In this paper, the authors describe recent developments in DNA technology. Key cases involving DNA evidence in Australia and overseas that occurred between 2003 and 2014 are used to illustrate the benefits and potential issues that can arise when new DNA techniques are applied to criminal investigations. Empirical data on the value of DNA evidence and DNA databases on investigative and court outcomes are outlined, demonstrating strong support for the value of DNA evidence to investigations and prosecutions.

The techniques and applications for DNA evidence described here, and future developments, clearly have important implications for policymakers, practitioners and legislators. Equally, the Australian criminal justice system will need to continue to adapt to accommodate new developments in this field.

Chris Dawson APM
Director

Recent developments in DNA evidence

Marcus Smith and Monique Mann

DNA evidence has made a significant contribution to criminal investigations in Australia and around the world since it was widely adopted in the 1990s (Gans & Urbas 2002). The direct matching of DNA profiles, such as comparing one obtained from a crime scene with one obtained from a suspect or database, remains a widely used technique in criminal investigations. A range of new DNA profiling techniques continues to be developed and applied in criminal investigations around the world (Smith & Urbas 2012).

This paper is the third in a series by the Australian Institute of Criminology (AIC) on DNA evidence. The first, published in 1990 when the technology was in its relative infancy, outlined the scientific background for DNA evidence, considered early issues such as scientific reliability and privacy and described its application in early criminal cases (Easteal & Easteal 1990). The second, published in 2002, expanded on the scientific background and discussed a significant number of Australian cases in a 12-year period, illustrating issues that had arisen in investigations, at trial and in the use of DNA in the review of convictions and acquittals (Gans & Urbas 2002).

There have been some significant developments in the science and technology behind DNA evidence in the 13 years since 2002 that have important implications for law enforcement and the legal system. These are discussed through a review of relevant legal cases and the latest empirical evidence.

This paper is structured in three sections. The first examines the scientific techniques and how they have been applied in police investigations, drawing on a number of recent cases to illustrate them. The second considers empirical research evaluating DNA evidence and databases and the impact DNA has on investigative and court outcomes. The final section discusses significant cases that establish legal precedent relating to DNA evidence in criminal trials where significant issues have arisen or new techniques have been applied that have not yet been widely discussed in the literature. The paper concludes by reflecting on implications for policy and practice.
DNA profiling

DNA is unique between individuals and can be used as a means of identification. Repetitive sections of DNA, called short tandem repeats (STRs), vary between individuals. A DNA profile is created by analysing the number of STRs that occur at specific points in an individual’s DNA. DNA profiling uses ‘non-coding’ regions of the human genome that do not provide any information about the individual beyond identification. DNA profiles are a series of numbers that can be used for identification. DNA profiling uses non-coding regions of DNA rather than coding regions that directly influence human traits. Aside from gender, these profiles provide no biological information about the individual, such as their physical appearance. A complete match between a DNA profile from a crime scene sample and a DNA profile from a suspect sample provides probabilistic support for inferring that the samples are from the same person. This process is known as direct matching. A non-match provides evidence that the samples are not from the same person (Butler 2011). However, as discussed by Gans and Urbas (2002), there are alternative hypotheses that could account for a match, such as sample contamination in the laboratory or at the crime scene. Thus, DNA evidence must always be considered in the context of the other available evidence in a case. The consequences of laboratory error are illustrated by the 2009 Australian case R v Jarra (see Table 1), in which the contamination of DNA evidence resulted in an individual being wrongly convicted and imprisoned.

DNA profiling is now routinely used in criminal investigations in Australia. For example, in sexual assault cases where the offender deposits DNA on the victim’s clothing, a burglary where the offender leaves DNA at the scene or a homicide where the victim’s hair is found on the clothing of a suspect.

New techniques

Several new techniques have been developed in the past decade that extend beyond the process of direct matching as described above, and have increased the way in which DNA may be used as part of the criminal justice process. These developments, and the potential implications and considerations for criminal justice agencies, are described briefly below.

Familial searching

A DNA profile can demonstrate genetic relationships when the profiles are matched on the basis of the number of STR markers that are shared. For example, a parent would be expected to share half of their STR markers with their child, because half of a child’s genetic code is received from each parent. This is the basis of familial searching—a new application of DNA profiling that is increasingly being used in criminal investigations around the world (McCarthy 2011).

Familial searching is used in criminal investigations where DNA evidence is found at a crime scene and a match cannot be established with any of the DNA profiles held on the databases available to police. While complete DNA profile matches may not be found on the database, these searches may identify partial matches. These partial matches may indicate the DNA profiles of genetic relatives in the database. Where a partial match is identified, this can provide new investigative leads and a greater chance of identifying suspects (Greely et al. 2006). The English case R v Harman provides an example of how familial searching is able to provide police with additional options where they are not otherwise able to progress with an investigation (Table 2).

Kinship matching

Kinship matching is based on the same principles as familial matching. It commonly refers to the use of this technique in civil investigations, such as an airline crash involving multiple fatalities where unidentified bodies are found, or in isolated cases of unidentified bodies. In kinship matching, family members voluntarily submit samples of their DNA to investigators. Where a partial match is found, this can reduce the time required to identify bodies in natural disaster and missing person cases. Kinship matching was used in the forensic identification process associated with the 2014 Malaysia Airlines Flight 17 disaster in the Ukraine to identify victims from all over the world, including Australia (Netherlands Forensic Institute 2014).

mtDNA profiling

Mitochondria are structures within the cell responsible for the production of energy. Each mitochondrion has its own DNA, and there are usually several hundred mitochondria per cell. Mitochondrial DNA (mtDNA) is maternally inherited, and is identical between siblings and maternal relatives. Mitochondrial DNA profiling can be used to provide greater specificity in familial searches where a number of partial matches are obtained. As there are multiple copies of mtDNA per cell, it can be used in the identification of degraded samples such as skeletal remains, where there may be insufficient nuclear DNA to create a profile (Coble et al. 2004). Because the mtDNA profile can be traced back maternally for generations, it can also be used to trace an individual’s geographic ancestry back to specific continents or countries (Kopec 2014). Their likely ethnic background can be used to obtain a likely physical description of a suspect. Mitochondrial DNA has been used on a small number of occasions in Australia—most notably, in the case Aytugrul v The Queen (see Table 1).

Y-STR DNA profiling

The Y chromosome is a paternally inherited sex-determining chromosome. Y-STR profiling can also be used to narrow familial searches where a number of partial matches are obtained. It has forensic application in distinguishing male and female DNA (or distinguishing multiple male offenders) in sexual assault cases where samples taken from the victim are mixed and it is difficult to obtain an accurate profile of the suspect (Roewer 2009). Y-STR profiling was used in the Australian case R v Priestley to separate male and female DNA and identify the DNA profile of the offender (see Table 1).
Low copy number analysis

The amount of DNA left on an object after contact is influenced by a range of factors, such as the duration or type of contact. Low copy number (LCN) analysis involves the use of techniques to multiply small amounts of DNA, obtained from only a few cells. This technique was applied in the high-profile Murdoch v The Queen case in the NT Supreme Court (see Table 1). However, it has the potential to create erroneous results due to a high potential for contamination (Gans 2007; Lowe et al. 2003).

DNA phenotyping

In comparison with the other forms of DNA profiling discussed above (which utilise non-coding regions), DNA phenotyping uses coding regions of the genome. It has been less widely used than other techniques and has been more controversial. This is because DNA phenotyping can determine whether an individual has specific genes that are relevant to physical features such as hair and eye colour, height, ethnic background, facial features, or predisposition to specific psychological conditions (Kayser & Schneider 2009; Koops, Prinsen & Schellekens 2006).

DNA phenotyping could be used in cases where a DNA match cannot be found on a database, and provides the option of obtaining information about a suspect where there is little evidence. This technique is legal and regulated in parts of the United States and the United Kingdom. As of September 2014, the US National DNA Index System (NDIS) contained 11,164,117 convicted offender and detainee profiles, 2,026,761 arrestee profiles and 583,444 forensic profiles. The Combined DNA Index System (CODIS) had 261,703 hits between individuals and crime scenes associated with 250,230 investigations (FBI 2014).

As of 30 June 2014, the United Kingdom’s National DNA Database (NDNAD) contained 4,936,021 individual profiles, 5,753,148 suspect samples and 463,562 crime scene samples. There were 477,966 crime scene matches for one or more subjects for all offences, 3,153 crime scene matches to one or more subjects for murder or manslaughter and 6,972 crime scene matches to one or more subjects involving rape cases (Home Office 2014).

In November 2014, the Commonwealth Minister for Justice, the Hon Michael Keenan, announced that Australia had entered into a pilot program with the United Kingdom, the United States and Canada enabling international sharing of DNA profiles (Keenan 2014).

The large number of DNA profiles collected and stored by law enforcement agencies and the extent to which police regularly use this information to investigate serious crimes demonstrate the value of DNA evidence as an investigative tool. One of the gaps in research undertaken to date on the use of DNA is developing an evidence base to support the rapid expansion of DNA databases and application of new techniques.

Empirical evidence

While there is considerable literature describing the techniques of DNA profiling from a forensic science perspective, there is a lack of empirical research available on the contribution of DNA evidence in criminal investigations. It is important to understand how police use DNA evidence during investigations and the impact of DNA profiling on criminal justice outcomes (Julian et al. 2011).

Dunsmuir, Tran and Weatherburn (2008) undertook empirical research using Australian data for the NSW Government. This evaluation examined the impact of mandatory DNA testing of prison inmates in New South Wales (and the associated expansion of the DNA database) on a range of outcomes including clear-up rates (the percentage of cleared incidents within 180 days), charges (the percentage of recorded incidents resulting in charges within 180 days) and charge to clear-up rates (the percentage of cleared incidents resulting in charges within 180 days) across eight crime types. For sexual assault, robbery with a firearm, robbery without a firearm, break and enter dwelling and break and enter non-dwelling, DNA profiling was found to have consistently positive impacts for clear-up, charge and charge to clear-up rates. For sexual assault, a positive linear trend was identified, starting in mid 2002, and resulting in an 18 percent improvement in the clear-up rate, and a 50 percent improvement in the charge rate 12 months later. For robbery with a firearm, a positive linear trend was identified in mid to late 2005, resulting in a 53 percent improvement in the clear-up rate, and a 70 percent improvement in the charge rate 12 months later. The contribution of DNA evidence to the offences of assault, motor theft and stealing from a motor vehicle was less clear. Overall, the study demonstrated there are improvements to investigative outcomes with mandatory DNA testing and the expansion of DNA databases.

DNA databases

The Australian National Criminal Investigation DNA Database (NCIDD), managed by CrimTrac, has been in operation since 2001. The NCIDD continues to expand and now holds more than 830,000 DNA profiles and conducts 68,000 matches from crime scenes to individuals, and more than 72,000 matches from crime scene to crime scene each year. The NCIDD is used by police in all jurisdictions to upload profiles and conduct searches (CrimTrac 2014).

In 2015, the NCIDD will be upgraded to include additional capabilities. This database will be among the most advanced in the world, incorporating familial searching and kinship matching capabilities, as well as mtDNA and Y-STR profiling (CrimTrac 2014).

Most developed countries have DNA databases, with the largest in the United States and the United Kingdom. As of September 2014, the US National DNA Index System (NDIS) contained 11,164,117 convicted offender and detainee profiles, 2,026,761 arrestee profiles and 583,444 forensic profiles. The Combined DNA Index System (CODIS) had 261,703 hits between individuals and crime scenes associated with 250,230 investigations (FBI 2014).

As of 30 June 2014, the United Kingdom’s National DNA Database (NDNAD) contained 4,936,021 individual profiles, 5,753,148 suspect samples and 463,562 crime scene samples. There were 477,966 crime scene matches for one or more subjects for all offences, 3,153 crime scene matches to one or more subjects for murder or manslaughter and 6,972 crime scene matches to one or more subjects involving rape cases (Home Office 2014).
Roman et al. (2008) conducted a randomised study of the effect of DNA technology in property crimes across five locations in the United States. The study was rigorous, including random assignment to experimental (treatment) and control conditions, reaching the highest level on the Maryland Scientific Methods Scale (SMS) (Farrington et al. 2002). In both treatment and control groups, traditional investigative techniques were used; however, the treatment group also incorporated DNA profiling during investigations. These results indicated that the use of DNA evidence in property offence investigations resulted in twice as many suspects identified, arrests and cases accepted for prosecution. Further, it was found that DNA profiling was five times more likely to result in the identification of a suspect compared with fingerprint evidence.

Briody (2002, 2004, 2006) examined the impact of DNA evidence on court outcomes for sexual offences, homicide and property offences in Queensland. In each of these studies, 150–200 cases were included, half where DNA evidence was presented and the remaining half assigned as comparison-control cases. In sexual offence cases, DNA evidence doubled the likelihood that a case reached court, and the presentation of DNA evidence by the prosecution resulted in a 33-fold increase in the likelihood that a jury would find the offender guilty. The presence of DNA evidence also increased the likelihood of a custodial sentence being imposed and, on average, increased the length of the custodial sentence by two months. In relation to homicide cases, those involving DNA evidence were more than 14 times more likely to reach court, and juries were more than 23 times more likely to convict. Finally, for property crimes, the study indicated that cases with DNA evidence had an increased probability of reaching court, and the offenders were more likely to plead guilty.

In an international review that included the research evidence described above, Wilson, Weisburd and McClure (2010, 2011) concluded that there is strong empirical evidence to support the use of DNA testing in the investigation and resolution of crime, including property offences and serious offences such as assault, homicide and rape. The empirical evidence demonstrates that DNA profiling does have a positive impact on criminal justice outcomes.

As discussed, DNA evidence has been found to significantly increase the likelihood that a defendant will be convicted at trial (Briody 2004). However, interviews with real jurors and jury simulations have questioned whether jurors understand and apply the science presented at trial (Findlay & Grix 2003). Further, Goodman-Delahunty & Hewson (2010) found that the use of DNA evidence tripled the conviction rate in comparison with an identical case that involved inconclusive DNA evidence. One option that has been proposed to address this is to provide standard tutorials for jurors prior to cases to ensure a sufficient level of understanding (Wheate 2008). These issues are particularly important in the context of the new forms of DNA profiling that have been discussed in this paper. It is important that jurors understand new techniques when deciding guilt or innocence in cases where these forms of DNA evidence are presented.

### Significant cases

Gans and Urbas (2002) examined case law on DNA evidence from its first use in Australian criminal proceedings in 1989 through to 2002. These cases establish the legal admissibility and use of DNA in criminal trials, as well as issues such as the validity of commercial profiling kits (R v Gallagher [2001] NSWSC 565, the ‘prosecutor’s fallacy’; R v Keir [2002] NSWCBA 30), the scientific acceptability of techniques such as polymerase chain reaction (R v Jarrett 1994 62 SASR 443) and cases that have been overturned on the basis of new DNA evidence (R v Button [2001] QCA 133). More recent DNA evidence cases in Australia and overseas demonstrate that the legal system will continue to adapt to apply new scientific developments in this field. Tables 1 and 2 summarise significant cases involving DNA evidence in Australian and overseas jurisdictions. These cases have highlighted the application of new techniques to solve crimes that would have previously remained unsolved, as well as issues that need to be considered when using this evidence in the future. The new techniques that have been discussed are familial searching, DNA phenotyping, mtDNA profiling, Y-STR profiling and LCN analysis. Examples of Australian and overseas cases that have applied all of these techniques between 2003 and 2014 are briefly discussed in terms of key issues and implications. As the facts of these cases demonstrate, these new techniques have made a significant contribution to the investigation of serious crimes, particularly in overseas jurisdictions.

Other cases included in the tables have had significant implications for criminal trials and evidence law. This includes considerations associated with the weight given to DNA evidence in circumstantial cases, the expansion of DNA databases, the extrapolation of statistical estimates from subpopulation databases and the continuing potential for human error such as laboratory contamination and DNA transfer to impact on the validity of DNA profiling. All of these issues indicate that, despite ongoing technological advancements, DNA evidence must continue to be considered as only one part of the overall evidence in a case, and that obtaining and using DNA evidence must follow established protocols and legal principles.

Overall, this summary of significant cases highlights the need to continually review new case law on DNA evidence and maintain an awareness of new technology that can be applied in the field of DNA evidence, both in terms of their potential contribution to investigations and an awareness of potential issues that could arise.
Investigations. Both the empirical evidence and case law have demonstrated the valuable contribution of DNA evidence in the criminal justice system. These benefits also appear to extend to civil matters, such as the identification of victims in disasters. Further quantitative research to establish how new techniques such as familial searching, Y-STR profiling and mtDNA profiling contribute to investigation and prosecution outcomes beyond the analysis of individual cases would be beneficial. Further research that builds on previous studies and examines the impact and cost-effectiveness of large-scale DNA databases would also contribute positively to the available evidence.

Analyses of DNA evidence cases in Australia and overseas in the past decade demonstrate that the legal system has largely adapted to and accommodated new scientific developments in this field. The techniques reviewed in this paper have made significant contributions to criminal investigations. However, issues such as the weight given to DNA evidence in circumstantial cases, the extrapolation of statistical estimates from subpopulation databases and sample contamination all impact on the validity of DNA profiling. Despite ongoing technological advancements, DNA evidence must continue to be considered as only one part of the overall evidence in a case.

### Table 1 Summary of cases that highlight legal issues in the use of DNA evidence, 2003–14

<table>
<thead>
<tr>
<th>Year</th>
<th>Case</th>
<th>Citation</th>
</tr>
</thead>
<tbody>
<tr>
<td>2007</td>
<td>Hillier v The Queen [2007] HCA 13</td>
<td>Hillier was found guilty of the murder of his former partner (and mother of his two children) after DNA was located on the victim’s pyjamas. It was argued by the prosecution that Hillier was motivated to murder his wife because he lost custody of their children. He was alone on the night the murder occurred, but there was circumstantial evidence consistent with his innocence. The defence argued that the DNA found on the victim’s pyjamas could have been transferred via their children. The conviction was quashed on the basis that the DNA evidence was considered in isolation from the other evidence. The weight accorded to DNA evidence must be considered in the context of all of the evidence presented by the prosecution.</td>
</tr>
<tr>
<td>2007</td>
<td>Murdoch v The Queen [2007] NTCCA 1</td>
<td>In a remote area of the Northern Territory, the accused signalled for a car to pull over and proceeded to shoot one victim and abduct the other. The accused relocated the vehicle driven by the victims, leaving DNA on the steering wheel and gearstick. Biological samples were analysed using the low copy number (LCN) technique. Analysis showed that DNA taken from the gearstick was from two individuals—best explained by the DNA profiles of Murdoch and the deceased. LCN analysis was contested at trial due to uncertainty about the reliability of the technique. There were two legal issues. The first concerned the admissibility of LCN evidence. It was accepted that LCN has general acceptance within the scientific community and could be admitted as evidence. The second issue was contamination. It was decided that the possibility of contamination could be excluded beyond reasonable doubt.</td>
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<tr>
<td>2009</td>
<td>R v Jama (Unreported, Supreme Court of Victoria, Court of Appeal, 2009)</td>
<td>A woman was found unconscious in a toilet cubicle. While she did not have any recollection of what had occurred, she believed she was sexually assaulted and underwent a medical examination. DNA was linked to an individual who was later convicted of raping the woman. The court later found that the DNA sample taken from the woman was contaminated, and it was likely that no rape had occurred. The same doctor who examined the woman had taken swabs from another woman 28 hours earlier, who had engaged in sexual activity with the accused; however, no changes were made in relation to the earlier event. Jama was incarcerated for 16 months before the miscarriage of justice was discovered (Vincent 2010). The sole evidence in the crime was contaminated DNA evidence. The case highlights the potential for miscarriages of justice to occur when too much reliance is placed on DNA evidence, or it is the sole evidence in a case.</td>
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<tr>
<td>2010</td>
<td>Forbes v The Queen [2010] HCA Trans 120</td>
<td>Forbes was convicted of sexual assault. DNA found on the victim’s trousers and bra was the sole evidence linking him to the crime. Two expert witnesses testified that there was ‘extremely strong’ and ‘strong’ evidence that the DNA profile on the victim’s clothing belonged to Forbes. This evidence was challenged on appeal. It was not possible to prove the guilt of Forbes beyond reasonable doubt. Statistical estimates of the frequency that Forbes’ DNA profile occurs in the population were calculated on the basis of a sample of 620 people (Gans 2011).</td>
</tr>
<tr>
<td>2012</td>
<td>Aytugrul v The Queen [2012] HCA 15</td>
<td>Aytugrul was convicted of murder after mitochondrial DNA analysis of a hair sample found under the victim’s thumbnail. The conviction was appealed because the DNA evidence was presented in court as an exclusion percentage of 99.9 percent, as well as a frequency ratio. The prosecution said to be unfairly prejudicial. The probability of the DNA belonging to the accused was presented in court as an exclusion percentage of 99.9 percent, as well as a frequency ratio. The defence argued that presenting the exclusion percentage of 99.9 percent would direct a jury to round this figure to one, indicating certainty of guilt. The main issue concerns how DNA evidence is presented in court. In this case, presenting an exclusion percentage caused the probative value of the DNA to be outweighed by its prejudicial value (Ustras 2012).</td>
</tr>
<tr>
<td>2014</td>
<td>Fitzgerald v The Queen [2014] HCA 28</td>
<td>During a burglary, one victim was murdered and serious brain injuries were inflicted on another. Fitzgerald was convicted of murder after DNA evidence linked him to a digger knife at the crime scene. There was no other evidence linking him to the crime. The conviction was appealed on the basis that the verdict was unreasonable as there were other possible ways the DNA could have been transferred. The DNA evidence was not sufficient to prove guilt beyond reasonable doubt, because there was no information about the circumstances in which the DNA was transferred. The DNA could have been deposited as a result of secondary transfer such as shaking hands with someone who did participate in the burglary.</td>
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</table>

### Future directions and implications for policy and practice

This paper has briefly reviewed developments in DNA evidence in Australia in the past decade. This has included new scientific techniques and a review of empirical evidence about the contribution of DNA evidence in the criminal justice system. Significant cases involving DNA evidence in Australia and overseas have also been examined.

The paper has highlighted recent techniques and how they can be applied in investigations. Both the empirical evidence and case law have demonstrated the valuable contribution of DNA evidence in the criminal justice system. These benefits also appear to extend to civil matters, such as the identification of victims in disasters. Further quantitative research to establish how new techniques such as familial searching, Y-STR profiling and mtDNA profiling contribute to investigation and prosecution outcomes beyond the analysis of individual cases would be beneficial. Further research that builds on previous studies and examines the impact and cost-effectiveness of large-scale DNA databases would also contribute positively to the available evidence.

Analyses of DNA evidence cases in Australia and overseas in the past decade demonstrate that the legal system has largely adapted to and accommodated new scientific developments in this field. The techniques reviewed in this paper have made significant contributions to criminal investigations. However, issues such as the weight given to DNA evidence in circumstantial cases, the extrapolation of statistical estimates from subpopulation databases and sample contamination all impact on the validity of DNA profiling. Despite ongoing technological advancements, DNA evidence must continue to be considered as only one part of the overall evidence in a case.
Table 2 Summary of cases involving the application of new DNA techniques, 2003–14

<table>
<thead>
<tr>
<th>Year</th>
<th>Case Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>2003:</td>
<td>R v Harman (Unreported, United Kingdom)</td>
</tr>
<tr>
<td>2011:</td>
<td>R v Delroy Grant (Unreported, United Kingdom)</td>
</tr>
<tr>
<td>2012:</td>
<td>Maryland v King [2012] Supreme Court of the United States</td>
</tr>
<tr>
<td>Ongoing:</td>
<td>Franklin (United States)</td>
</tr>
</tbody>
</table>

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All URLs are correct at May 2015.


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Miller G 2010. Familial DNA testing scores a win in serial killer case. Science 329: 262
Miller G 2010. Familial DNA testing scores a win in serial killer case. Science 329: 262