring fingerprint matches, and in the US, the admissibility of fingerprint evidence has been challenged (thus far unsuccessfully) in a series of Federal and State cases starting in 1999 [4].

In the past several years, it has become commonplace in the courts, in the media, and in much of the technical literature, to contrast the scientific and objective evidence supplied by DNA profiling, with the experiential or subjective opinions given by traditional forensic experts.

The traditional role of expertise in forensic science is well recognised and it is something of a stereotype to visualise the distinguished, greying individual on the stand saying, ‘my opinion is based on my many years of experience in this field’. Whereas we do not for one moment deny the value of experience, we claim, as a matter of principle, that the scientist should, as far as possible, support his/her opinion by reference to logical reasoning and an established corpus of scientific knowledge. This is what we mean by ‘transparency’: the former ‘in my experience’ justification we refer to as ‘obscurity’ [5].

For decades, courts in the UK and US had permitted fingerprint examiners to declare categorically whether a suspect’s fingerprint matched or did not match a ‘latent print’ found at a crime scene. The 16-point standard used until 2001 in the UK was not a probabilistic measure; instead, it provided a conventional threshold for deciding if two prints displayed a sufficient number of detailed similarities to justify declaring a match. Other criminal justice systems used different numbers of points (Australia, for example, used 12) or no points at all. Two basic assumptions supported the courts’ trust in the person of the fingerprint examiner: first, it was assumed that no two individuals share the same fingerprint patterns, that each person’s fingerprints provide a unique ‘signature’ (indeed, historically fingerprints were sometimes used as signatures); and, second, it was assumed that differences between fingerprint patterns are sufficiently obvious that trained examiners should never mistake one individual ‘signature’ for another (Fig. 1). The courts trusted fingerprint experts not so much because of the examiners’ esteemed personal qualities, but because fingerprints were believed to be a certain means of individual identification and the job of detecting fingerprint matches was deemed trivial for trained persons, given sufficient evidence. The ‘greying individual on the stand’ was trusted because he declared what anyone with sufficient training could see with his or her own eyes.

Trials of technique

When a research group headed by Sir Alec Jeffreys of the University of Leicester, UK first introduced DNA profiling in the mid-1980s, they dubbed it ‘DNA fingerprinting’ and claimed that it was a sure means of individual identification, and it was quickly developed as a forensic tool [6]. Although novel in many respects, and based on entirely different principles, the new technique was named and described in a way that was parasitic upon the credibility of fingerprinting. Unlike ABO blood typing and serum analysis, the new technique was heralded as a unique, certain and even absolute means of identification. Jeffreys stated that ‘…the pattern is so varied (hypervariable) that any particular combination of the segments is as unique as a fingerprint’ [7]. A Home Office spokesman declared: ‘The procedure is very complicated but it provides the scientists with a DNA fingerprint which has been shown to be specific to a particular individual’ [8]. A journalist extrapolated, ‘Suppose we could test a million people every second. How long would it take to find one exactly the same? The answer is, the universe itself would die before we found one the same. It is simply an incomprehensible number’ [9]. The Economist declared it ‘...the perfect fingerprint: unfakeable, unique, and running in families’ [10].

Such analogies with fingerprinting are most apt in relation to the earliest technique that Jeffreys and his colleagues developed. This was the multi-locus probe (MLP) technique, which used chemical ‘scissors’ – restriction enzymes – to dissolve DNA into fragments. A DNA ‘fingerprint’ is not a direct trace of a person’s DNA. Instead, the MLP technique visualizes selected sequences of single-strand DNA and compares their sizes (Fig. 2).

The MLP technique used markers that bind to an indefinite number of chromosomal sites, resulting in a complex pattern of bands. Jeffreys and other proponents of the MLP technique believed that when two samples were compared, it was virtually impossible (except with identical twins) that an entire pattern of bands would match, although precise estimates could not be given for the likelihood that any given band, or number of bands, would match [11]. Matching bands were analogous to matching ‘points’ in a fingerprint comparison, in the sense that they were visually identified as elements in a holistic comparison.

MLP ‘fingerprints’ were quickly replaced in the late 1980s by the single-locus probe (SLP) technique (Fig. 3). This involved the isolation and marking of a limited
number (4, 6, 8 and sometimes more) of noncoding DNA regions known as variable number tandem repeat (VNTR) sequences. Selected VNTR sequences were shown to be hypervariable in the human population, and these were marked by means of radioactive probes.

Population studies (using, for example, blood taken from blood banks or from volunteers on police forces) generated probability measures for the frequency of SLP patterns in human populations and selected ‘racial’ subpopulations [e.g. Caucasian, (South) Asian and Afro–Caribbean in the UK]. Although the SLP technique enjoyed the advantages of greater control and more precise quantification, it also became subject to heated disputes in the courts and scientific literature.

SLP results were presented in probabilistic form, but the resulting estimates often seemed to connote near-absolute identity. Estimates of the chance that two, randomly chosen and unrelated, individuals would share the same combination of alleles in a DNA profile sometimes approached less than one in hundreds of millions. Consequently, the analogy with fingerprinting was left unshaken in hundreds of trials in the US and UK during the late 1980s. However, in the 1989 double-murder case NY versus Castro [12], the defense, with the assistance of some prominent scientists as expert witnesses, successfully challenged the admissibility of the DNA evidence. Although the defendant, José Castro was later convicted in the trial, the pre-trial admissibility hearing which ran for several days became a key turning point for DNA profiling. Critics questioned the scientific status and adequacy of forensic methods, and they attacked the population genetic assumptions and statistical methods used for estimating the likelihood of matches.

The ‘DNA wars’ lasted for a few years in the early 1990s, and were an instance of a dispute that crossed back and forth over the boundaries between science and law. In an effort to resolve the dispute, the US National Research Council conducted two high-profile investigations of forensic DNA profiling [13]. Although the instruments and techniques used in forensic DNA profiling were commonplace in biomedical research and diagnostics,
the investigations brought into relief questions about the specificity of forensic practice. Forensic evidence (particularly crime scene evidence) is collected in uncontrolled environments, and many of the police agents who collect such evidence have little or no technical training in methods for preserving the integrity of biological samples. Forensic scientists and technicians often lack the credentials of personnel in university and medical laboratories, and they work under severe caseload pressures. Moreover, in the US, and to a lesser extent the UK, criminal casework is often contracted to private firms, which use distinct techniques and have proprietary interests in the methods they use. The variability of laboratory practices, the uncontrolled conditions and involvement of nonscientific personnel, created doubts about possible sources of error and a general lack of transparency. Criticisms were also made about the assumptions and calculative procedures used to develop estimates of the probability of false-positive (or ‘adventitious’) matches between a given suspect’s profile and a crime stain profile.

Legal, technical and administrative fixes

Disputes about DNA profiling were aired in the pages of Nature, Science and other more specialized journals, but trials, pre-trial admissibility hearings and appeal cases provided a significant forum for presenting and resolving the arguments. Legal standards of admissibility, and legal procedures for presenting and resolving arguments, were integral to the conduct of the dispute. This was not a ‘scientific’ controversy in any straightforward sense, as lawyers and judges played a crucial role in opening up and closing off questions about the reliability of forensic DNA profiling. In addition, legal considerations of burden of proof and reasonable doubt, and inferences about what juries could or could not understand, framed the way the courts handled questions about the reliability and certainty of ‘scientific’ evidence [14].

Several technical changes in the procedures of DNA profiling bypassed former sources of argument. By the mid-1990s, criminal justice systems in the UK, the EU and the US converged on a new system, the multiplex short tandem repeat (STR) system, which uses hypervariable DNA sequences of relatively short length. This technique uses the polymerase chain reaction (PCR) to ‘amplify’ the amount of analyzable genetic material in a sample, and thus can use much smaller amounts of initial sample than the MLP and SLP techniques. STR markers chosen for a profile system – such as the FBI’s system which uses 13 different markers – are taken from distinct chromosomal sites, and they are visualized by means of laser scans which produce color-coded graphic outputs (Fig. 4). This system disambiguates, automates and ‘black boxes’ the reading of profile patterns, thus bypassing previous sources of dispute. It also takes DNA profiling further away from the analogy with fingerprinting.

Court decisions in favor of admitting DNA profiling and international convergence upon the STR system for developing DNA databases, provided a legal and organizational context in which the system became entrenched even while questions remained unresolved about sources of error and statistical uncertainty. Weighty advisory panels such as the NRC recommended administrative procedures for making ‘conservative’ probability estimates and conducting proficiency tests. Judicial citations of such recommendations had decisive impact in the legal system, even though questions remained about how closely such recommendations are followed [15].

An inversion of credibility

Fingerprint examiners were trusted because their judgments were expressed as absolute and unproblematic determinations. An examiner’s ‘subjective’ judgement had impersonal authority because, in principle, anybody with adequate training could reach the same conclusion. A forensic scientist testifying in an early death-penalty rape–murder trial made a similar claim about a DNA profile match, saying that detecting it was a ‘very simple straightforward operation…there are no objective standards about making a visual match. Either it matches or it doesn’t. It’s like you walk into a parking lot and see two blue Fords parked next to each other’ [16]. Others argued that it was far from a simple operation, and courts and advisory panels found the lack of ‘objective standards’ to be a severe problem. Currently, the STR system of DNA profiling is widely heralded as ‘objective’. Its objectivity is a matter of credibility – effective belief in validity – based upon a combination of ‘mechanical objectivity’ [17] and administrative science. Trust is invested in a combination of automated systems, impressive numbers [18], graphic outputs, and bureaucratic procedures.

Not only has STR profiling become more distant from its predecessor than the early name DNA ‘fingerprinting’ suggested, it has become a new ‘gold standard’ – the model of a science – and a basis for making invidious comparisons with other forensic sciences, including fingerprinting. Fingerprinting now seems quaint and outmoded: a legacy of the late-19th century; with no connection with the genetic revolution, which developed without stringent demands for quantitative estimation and dense administrative controls, it is undergoing repeated challenge in the courts, and will no doubt be refashioned to upgrade its scientificity.
What should not be forgotten, however, is that the DNA revolution in criminal justice was initially mapped on to an older forensic technology, and it was framed and legitimated through the ancient institution of the courts. Conservative reactions, expressed by the likes of District Attorney Castor who prefers to trust traditional evidence over DNA test results, might become a source of ridicule, but they express something that must never be forgotten: that forensic DNA test results are embedded in the details of criminal cases that will always, and should always, involve public consideration of the singular elements of a case. The meaning and significance of ‘scientific’ testing cannot be divorced from such consideration.

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References
1 Eddie Joe Lloyd, quoted in the New York Times (26 August 2002). Lloyd had been sent to prison 17 years ago and was released in 2002 when DNA testing showed a mismatch between his DNA profile and the profile developed from evidence at the crime scene
2 This story and the quotations from Mr Castor and Dr Blake are taken from Rimer, S. (2002) Convict’s DNA sways labs, not a determined prosecutor. New York Times (6 February)
7 Jeffreys, quoted in The Times (London), 3 December 1985
8 The Times (London), 27 November 1985
10 Anon (1986) 'Cherchez la gene'. The Economist (January), 68–69
11 In some cases, forensic scientists counted the number of matching bands within a sector of an autoradiograph, and estimated the probability of a matching profile by assigning a constant figure (for example 0.25), which represented the average likelihood of matching MLP alleles in the human population, to each matching band, and then multiplying that figure by itself for each match. For ten matching bands, the result would be 0.25 to the tenth power
12 People versus Castro, 545 N.Y.S.2d 985 (Sup. Ct. 1989)

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