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DNA Identification in the Criminal Justice System

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DNA profiling and the forensic use of DNA evidence have undergone considerable development since the Australian Institute of Criminology first examined this topic in 1990 in Trends and Issues *no. 26. Some of the laboratory techniques described in that report have since been refined so that more precise DNA profiling is now possible, and a greater range of criminal investigations can benefit from the use of such forensic techniques. Moreover, the proposal in that report for a national DNA database has now been advanced, with the establishment on 1 July 2000 of the CrimTrac agency. However, many of the issues raised in relation to scientific reliability, standardisation of profiling techniques, laboratory accreditation and quality control, improved population and data analysis, and privacy are still the subject of disputation in legal proceedings.*

This paper examines the science of DNA identification and its use during criminal investigations and in criminal proceedings, including criminal trials, appeals and post-conviction proceedings. It describes the main benefits and costs of the increasing role of DNA identification in the criminal justice system.

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The Science of DNA Identification

A Natural Identifier

Deoxyribonucleic acid (DNA) is a long molecule, found in the cellular nuclei of living organisms. Since 1954, scientists have recognised that the chemical structure of an individual's DNA encodes information about that individual's inherited characteristics. The present limits on genetic science mean that a direct analysis of a person's DNA will yield only limited information about individual characteristics, although some research suggests that investigators may in the future be able to discern specific physical traits such as hair, eye and skin colour from forensic samples (National Institute of Justice 2000, pp. 18–19; van Oorschot et al. 2001). Rather, the current utility of DNA analysis to the criminal justice system arises from the comparison of DNA from two sources, such as DNA from a crime scene and DNA from a suspect, to determine the relationship between those sources.

Traditionally, the identification of a person has required the observation of that person's entire body or of localised special characteristics such as fingerprints, blood group or hair type. By contrast, DNA analysis allows identification by reference to the information contained in any human nucleic cell, irrespective of which part of the body the cell comes from. The DNA in a human cell is unique, the product of sexual reproduction that combines half of the mother's DNA and half of the father's DNA. Every cell in an individual's body is the result of cellular division, which copies the DNA in the newly fertilised cell into every other nucleic cell. As a result, DNA in a cellular nucleus is identical throughout a human body but variable between any two humans, making it a natural alternative to artificial human identifiers, such as names or tax-file numbers. The notable exception is identical twins, who develop from a single fertilised cell and hence have identical nuclear DNA.

The technique of "DNA identification" compares the DNA of two bodily samples to ascertain whether or not they came from the same human being. Identity of DNA in the cells across both samples implies that the samples are derived from the same person (or identical twins); non-identity implies different human sources. Alternative comparative techniques can

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be used to determine whether or not there is a familial relationship between the two human sources. For example, a matrilineal relationship can be inferred from a comparison of DNA in mitochondria, which pass from mother to child unchanged by sexual reproduction.

DNA Profiling

Comparison of human DNA molecules does not require analysis of the entire DNA molecule, as about 99.9 per cent of DNA is common to all people. Rather, DNA comparison need only focus on a portion of the remaining 0.1 per cent of human DNA that is sufficiently variable to be unique to individuals. Such variable DNA—termed "non-coding" (or "junk") DNA—plays no direct role in the development of human characteristics (Trent 2000, p. 52).

Modern comparative techniques compare only a small set of features of non-coding DNA. Such sets of features are known as DNA profiles and can be represented as an ordered series of numbers. That DNA profiles are easily quantified represents a further advantage over other unique human features, such as appearance and fingerprints, as it allows for automated analysis. The features comprised in a DNA profile must be sufficiently variable throughout the population to have an acceptable statistical likelihood that the profile is unique in that population, but also sufficiently regular to be amenable to cheap and efficient mass analysis. While several varieties of DNA profiling have been used in the past (National Institute of Justice 2000; Butler & Becker 2001; Freckelton & Selby 2002, ch. 80), the future of DNA identification in Australia is likely to be dominated by the type of profiling in present use (Box 1). Any significant future changes in profiling would render contemporary investigative databases obsolete.

Laboratory technicians do not "read" a DNA profile from a bodily sample. Rather, they construct a profile by inference from the outcomes of a series of procedures performed on that sample. Contemporary profiling techniques (Box 1) are increasingly automated, but the elimination of artefacts of the profiling process requires careful judgments by properly trained scientists (Roberts 1998, p. 32; Kaye & Sensabaugh 2000, pp. 516–17.) Accordingly, a DNA profile generated from a sample by contemporary procedures must be understood not as a fact about a sample but rather as an interpretation of that sample. Future developments may allow initial profiling to be done by non-technicians outside of the lab (National Institute of Justice 2000, p. 30).

An attempted comparison (or "matching") of two DNA profiles in order to determine whether they are related will yield one of three possible results (Table 1).

DNA Identification in Criminal Investigations

Linking People and Crimes

Crime investigators utilise DNA profiles from two sources: human bodies and small samples of human bodily material. DNA profiles can be obtained from human bodies by analysing samples removed from those bodies. Forensic procedures that can be used to obtain such samples (whether voluntarily or involuntarily) include blood sampling by injection, pulling out hair at the root and taking swabs from inside the mouth, known as buccal swabs.

In many cases, DNA profiles can be obtained from bodily samples that have become separated from a human body. Contemporary profiling techniques can generally be used on such tiny samples as the root of a pulled hair, saliva on a cigarette butt, a square-centimetre blood stain, skin cells from clothing or three micrograms of semen from a vaginal swab; standard or alternative techniques will sometimes succeed on other, less optimal, samples such as shed hair or skin cells from a handled object (Kaye & Sensabaugh 2000). Investigators will be interested in such samples if they suspect that they became separated from a person's body (usually either victim or offender) at the time of the commission of a crime, thus providing a potential insight into details of that crime.

The most important use of DNA identification by crime investigators is to compare a profile believed to be from a crime perpetrator (for example, derived from semen in a rape victim's vagina, or blood, hair or skin cells at a crime scene or on a victim's body) with a known person's profile. Other uses of DNA identification include:

- comparing a profile from foreign samples on a suspect's body or possessions with a victim's profile (to test the suspect's prior contact with the victim);
- comparing a profile from an unidentified person or corpse with a known person's profile (to test identity); or
- comparing profiles in two crime scene samples (to infer the details of a crime or the common involvement of one person in separate crimes.)

DNA matching can be used at various stages of an investigation. If a known person is a suspect at the time of the matching, then a positive match between crime scene DNA and that person will help to confirm the existing suspicion, while a negative match will tend to negate that suspicion. However, DNA matching can also be used before suspicion has fallen on a single individual by comparing the unknown sample profile to samples taken from a group of persons, such as all adult males within a locality. A positive match

Box 1: DNA profiling in contemporary Australian forensic laboratories

Since January 1999, all Australian forensic laboratories regularly involved in criminal casework have used a commercial profiling kit, *Profiler Plus*, owned by US-based Perkin Elmer Corporation. *Profiler Plus* analyses nine points in human DNA where short sequences of proteins are repeated a variable number of times. The profile consists of the number of repetitions at each point. For a sample from a single individual there will be up to two numbers at each of the nine points, one from each parent. "Mixed" samples from more than one individual (for example, a post-coital vaginal swab) will produce a more complex profile, requiring considerable interpretation. This general approach to profiling based on "short tandem repeats" (STRs), which has largely replaced "restriction fragment length polymorphism", or RFLP, is favoured in contemporary forensic laboratories because it is amenable to the analysis of small, degraded and mixed samples typically present at crime scenes. However, other techniques can be used in individual investigations where appropriate. The *Profiler Plus* system can theoretically distinguish over 10 billion possible variations of human DNA.

The key steps of the analysis are the introduction of fluorescent primers that attach to the beginning and end of the nine repeating portions of DNA, the simulation of the natural process of replication at those portions (using the "polymerase chain reaction", or PCR, method), and the passage of an electric current that separates the results by length. The resulting pattern of fluorescent primers can then be visually examined to discern the number of repeating portions at each analysed point of DNA.

Table 1: *Possible outcomes of DNA matching*

Outcome	Description	Explanation/interpretation
Null result	Profile comparison is not possible	This will occur when one or both samples are of insufficient quantity or quality (for example, because of contamination by DNA from microbes) to yield an adequate DNA profile.
Negative result (exclusion)	The profiles are different—no DNA match	This is conclusive evidence that the two samples are derived from different individuals.
Positive result (inclusion)	The profiles are the same—DNA match	This is evidence that the two samples are derived from the same human being (or identical twins). Note that there are several important alternative hypotheses for a positive result (see Table 3).

with one person will cast strong suspicion on that person, while a negative match to all persons will cast suspicion away from the entire group. Such mass screenings may occur as part of a single investigation, where the group is drawn from a particular location or shares an occupation associated with the crime. The largest mass screening in Australia to date was the April 2000 investigation following the rape of an elderly woman in the New South Wales town of Wee Waa, during which most of the town's 600 male residents volunteered mouth swabs for DNA testing (Moldofsky 2000).

The most important method of mass comparison is through the use of databases of DNA profiles from known persons, each of which can be easily compared with every crime profile, potentially yielding "cold hits", that is, entirely unsuspected links between known persons and crimes. This method has resulted in a significant number of convictions in jurisdictions such as the United Kingdom, New Zealand and the United States (Loftus 1999; Napper 2000; Tracey & Morgan 2000, pp. 644–5). All Australian jurisdictions have acted to create such databases, with samples drawn from volunteers, some crime suspects and certain categories of offender. With common protocols, different databases can be linked to expand the group of known persons whose profiles are regularly screened against crime scene samples (Haesler 2001). This is the idea behind the establishment of the National Criminal Investigation DNA Database administered by CrimTrac (Ellison 2001).

Investigative Results of Matching

If all parts of a known person's profile are present in a crime sample, then that person remains a possible source of that sample and, depending on other evidence, a potential suspect. Clearly, such an inclusion justifies further investigation of that person's involvement in the crime. Correct inclusions will increase the accuracy of investigations and the chance of convicting the perpetrator. Indeed, a suspect may respond to an inclusion by confessing to the crime. If a known person's profile is inconsistent (even in part) with a profile from a crime sample then that person is excluded as a source of that sample. In the right circumstances, exclusions may divert resources from fruitless inquiries and point investigators to the real perpetrator. More importantly, a timely exclusion that clears a suspect may save that suspect from the ordeal of a criminal investigation and even an erroneous conviction based on unreliable traditional investigative methods. In fact, the first investigative use of DNA identification, the *Pitchfork* case in England in 1986, produced an exonerating exclusion as well as a conviction. In that case, a 17-year-old mental hospital worker had confessed to one of two murder/rapes, believed to have been committed by the same person. Police sought DNA analysis in the hope of proving that this suspect had committed both crimes, but the testing instead excluded him altogether. The real offender was subsequently identified after he bribed someone else to stand in for him in a mass screening of local men, and a positive DNA match then led to his conviction for both crimes (National Institute of Justice 1999, p. 1; Napper 2000, p. 65).

It is important to recognise that not all inclusions or exclusions will be of value in an investigation or trial. An inclusion will be of limited significance where identity is not disputed (for example, many rapes where the defendant concedes intercourse but argues that the victim consented) or where there are innocent explanations for the presence of a suspect's DNA at a crime scene or on a victim's body (for example, the suspect is an acquaintance of the victim or a resident of the crime scene). Likewise, many exclusions will also be equivocal, because the presence of another person's DNA at a crime scene will not necessarily disprove the involvement of the suspect in the crime. The utility of DNA identification is also entirely dependent on the correct characterisation of crime scene samples, which may be compromised by poor crime scene management or even deliberate misconduct.

A persistent danger is the possibility of false inclusions—that is, matching profiles from samples with different human sources. There are several alternative hypotheses for a positive DNA match that must be considered (Kaye & Sensabaugh 2000, pp. 520–34; Freckelton & Selby 2002, ch. 14); see Table 2. To date there have been no known false DNA matches in Australia, but there have been at least two noteworthy instances overseas. In 1999, UK police arrested and charged a man with burglary following a DNA database match. The profiling technique then in use indicated that the chance of an unrelated person having the same profile as the burglar was one in 37 million. However, a more discerning DNA identification technique later proved that the initial match was false (Concar 2001.)

Also in 1999, the New Zealand DNA database linked two Wellington murders to a Christchurch man who was on the database as a victim of an unrelated crime. An inquiry later concluded that the murder samples were contaminated by the victim's sample in the testing lab in Auckland (Eichelbaum & Scott 1999.) Changes to profiling techniques since these errors occurred have reduced some of these risks, but cannot eliminate them entirely. False exclusions, which can occur through errors in the handling of samples or the reporting of results, are of less concern as they can be largely avoided through improved protocols.

DNA Identification in Criminal Proceedings

Evidence in Criminal Trials

Presentation of DNA identification evidence in a criminal trial can be difficult for a number of reasons. First, DNA identification evidence comprises a number of facts, including the circumstances in which the relevant body samples were obtained, their

secure transportation to a laboratory, their analysis and the detection and recording of DNA profiles. The need to prove all of these facts can be avoided with the agreement of the defendant. However, defendants are not obliged to agree, potentially resulting in long and complex proceedings or difficulties in fact finding (Redmayne 1995).

Moreover, evidence law requires opinions about forensic interpretation to be presented by a person with specialised knowledge based on training, study or experience that substantially or wholly supports the opinion. The interpretation of DNA evidence requires expertise from several fields, notably genetics, statistics, laboratory technique and crime-scene analysis (Roberts 1998, p. 36). On a number of occasions, Australian courts have permitted a person qualified in a single field to present an opinion based on several fields.

Further, there is considerable controversy about how DNA evidence ought to be presented in a way that is both accurate and intelligible to people without a scientific background, especially jurors (Roberts 1998, p. 36; Heyes 2001, p. 13; Jowett 2001). Jurors deliberating on a verdict must consider the DNA evidence along with all other evidence in the trial in deciding whether or not the defendant's guilt has been proved beyond reasonable doubt. Australian trial lawyers and judges are not permitted to define the words "beyond reasonable doubt" in addressing the jury, but their comments on the use of DNA evidence may critically affect the way in which jurors approach their task. A major concern relates to evidence of the probability of a coincidental error, which may be misunderstood as a statement of the probability of the defendant's innocence (the "prosecutor's fallacy"), or as an estimate of the number of potentially guilty persons in the population (the "defendant's fallacy"). Disputes about the presentation of the risks of error may also result in jurors being confronted with contradictory interpretations of DNA evidence by opposing experts (Jowett 2001).

Australian Cases

DNA evidence has featured in numerous Australian cases dating from 1989 to the present (Freckelton & Selby 2002, ch. 14); see Table 3. The general principles established by these cases can be summarised as follows:

- DNA evidence is admissible in Australian criminal trials, subject to the evidentiary requirements that it be relevant to the facts in issue, is presented in an appropriate manner by qualified witnesses, and that it does not cause unfair prejudice to the accused.
- Profiling techniques commonly used by Australian laboratories provide an acceptable basis for the comparison of forensic samples in order to provide evidence of an accused person's contact with a victim or crime scene.
- Scientific opinion on the interpretation of DNA evidence may be admissible provided that it is given by persons with specialised knowledge based on training, study or experience.
- Statistical evidence of the probability of anyone other than the accused having the same DNA profile as a given profile (for example, from a crime scene) may be presented, but care must be taken to explain the basis on which the calculation is made, including relevant characteristics of the population database used, and the presentation must avoid misrepresenting this probability (for example, the prosecutor's or defendant's fallacies).

Significant issues that have not yet been fully addressed in Australian courts include the statistical interpretation of DNA database matches and the admissibility of improperly or illegally obtained DNA evidence.

DNA Identification in Post-conviction Reviews

Criminal Appeals

Where people may have been wrongly convicted, for example on the basis of mistaken eyewitness identification, exculpatory DNA evidence may form the basis of an appeal against conviction. In the United States, post-conviction DNA testing has been used in numerous appeals to overturn wrongful verdicts (Connors et al. 1996; Scheck, Neufeld & Dwyer 2000). Some of these cases have involved prisoners awaiting execution for capital crimes (Scheck, Neufeld & Dwyer 2000; Liebman, Fagan & West 2000).

In Australia there have also been several prominent miscarriages of justice, including convictions based substantially on questionable scientific evidence (Carrington et al. 1991). However, DNA identification has not so far featured in the post-conviction detection of such errors by Australian appeal courts. A notable exception is the recent case of *Button*, in which the Queensland Court of Appeal unanimously accepted that a DNA test conducted after a rape conviction indicated that someone other than the appellant had committed the offence (see Table 3).

The introduction of new or "fresh" evidence in Australian criminal

Table 2: *Main alternative hypotheses for a DNA match*

Scenario	Description	Likelihood	Ways to reduce error
Coincidence	Crime sample comes from an unrelated person with the same DNA profile.	A slight possibility for contemporary profiling methods. The possibility can be estimated by sampling profiles from the population of possible offenders. The statistical risk is increased if both suspect and offender come from a genetically isolated group or if the suspect was located through a database search.	• Increase the number of DNA features profiled. • Test or exclude possible suspects, for example, by mass screenings or database searches.
Kinship	Crime sample comes from a related person with the same DNA profile.	A higher possibility than the coincidence scenario if the suspect and perpetrator are first cousins or closer. If the suspect and perpetrator are identical twins then the likelihood is 100 per cent. Risk can be estimated using straightforward population genetics.	• Increase the number of DNA features profiled. • Test or exclude close relatives, especially identical twins.
Contamination	Crime sample is contaminated by DNA from suspect sample.	A possibility if the suspect sample has ever been near the crime sample. The risk of a false inclusion is higher where the profiling process replicates small amounts of DNA (as currently occurs in Australian labs; see Box 1).	• Separate profiling of suspect and crime samples. • Introduce stringent crime scene and laboratory protocols to avoid contamination. • Preserve part of the crime sample before testing (not possible for some crime samples).

appeals is not a straightforward matter. Courts of Criminal Appeal are statutorily empowered to receive evidence if this is deemed "necessary or expedient in the interests of justice". However, there is a general common law requirement that the evidence be "cogent" and "fresh". Australia's highest appellate court, the High Court, has no power to receive fresh evidence in a criminal appeal and so may be unable to hear an appeal based on new DNA evidence (Urbas forthcoming).

Review Commissions and Innocence Panels

Some jurisdictions have established formal independent review bodies to investigate suspected wrongful convictions and, if necessary, to refer them to courts of criminal appeal for reconsideration. An example is the Criminal Cases Review Commission in the United Kingdom, established under legislation in 1995 and in operation since 1997 (CCRC 1998). Less formal review bodies are the various "justice" or "innocence" panels set up by legal academics and defence lawyers in an attempt to assist convicted persons. The best known of these is the Innocence Project founded by attorneys Barry Scheck and Peter Neufeld in 1992 at the Benjamin N. Cardozo School of Law in New York, which has used DNA evidence to overturn convictions in over 100 cases (Scheck, Neufeld & Dwyer 2000; Innocence Project 2002).

The government of New South Wales has announced the creation of an Innocence Panel, with 10 members headed by a District Court judge, and including representatives of the New South Wales Police Service, the Director of Public Prosecutions, the New South Wales Privacy Commissioner and victims of crime (Australian Law Reform Commission & Australian Health Ethics Committee 2001, p. 413). Other Australian jurisdictions may act to establish similar bodies (Harvey 2001). University-based innocence projects have also been established at the University of Technology in Sydney (Liverani 2001) and Griffith University in Queensland (Alternative Law Journal 2001).

Conclusion: Benefits and Costs of DNA Identification

The most obvious benefit of the use of DNA identification in criminal investigations arises when the technique generates a link between a suspect and a crime that ultimately leads to the conviction and punishment of the criminal. This may also avoid costly alternatives such as the use of less efficient traditional investigative techniques, which can in some cases lead investigators to target the wrong person.

Proponents of DNA identification point out that a negative result in a DNA comparison may well prevent a miscarriage of justice, as innocent persons may be removed from suspicion at early stages of an investigation. Availability of DNA evidence may also affect offender behaviour in useful ways, by prompting admissions or incriminating attempts to evade DNA profiling or explain away a profile match. Particularly with the introduction of DNA databases, some offenders may even be deterred from further criminal activity by the increased risk of detection.

The risks of false or misleading results from DNA identification are not cause to reject its use by crime investigators, particularly where there is independent evidence about a suspect's guilt or innocence. Rather, there is obvious cause for considerable

Table 3: *Significant Australian DNA cases*

Jurisdiction	Case	Description
Australian Capital Territory	*Desmond Applebee* (see Director of Public Prosecutions 1989, p. 84)	First use of DNA evidence in Australian criminal proceedings. The accused was charged with sexual assault and initially denied any contact, but altered his defence to consensual intercourse after DNA evidence was admitted as part of the Crown case. He was convicted by a jury.
New South Wales	*R v Tran* (1990) 50 A Crim R 233	Conflicting expert evidence on DNA test results held inadmissible due to tendency to produce a misleading and confusing impression for the jury.
Victoria	*R v Lucas* [1992] 2 VR 109	DNA evidence tendered to establish source of a bloodstain on a wall by reference to DNA samples was held inadmissible due to its probative value being outweighed by its possible prejudicial effect.
Victoria	*R v Percerep* [1993] 2 VR 109	Rejection at trial of DNA evidence on grounds of scientific disagreement and imprecision resulting in low probative value, confirmed on appeal.
South Australia	*R v Jarrett* (1994) 62 SASR 443	Laboratory process of constructing DNA profiles using PCR techniques judicially considered and accepted. The question whether forensic experts had performed analysis competently was held to be a matter for the jury to decide.
New South Wales	*R v Milat* (1996) 87 A Crim R 446	On the issue of minimum size of statistical databases used for calculation of match probabilities, the court held that databases of several hundred were adequate.
New South Wales	*Pantoja v R* (1996) 88 A Crim R 554; *R v Pantoja* [1998] NSWSC 565	On appeal, DNA evidence was ruled inadmissible as the databases used by the prosecution were not shown to be statistically valid. (The accused was a member of the Quechua Indians population sub-group.) On re-trial, DNA evidence was admitted and the accused was convicted. A second appeal against conviction failed.
South Australia	*R v Karger* [2001] SASC 64	Murder trial involving a three-month preliminary hearing to review the scientific acceptability of the *Profiler Plus* system. The DNA evidence was admitted.
New South Wales	*R v Gallagher* [2001] NSWSC 462	Murder trial was suspended after a defence challenge to the *Profiler Plus* system. The DNA evidence was subsequently held admissible, but the accused was acquitted.
Queensland	*R v Button* [2001] QCA 133	First Australian appeal overturning a conviction on the basis that DNA evidence not presented at trial but considered on appeal indicated that someone other than the appellant had committed the crime.
New South Wales	*R v Sing* [2002] NSWCCA 20	Appellate court held that evidence of correct DNA testing procedures should have been presented at trial by forensic experts who conducted specific testing, rather than evidence of general procedures and instructions given by supervisors. Appeal allowed.
New South Wales	*R v Keir* [2002] NSWCCA 30	Murder conviction was quashed and a new trial ordered after the NSW Court of Criminal Appeal found that the trial judge's directions to the jury contained the "prosecutor's fallacy".

caution, especially when a DNA inclusion is the only incriminating evidence, as will often be the case when a DNA database generates a "cold hit". As many defendants will be poorly placed to maintain their innocence in the face of an apparent inclusion, and may even choose to plead guilty, the burden of fully investigating the possibility of a false or misleading inclusion will fall mainly on the state. Care must also be taken to ensure that criminal juries understand the risk of error.

Costs of increased reliance on DNA technology in criminal investigations include not only the obvious financial costs of scientific expertise, laboratory equipment and the administration of information databases (Tracey & Morgan 2000, pp. 663–7). A further, unquantifiable cost of the use of DNA evidence is a possible reduction in individual freedoms, notably the right to privacy. The use of DNA evidence involves invasions of bodily integrity and the scrutiny of individual genetic information, some of which may be coerced, both lawfully and otherwise.

While the infringements of privacy from DNA sampling procedures and the profiling of non-coding DNA are, arguably, minor, the increased use of DNA evidence by investigators may lead to "function creep", whereby more intrusive infringements become acceptable—for example, the scrutiny of DNA by public and private organisations to discern inheritable characteristics. The investigative power of DNA evidence may also create pressures to cooperate with criminal investigations that undermine the privilege against self-incrimination (Gans 2001). Reviews of relevant legislation in each Australian jurisdiction by law reform bodies provide an opportunity to monitor and reduce the costs of the use of DNA identification in the criminal justice system (Australian Law Reform Commission & Australian Health Ethics Committee 2001; New South Wales Legislative Council 2002.)

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