The Forensic Use of DNA Profiling

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The application of DNA profiling in the criminal justice system is an important issue facing Australian courts and criminal investigators today. The technology is changing rapidly and several new techniques are becoming available. Increasingly, legal advisers are required to come to grips with this kind of scientific evidence.

DNA profiling has been described as a powerful breakthrough in forensic science by many. However, the scientific validity of its application by individual laboratories has been called into question by some. The implementation of DNA profiling also raises the issue of privacy. Should it be compulsory for all suspects to allow access to body samples? How long should the information be stored? This Trends and Issues canvasses these matters and outlines our current knowledge of DNA technology. The authors raise a number of issues that limit the use of profiling in forensic investigations, and stress the importance of the establishment of national standard techniques and the establishment of population frequency databases that reflect Australia's particular ethnic composition.

The Forensic Use of DNA Profiling is a major contribution to the debate on law reform. In the interests of justice, laws in all Australian jurisdictions need to be enacted which protect an individual's privacy, but allow our investigators access to a technology which can help in excluding the innocent.

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For a long time forensic scientists have been interested in using genetic information to match crime scene evidence with suspects in criminal investigations. Until recently the lack of a suitable experimental approach prevented them from achieving this end. However developments in molecular genetics during the last decade have changed the situation. In 1980, American geneticists discovered a region of DNA (the molecule that contains the genetic identity code in all life forms) that does not hold any genetic information and which is extremely variable between people. Since that time many other similar areas have been found. They consist of short sequences repeated many times with the number of repeats varying enormously among individuals. Thus a vast amount of DNA diversity has been found. Techniques for analysing this variation have also been developed and continue to improve. These developments in molecular biology when combined with the application of population genetic principles allow forensic scientists to achieve exclusion and a degree of individual identification not previously possible in the context of criminal inquiries.
Initially referred to as DNA fingerprinting, the promotional literature of the private companies involved claimed that their techniques had ‘the power to identify one individual in the world’s population’ and ‘that the chance that any two people would have the same DNA print was one in 30 billion’ (Neufeld & Colman 1990). It is important early in the present document to stress the inaccuracy of such commercial hype which implied an ability to provide a genetic code unique to an individual which is as discriminating as a fingerprint. The product of the existing technology is a profile, not a ‘fingerprint’. It is unlikely to be unique. An estimate of probability of a match between two genetic samples occurring by pure chance needs to be made. The discriminating power of the method depends on the number of variants that exist and the frequency with which each occurs in the population. Therefore, the outcome is normally a statistical probability that is substantially less than ‘one in 30 billion’ when all factors are taken into consideration. This does not mean that profiling is not a valuable tool to be used by forensic work. It has the potential to be extremely powerful. Certainly it is not an infallible identifier of an individual but it can be a reliable technique if used correctly; it is by no means foolproof.

### A Complex and Rapidly Changing Technology

Most Australian and overseas forensic laboratories are currently using some type of RFLP (Restriction Fragment Length Polymorphism) analysis. In the regions of DNA between genes (non-coding regions) some sequences are arranged adjacent to each other in blocks on a chromosome; in other words, the sequences are tandemly repeated and are extremely variable in the number of repeats, rather than in the base composition of the sequences. For instance, one person might have 13 repeats of a sequence at a certain locus; another might have 33. Labelled VNTRs (Variable Number Tandem Repeats), these areas of DNA do not contain genetic material; thus, any genetic ‘secrets’ are not revealed in the profile. The RFLP method cuts out a specific VNTR which would vary in length depending on the number of repeats.

In Figure 1 the bands representing the DNA profiles of three suspects, a crime scene sample and the victim are portrayed. Note that in this example, the profile of suspect 2 matches that of the crime scene.

The procedures are complex, involving at least eight stages, that can employ a variety of different processes (protocols) and materials. The end result is an image on an X-ray-like film (autoradiograph) that reveals the length of the RFLP in the form of a band. The length is measured by the distance travelled in an electric field; smaller fragments move farther and would appear in a position on the autoradiograph that corresponds to its length. The final step, the comparison of bands from two samples, requires an understanding of population genetics and molecular biology.

DNA profiling is in a period of rapid discovery and change. The PCR (Polymerase Chain Reaction) method, although still in the experimental stage, may well revolutionise the entire technology. PCR can replicate the DNA in one cell multi-million fold even if it is substantially degraded. This means that smaller more degraded samples can be used and that non-intimate body samples such as saliva and hair which can be acquired...
less invasively from suspects could be more easily analysed (Easteal 1990)

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**The Forensic Use of Profiling**

Initially, forensic profiling was only available from three private American and British companies: Lifecodes (USA), Cellmark Diagnostic (UK; USA) and Forensic Science Association (USA). Such privatisation and patenting of techniques has been widely criticised in the literature for a number of reasons including the consequent restrictions on information sharing and the vested interests of such company scientists in their court presentation and interpretation of profiling data. In the United States, the public sector began to conduct forensic DNA tests in late 1988 starting with the FBI laboratory and followed by various states such as Virginia and New York, and profiling rapidly gained popularity.

Police in Britain first used the technology in 1985, and Britain, along with Switzerland and Germany, shares the lead in such work in Europe. It was used for the first time in British courts in October 1987, when a man accused of rape, changed his plea to guilty when faced with the incriminating DNA evidence. Also, in 1987, the celebrated Pitchfork (see Read 1989) case eliminated an innocent 17-year-old charged with two rape-murders and identified the perpetrator using DNA profiling. Over 5,000 samples from local men were obtained. It should be noted that the defendant, Pitchfork almost succeeded in misleading the police. After first refusing to allow a blood sample to be taken, Pitchfork later persuaded a friend to substitute his own sample. This shows the enormous importance of security procedures with mass testing.

The forensic science laboratory of Britain’s metropolitan police has frequently been called upon to produce profiles; over 700 in London in 1989. Most were undertaken to determine whether the right person was in custody; in 28 per cent of the cases, suspects were released after DNA analysis excluded them as matches with crime scene evidence (Thompson & Ford 1990).

In Australia, the limited amount of profiling has been done both by private companies and, increasingly, by state forensic laboratories. Two states, New South Wales and Victoria, have implemented the Lifecodes technique. That company offers a training and accreditation package with a supply of Lifecodes’ patented radioactively labelled probes, technical advice and a population database that includes United States Caucasian population frequencies (Read 1989). The South Australian forensic laboratory has not opted for the Lifecodes system but is working on developing PCR based methods. Tasmania is currently adopting the FBI methodology.

The actual presentation of DNA profiling evidence in Australian courts has been limited in comparison to overseas. Its first use in criminal proceedings was in the ACT in 1989 when a man charged with three counts of sexual assault changed his defence during the trial from ‘I wasn’t there’ to a defence that the woman consented (Coelli 1989). This change was motivated by profiling evidence that matched his blood with crime scene evidence. In this case, the accused only gave a blood sample after police allegedly persuaded him that they were entitled to take it under the same law that allows for hand fingerprinting. This has raised some question of civil liberties in the press (Reform 1989; Scutt 1990; Coelli 1989).

A Tasmanian Supreme Court criminal trial involved genetic profiling with blood samples analysed from over 20 people. In addition, DNA evidence has been submitted by Genetic Technologies Corporation (Australasian licensee for Lifecodes Corporation) in three criminal cases - one in Queensland, two in New South Wales. It was challenged in two and found admissible. Victoria State Forensic Science Laboratory has provided DNA profiling evidence in 15 trials/hearings to date.

It should be noted that the potential power of DNA profiling in criminal investigations would not necessarily be reflected in the numbers of court cases. Much of its impact is behind the scenes; excluding suspects and reducing court load through plea changes and bargaining. Thus, only a small proportion of cases involving DNA evidence would in all likelihood, culminate in judicial proceedings.

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**Current Limitations on the Use of Profiling**

It should be pointed out that there are a number of factors that can restrict the utilisation of profiling in forensic investigations. These include:

- An adequate amount of undegraded and uncontaminated DNA must be extractable from the crime scene. DNA does degrade with prolonged exposure to sunlight, heat and humidity. Additionally, it may become contaminated with yeast, bacteria, or fungus although such contaminants can be detected if proper controls are employed in the laboratory.

Aside from the above factors, obviously the ability to use the technique is dependent upon the perpetrator leaving a sample at the crime scene, such as blood, semen, hair, soft tissue. For example, remnants of sperm will not always be left at rapes since the rapists are often impotent and do not ejaculate (Scutt 1990). Furthermore, if the rapist has had a vasectomy, his semen would contain little or no DNA.

- Some aspects of sex crimes and how such acts are often perceived in society may also mitigate against the use of DNA profiling. Most rape victims for instance do not report the crime or it is reported too late for effective vaginal smears to be obtained. Victims also often refuse to have a
Forensic medical examination or to press charges (Freckelton 1990a). Additionally, in many rape cases, the issue of whether sex took place is not contested; the question of victim consent is frequently the main question.

- Forensic scientists can also be limited by law enforcement officers’ legal inability to obtain suspects’ non-intimate and/or intimate body samples for matching or exclusionary purposes. In the United States, constitutional treatment of the collection of hair or saliva samples under the fourth amendment has been varied; a search warrant is usually required, however probable cause is sufficient if exigent circumstances are found to exist. One state court has held that DNA evidence acquired from hairs plucked by police officers was admissible since the defendant was being arrested and the hairs were in plain view (Renskers 1990). However, the federal courts have not yet resolved the issue of whether involuntary removal of hair samples constitutes legal search and seizure. Although the United States Supreme Court has recognised the constitutionality of compelled blood-taking for blood-alcohol tests, probable cause and a warrant must exist prior to obtaining blood for DNA samples. Additionally, a defendant’s refusal to submit to such a test will probably be admissible into evidence at the trial. This is also true in England. If a suspect refuses to provide a blood sample, (s)he is warned that the refusal may be treated in any proceedings against him as corroborating relevant prosecution evidence’ (Read 1989, p. 1156). This power of persuasion may reflect existing legislation. Police powers of persuasion may reflect existing legislation. McLeod (1990) believes that thus far the courts have shown a readiness to allow quite generous interpretations of the existing legislation. Police powers of persuasion are certainly an additional factor.

- Cost and time required for profiling may prohibit its extensive use since it is fairly expensive, labour intensive and time consuming. However, increased use and technological changes may well reduce these variables. Recent RFLP technique modifications by the FBI laboratory for example, have resulted in a significant reduction in both time and cost (Budowle & Baechtel 1990). Increased use of PCR methods would also significantly cut down on the hours, work and money involved.

- The lack of standardisation limits investigators’ ability to match ‘profiles’ generated from different laboratories. It also may prohibit adequate additional reliability tests for the courts.

- The fallibility of the technique may also limit its effectiveness as evidence. The potential for error and the steps which can be taken to enhance the reliability of DNA profiling forensic analysis will be discussed further below.

- The lack of a national database also limits the use of profiling. Much of the potential uses lie in addressing old unsolved cases; cases where the assailants were unknown; and serial homicides and rapes which all require a data bank of DNA profiles. Byers (1989) notes that a database and the routine use of the techniques could render standard alibi defences insignificant; greatly reduce the importance of eyewitness
testimony; speed up the court calendar and clear up unsolved crimes.

Is DNA Profiling Scientifically Reliable?

Initially, profiling evidence went virtually unchallenged in the judicial realm overseas and was admitted in at least nine American states and by several appellate courts. Lifecodes has provided testimony on its DNA analysis findings in over 100 cases in the past three years. Then, in 1989, a pretrial hearing in a New York City murder trial (The People v. Castro, unreported decision, 14 August 1989, New York Supreme Court) suddenly propelled DNA profiling into the news with some doubt cast upon its scientific acceptability. The defence was challenging the admissibility of the DNA evidence with a 'Frye hearing' which tests the reliability of scientific procedures by determining their acceptance by experts in the relevant fields.

The defendant in this case was charged with killing a woman and her baby. Blood was found on his watch which was analysed by Lifecodes for a match with the victim's. Four scientists involved in the hearing met outside the court to discuss the scientific merit of the case. The result as Hoeffel (1990) states was that the 'infallibility mystique' of profiling was successfully questioned. The scientists concluded that 'serious doubts [exist] as to the reliability of the DNA evidence based on the test procedures used and the interpretation of the results (Renskers 1990). They specifically stated that:

The DNA data in this case are not scientifically reliable enough to support the assertion that the samples... do or do not match. If these data were submitted to a peer reviewed journal in support of a conclusion, they would not be accepted (Lander 1989a, p. 504).

As a result of the experts' findings, the judge ruled that the exclusionary test would be accepted (the blood was not a match with the defendant's), but that the 'inclusionary' evidence - the match with the victim - was not admissible since the third prong of the Frye test had not been met (actual testing procedures). Thus, the scientific validity or worth of the technique itself was not faulted. The evidence relating to whether the blood was the victim's was excluded because the Lifecodes' laboratory did not keep sufficiently accurate records and did not conduct the tests in a generally scientific manner (Petrovich 1990). Specifically, discrepancies between the forensic report to the court and the laboratory findings were found. Three bands existed in the victim's lane; five in the 'watch' lane. The report did not mention the extra two bands. If the two bands reflect human DNA, then there was no match. Lifecodes claimed that they were not human but the result of bacterial contamination; yet no tests with bacterial probes were done (Longobardi 1989).

Lander (1989a), a key expert witness for the defence, pointed out a large number of problems with the procedures used in the Castro profiling. These include the lack of gender control; analysis of degraded DNA; probe contamination; population genetics problems in the calculation of probabilities (Lifecodes reported the odds of a match was due to chance at 1:100 million; Lander believes 1:24 is the correct probability); and the lack of objective criteria with lane-to-lane comparison for declaring matches.

Subsequently, scientists and lawyers have protested the premature forensic use of techniques lacking necessary validation and reliability checks. The director of Britain's Metropolitan Police forensic science laboratory explains how he perceives the American situation:

What happened in the U.S. was that a new company without any experience in forensic science, and a little naive about what can happen in court, was caught out. (Thompson & Ford 1990, p. 24)

However, it is important to stress that the Castro case did not question the fundamental scientific value and acceptance of DNA profiling. In fact, the Congressional Office of Technological Assessment which was mandated to examine the efficacy of the technique recently concluded that DNA profiling is 'reliable and valid when properly performed and analysed by skilled personnel' (Anderson 1990, p. 499). However, the Castro case did expose the potential for error in a laboratory's application. The result has been a push for various measures outlined below to ensure the dependability and soundness of profiling results.

Standardisation

Task forces such as the New York State DNA Forensic Panel have concluded that since dissimilar information cannot be compared, national standards need to be set in place for all testing procedures, analysis, interpretation and coding of data. Renskers (1990) emphasises that unless uniform standards are soon implemented, there is a great risk that the credibility of DNA forensic evidence could be 'compromised' through mishandling, mismanagement and improper analysis. Laboratories should follow written protocols that cover every step; this yields the highest reliability of results. If standardised, each laboratory's finds become not only useful to all the other agencies doing criminal forensic work but also permit more efficient and effective on-going quality control, accreditation and training of personnel (Beeler & Wiebe 1988). Rose and Keith (1989) also believe that standardisation of reagents and techniques is 'critical' but note that given the difference in view about what is the best system, a compromise would be the employment of a completely standardised core system by all forensic laboratories along with any additional systems that the individual laboratory might choose to use.
Perhaps more than any other stage - the interpretation of the results, the 'profile' has caused the greatest outcry for uniform standards. This includes the statistical processes used in defining a 'match' and the actual process of interpreting the bands. It is recommended that a scientifically acceptable standard of error should be established (Petrovich 1990). Recent court cases have also highlighted the unacceptability of visual matching of bands. All of the scientists testifying in court cases have also highlighted the established (Petrovich 1990). Recent acceptable standard of error should be recommended that a scientifically process of interpreting the bands. It is the statistical processes used in for uniform standards. This includes 'profile' has caused the greatest outcry - the interpretation of the results, the proficiency testing programs. The New York State Forensic Accreditation on DNA Analysis (1989 pp. 14-15) thus concluded that 'methodological problems unique to the forensic application of DNA technology . . . must be resolved and the most stringent controls be implemented'.

**Quality Control**

Hoefel (1990) states that much of the current problematic areas of DNA forensic analysis stems from a lack of proficiency testing and other on-going controls. Beeler and Wiebe (1988) agree and believe that all such laboratories need to be evaluated periodically with such controls as 'blind tests' that should be conducted by independent experts to ensure the accuracy of the laboratory's results. Further, strict quality assurance programs covering reagent production and test performance need to exist in order to determine when errors have been made (Rose & Keith 1989). Controls to determine procedural errors need to be part of such programs. The New York State Forensic Panel on DNA Analysis (1989 pp. 14-15) thus concluded that 'methodological problems unique to the forensic application of DNA technology . . . must be resolved and the most stringent controls be implemented'.

**Improved Population Data and Analysis**

It appears that several problems may be occurring in the reliable ascertaining of population probabilities in DNA profiling. To reiterate from above, a scientist must calculate the probability of a person having a particular DNA profile by chance through referring to accumulated population frequencies of the VNTRs that make up that profile. Some believe that a larger population database of these frequencies is required than already exists in order to improve the accuracy of test results (Byers 1989; Beeler & Wiebe 1988). The American Society of Human Genetics (1990) stress that more population reference bases are needed and believe that a central repository for such data should be compiled. Another major issue that has appeared in recent literature is the assumption apparently made by some forensic laboratories concerning the equilibrium of populations. For a particular VNTR to have the same frequency among all segments of a group, its members must be mating at random. If the subgroups tend to be endogamous (mate within their own group) genetic drift can act to cause changes in DNA variant frequencies of isolated populations. In fact, geneticists report that there are statistically significant ethnic differences for 18/20 RFLPs examined (Lander 1989b).

If there are differences in variant frequencies between population groups, it is vitally important to generate and employ data on these differences in forensic analysis when findings can markedly affect determinations of guilt or innocence or even life or death. Without subgroup sampling, an individual could appear guilty with a low probability given to pure chance of a match when in fact the probability was much higher. Let us say that the suspect is a Sri Lankan Australian and although variant A of a particular RFLP is very rare among mainstream Australians, it is markedly more common among Sri Lankans. Without the knowledge of and calculation of different sub-population frequencies, the suspect would appear to be a likely match if variant A is also found in sperm at the crime scene; if Sri Lankan variant distributions are known, the probability might change dramatically. However, note that the use of Sri Lankan frequencies cannot take place without consideration of other factors. If it is known that the perpetrator was Sri Lankan or if the crime took place in a Sri Lankan neighbourhood, sub-population frequencies would be appropriate. However, if the general population database accurately reflects the proportion of Sri Lankans in the Australian population, then this database should be used if there are no particular reasons, for example, location, for believing that the perpetrator is Sri Lankan.

As Kearney (1990) points out, Australia is composed of many recent migrants from different gene pools, Islanders, Aborigines or other...

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**Accreditation**

The American Society of Human Genetics (1989 p. 632) recommends that 'appropriate scientific bodies need to develop comprehensive standards of practice' which could then lead to accreditation of laboratories engaged in DNA technology for forensic purposes or paternity testing. In other words, the standards need to be in place before laboratories can be certified as appropriately incorporating them in procedures.

The New York State Forensic DNA Analysis Panel (1990) also included a strong recommendation for an accreditation process requiring that each laboratory: 1) must fully document their methods and maintain careful quality assurance records; 2) must be totally equipped for molecular biology with confidentiality of records and secure long-term cold storage capability; and 3) must implement proficiency testing programs.
relatively isolated population groups. He also stresses the need for population geneticists' expertise in calculating probabilities within Australia.

This latter point should indeed be stressed. This area of DNA profiling involves the knowledge and expertise of those in the specific field of population genetics. For example, Eric Lander's (1989a) understanding of genetic drift and equilibrium resulted in his disputing the 1:96,000,000 odds of a declared match in a rape case (Texas v. Hicks - see Lander 1989a). His argument was that the laboratory in this case failed to take into account the fact that the crime took place in a small, in-bred Texas town founded by a handful of families.

**National Databases**

Figure 2 shows the reciprocal relationship between national databases of profiles and population frequencies, quality assurance programs, and standardisation of all aspects of the technique. The latter would enable the development of regulations and norms that are a prerequisite for both programs of quality control and accreditation and databases. Concurrently, quality assurance processes enhance the validity and reliability of the databases whilst they in turn increase the acceptability and value of the DNA forensic evidence.

The integral relationship between the efficacy of DNA forensic evidence and a national database is being recognised overseas where attempts are underway to establish such centres. However, as in the United States, Australia is impeded by the lack of standardisation. Thus, one profile is not transferable from one method of analysis to another. Therefore, interstate searches, for example checking a suspect's profile against ALL known DNA profiles, cannot be done unless or until the technology is standardised and/or a national data centre is established.

The FBI is attempting to develop the prerequisite standards and controls to implement a national database which would include population data and an investigative section including profile data for unsolved homicides and rapes; profiles from convicted sex offenders; and DNA 'prints' of missing persons and unidentified deceased (Nimmich 1990). They believe that aside from improving the scientific validity of the procedures, law enforcement agencies could benefit in a number of ways:

- Convicted sex offenders with a high rate of recidivism would be profiled (in a standard way by all agencies). Then, if body fluid is recovered in a sex crime, that profile could be entered for any match with prior offenders.
- The same process could be conducted for non-sex offenders.
- Detection of serial rapists would be greatly enhanced (Bigbee 1989).

**Privacy Issues**

Two major privacy issues arise in the implementation of any sort of DNA profiling and databank. The first, obtaining the intimate samples from suspects has been addressed earlier; the second is the issue of potential abuse of the stored information. Part of the concern may in fact be based on the erroneous impression that DNA profiles contain all of the individual's genotypic data including disease states and other genetic information that could potentially be used against someone if the knowledge were abused. In reality, within our present scientific awareness, these areas are not diagnostic for either disease states or other genetic conditions.

However, the above is only true if the DNA of the individual is not retained, and only their profile is kept in a computerised form. This question - what exactly to store - has met with conflicting responses from the scientific and forensic communities. The American Society of Human Genetics (1990 p. 632) believes that actual samples should be retained 'so long as the permissible uses of such material are defined initially, and so long as adequate rules of access and disclosure are implemented'.

The New York State Forensic DNA Panel (1989) disagrees with retaining the samples. Although they strongly recommend the use of a database to improve the ability of identifying suspects, they believe that the technique should match the DNA extracted from an evidentiary sample with suspects' DNA coded information stored in a database computer. Such information would not be the 'print' but only the data obtained from coding that 'print' along with relevant demographic
information. The panel specifically recommends that the DNA sample is not saved and that if a conviction is reversed, the computer's soft copy as well as hard ones of that individual's profile should be destroyed.

There is no doubt that a DNA databank could potentially violate someone's civil liberties if: a) it contained sensitive and genetic revealing information, and b) confidentiality were abused. However, even if the first condition prevailed and actual samples were retained, steps can be taken both legislatively and in the design of the database to reduce the possibility of any abuse or leakage taking place.

Although the retention of only coded profiles would eradicate privacy concerns, limitations would then be placed on further analysis or on the ability to generate new profiles from existing samples based on a change in the technology.

**Conclusion**

Certain major changes in forensic DNA analysis need to take place in order to better assure the reliability of individual laboratories procedures and the admissibility of DNA evidence, preferably at a national level. These include: a) standardisation of protocols; b) standardisation of probes and markers; c) development of standards for accreditation, quality control and personnel; and d) establishment of population frequency databases that reflect Australia's particular ethnic composition. While some of the overseas developments in these areas can provide useful input, they are not entirely appropriate in Australia. They neither reflect our legislative and political constraints nor the Australian forensic science structure. Furthermore, overseas population frequency databases have limited relevance to this country's population makeup. Thus, the wholesale acceptance of overseas developments is not acceptable; instead, Australia should plan and implement its own DNA profiling program integrating some, but not all of the ideas initiated abroad.

An Australian-wide database of criminals' DNA profiles would further promote the reliability of results both by generating the standardisation and quality assurance necessary and by providing law enforcement officials with a vital tool for identifying some criminals.

However, a national database or database would involve the networking of DNA information by definition. It is therefore imperative (particularly if samples are retained) as Renskers (1990 p. 338) states that: Legislation providing for protection of samples and information yielded must accompany the establishment of DNA databanks ... The right to genetic privacy should be statutorily recognised and protected.

In addition, legislation in each state and territory needs to be enacted to clarify law enforcement officers' powers in the acquisition of body samples. Without both types of laws, the full potential utility of DNA profiling in forensic will not be realised.

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