THE ROLE OF DNA PROFILING IN CRIMINAL INVESTIGATION

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Introduction

DNA profiling has attracted a good deal of public attention in the last eight years. The practical application of DNA technology to the identification of biological material has had a significant impact on forensic biology, because it enables much stronger conclusions of identity or non-identity to be made. Legislation regulating the taking of blood samples from suspects has been enacted in Victoria (Crimes (Blood Samples) Act 1989; Crimes Legislation (Miscellaneous Amendments) Act 1991), and further amendments are proposed. Databases of DNA profiles are being compiled as an aid to criminal intelligence. Faced with powerful incriminating evidence, defendants are often persuaded to change their plea or else mount a legal challenge to the admissibility of DNA profiling.

This paper will firstly outline the procedures used in DNA profiling, without dwelling too much on the technical details. The basis of the technology can be described simply, and is not in dispute (see, for example, the decisions in People v. Castro, 545 N.Y.S. 2d, Supreme Court of New York, Bronx, 1989; R v. Lucas (1992) 2 V.R. 109). This will enable us to focus more clearly on the real issues in DNA profiling.

Contrary to popular perceptions, DNA profiling in its present form cannot identify an individual. This illusion was created by commercial organisations intent on marketing competing DNA profiling systems (see for example, Lander, 1989), and was fostered by the news media.

What is DNA?

DNA in an individual's chromosomes controls an array of visible characteristics (including race, colouring and sex) as well as invisible characteristics (such as blood groups and susceptibility to inherited diseases). The DNA is the same in all of the cells in the body of an individual.

It is almost certainly true that each person's DNA is unique (unless he or she has an identical twin). However, current techniques only allow an examination of a small part of the total variation between individuals.

The structure of DNA is a double helix, in which two strands are twisted around one another. Each strand is composed of a linear arrangement of building blocks called nucleotides or bases. There are four types of base: adenine, thymine, cytosine and guanine, represented by the letters A, T, C and G. The two strands are held together by specific and mutual attraction between the bases. Adenine in one strand only attracts thymine in the opposite strand, forming an A-T pair, and cytosine only attracts guanine, forming a G-C pair. The pairs of bases form bridges between the two strands, and are analogous to the rungs of a spiral staircase.

The sequence of the bases along one of the two strands constitutes the genetic code. A cell reads this code like a recipe. The recipe is basically the same in all members of the same species.
In higher organisms, including humans, only a small part of this code seems to contain meaningful instructions. The vast majority of the DNA in a cell has no known function, and this non-coding DNA varies very greatly from one individual to another. This is the DNA which is examined in most DNA profiling systems.

Because the highly variable, non-coding regions of the DNA are not associated with any known characteristics, no information about the individual can be gleaned by analysing these regions. There are two important consequences of this fact. Firstly, DNA profiling is a comparative technique. Analysing a bloodstain found at the scene of a crime tells us absolutely nothing about the perpetrator of the crime. It is similarities between a crime scene profile and a suspect's profile that indicates whether he is a possible source of the blood at the scene.

Secondly, collections of DNA profiles comprising a library or database can only be used in comparisons with other DNA profiles. No information about the individuals to whom the DNA profiles belong, or their relatives, can be obtained from such a database.

**How Are DNA Profiles Obtained?**

Small, highly variable areas of DNA in different chromosomes are examined. The section of DNA that contains the chosen area must be identified. There are two main techniques which may be used to do this: Restriction Fragment Length Polymorphism (RFLP) analysis (Wyman & White, 1980; Jeffreys et al. 1985; Baird et al. 1986), and Polymerase Chain Reaction (PCR) (Saiki et al.,1985).

In RFLP analysis, the long DNA helix is cut into a set of fragments. This can be done with one of a number of specific enzymes called restriction endonucleases, which cut the DNA of all humans at specific points in the DNA base sequence. Thus a set of fragments is obtained, one of which contains the region of interest. This fragment is identified using a short piece of synthetic DNA called a probe. The probe is designed so that its base sequence is complementary to (that is forms A-T and G-C pairs with) the base sequence of one of the two strands in the fragment of interest. The probe seeks out and binds to this fragment to the exclusion of all the other fragments. The fragment to which the probe is bound can be detected by attaching a chemical label (usually a radioactive atom) to the probe. The radiation emitted by the label makes a visible image on X-ray film.

The second way of identifying the fragment of interest is to use the Polymerase Chain Reaction. Short, specific, synthetic DNA molecules called primers are constructed so that their base sequences are complementary to the sequences of two areas, one at each end of the region of interest. The primers therefore bind to the ends of this region of the DNA, and an enzyme called a polymerase is then used to make copies of the segment of DNA between the primers. In two hours, a very large number of copies can be made, which can then be readily detected and analysed. This ability of PCR to make a large number of copies from a limited amount of starting material makes it ideal for analysing very small amounts of blood or semen, and tissue from decomposing remains in which most of the DNA is in very poor condition (Hagelberg et al. 1991; Sykes, 1991; Jeffreys et al. 1992).

**What are we looking for in these DNA fragments?**

The most distinctive features of variable DNA fragments are the length (or number of bases) and the base sequence. There are already available methods for detecting sequence differences, for example, in the HLA region (Saiki et al. 1989) and in mitochondrial DNA in humans (Wrischnik et al. 1987; Vigilant et al. 1989; Sullivan et al. 1991). Sequence analysis is undoubtedly the way of the future for forensic DNA typing.
However, at the moment, the most useful forensic DNA typing systems remain those systems which look at differences in the length of particular fragments from one individual to another. Such regions of the DNA have been found to contain multiple copies of a short DNA sequence, repeated one after another and joined head to tail (that is, in tandem). The length variation arises from different numbers of repeats of the core sequence. A region of the DNA containing a Variable Number of Tandem Repeats is called a VNTR locus (Nakamura et al. 1987).

A fragment in which the core sequence is repeated a hundred times will clearly be longer than one in which it is repeated only ten times. In theory at least, any number of repeats (up to the maximum observed for that particular VNTR locus) could occur in different individuals. The length of the fragment can be measured (and, for PCR systems, the number of repeats determined) by using electrophoresis to separate the DNA fragments on the basis of their lengths. In electrophoresis, the DNA fragments move through an agarose gel under the influence of an electric field. Shorter fragments move more quickly than longer fragments.

Specific fragments of interest can be visualised using labelled probes (in RFLP analysis) or simply by staining (for PCR products). The resulting pattern of fragments is called a DNA profile.

Different individuals rarely have similar patterns, especially when we combine RFLP results obtained from three or more different areas of the DNA (Risch & Devlin, 1992). Therefore, if the profiles of two different samples are similar, it is likely (but not certain) that they come from the same person. The likelihood increases as more areas of the DNA are examined.

**Building up a database of DNA profiles**

Currently, the database at the State Forensic Science Laboratory contains information largely from RFLP analysis. This reflects the accumulation of five years of experience with this form of DNA typing. In this method, the lengths of the fragments can be estimated by measuring their positions in the profile. This can be done using a digitising tablet and a computer. The resulting digitised profile can be stored in a computer database.

It is then possible to search through the database for matching profiles. This is done for several reasons.

Firstly, it indicates how common or rare is a particular profile. The rarer the profile, the stronger the evidence that it comes from a person whose profile it matches.

Secondly, if samples recovered from several crime scenes or victims have the same profile, it is likely that the same offender was responsible. DNA profiling was used to link together some of the rapes to which George Kaufman eventually pleaded guilty in 1989, and in the investigation of the Armadale, Parkdale and Frankston rapes to which Stephen Brown pleaded guilty in 1993. The State Forensic Science Laboratory is currently putting in place resources to allow the inclusion in the database of the large number of unsolved rapes which occur every year. It is anticipated that several more serial offences will probably be revealed by the recurrence of the same DNA profiles.

Thirdly, the profile obtained from the scene of an unsolved crime might match that of a person who had been charged in relation to a previous crime and whose profile was recorded in the database. Under current Victorian legislation, DNA profiles obtained from suspects can only be retained in such a database if the suspect is charged with a relevant offence within six months (or if an extension of this time limit is granted by a magistrate). The profile cannot be retained if the person is subsequently acquitted or successfully appeals against conviction, and there is no provision for obtaining blood samples from offenders after conviction.
Fourthly, it is feasible to set up a database of missing persons or their relatives, and to look for profiles which match those of unidentified bodies. It has even been suggested that members of aircraft crews and the armed services should have their DNA stored to aid in identification of victims of disaster or war (Weedn, 1991).

It should be pointed out that DNA profiling can assist in the identification of a body only if DNA samples are available that are known to come from the deceased person, from the parents of the deceased, or from a spouse and child of the deceased.

The impact of DNA profiling on the criminal justice system

In Victoria, approximately two thirds of all cases in which DNA profiling has been used have been sexual offences (Table 1). However, DNA profiling is useful in only a relatively small proportion of sexual offences. Of the 621 sexual offence cases submitted to the State Forensic Science Laboratory in the two years from July 1989 to June 1991, only 97 were analysed using DNA profiling (Table 2).

There are several reasons why so few cases were analysed. In about 30% of cases, no further examination was required (usually because no-one was ever charged, or the charges were dropped). In another 30% of cases, no suitable semen stains could be found.

Most sexual offences are fought on consent, and identification of the source of semen is not an issue.

Sometimes the semen sample may not be connected with the offence. For example, semen stains were found on a blanket from a bed on which an elderly widow said she had been raped by an intruder. A DNA profile of one of the stains was different from the DNA profile of the suspect. The remaining seminal stains were therefore profiled, and these yielded a third DNA profile, which matched neither the suspect nor the first seminal stain. The stains on the blanket must therefore have originated from two other men. Further questioning of the victim eventually elicited the information that her two daughters and sons-in-law had slept in the bed at various times in the past, and probably after the blanket was last washed. The suspect was implicated in the attack by other evidence. Among a large number of paint stains on his overalls, there was a bloodstain. The DNA profile of the bloodstain matched that of the victim. The suspect was tried and convicted of aggravated burglary and rape.

Of the ninety-seven sexual offences in which DNA profiling was performed in that two-year period, approximately equal numbers of suspects have been included and excluded. If a particular stain in fact comes from someone other than the suspect, there is a very high probability that DNA profiling will show the difference and exclude the suspect. This fact is now widely used by investigating police to screen.

Table 1 - Types of offence submitted for DNA profiling at the State Forensic Science Laboratory, Victoria, between 1 July 1989 and 30 June 1991

<table>
<thead>
<tr>
<th>Offence</th>
<th>Number of cases</th>
<th>Proportion of cases</th>
</tr>
</thead>
<tbody>
<tr>
<td>Homicide</td>
<td>25</td>
<td>15%</td>
</tr>
<tr>
<td>Sexual Offences</td>
<td>107</td>
<td>65%</td>
</tr>
<tr>
<td>Other assaults</td>
<td>14</td>
<td>8%</td>
</tr>
<tr>
<td>Burglary, robbery etc</td>
<td>10</td>
<td>6%</td>
</tr>
<tr>
<td>Miscellaneous</td>
<td>9</td>
<td>5%</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td><strong>185</strong></td>
<td></td>
</tr>
</tbody>
</table>
Of the sexual offences analysed by DNA profiling, almost one-third resulted in a guilty plea (Table 2). The reasons for entering a plea of guilty are not always revealed, but in several cases, a guilty plea was entered after incriminatory DNA evidence was present prior to or at the committal proceedings.

Twenty-two per cent of the cases went to a full trial. Most of these cases were fought on consent: identity was an issue in only 8 of the trials. Only in three of these eight cases, did DNA profiling provide positive evidence of a connection between the accused and the offence (no results were obtained in the other 5 cases) (Table 3).

In a few cases where identity was at issue in the trial, the defence has sometimes challenged the admissibility of the DNA evidence (Roberts 1993). A defence challenge was
upheld by a Justice in only one Supreme Court case in which the State Forensic Science Laboratory sought to present DNA evidence (R v. Lucas 2 V.R. 109 (1992)). In 1989, the dismembered body of James Pinakos was found buried in two packages on a beach. A bloodstain was found at the house where the accused had been living several months earlier when Pinakos disappeared. A DNA profile obtained from the bloodstain could not be compared with tissue from the body because the latter was too badly decomposed. However, comparison with the DNA profiles of the parents of the deceased indicated that the bloodstain could have originated from one of their children. The defence challenged the basis of the calculation of the probability of parentage in this case, and the prosecution decided to withdraw any probability estimates. The judge then ruled that the DNA evidence could not be admitted because a jury would have no means of assessing the significance of the match between the bloodstain and the parents' DNA profiles.

Since the Lucas case, several studies have substantiated the method of calculating probability estimates from general population databases (Risch & Devlin, 1992; Evett & Gill, 1991; Evett & Pinchin, 1991; Devlin et al. 1990, 1993; Balazs et al. 1990, 1992; Chakraborty & Kidd, 1991; Buckleton et al. 1991). The State Forensic Science Laboratory has gathered population data for the DNA typing systems in use at the Laboratory, from a representative sample of the Victorian population (Gutowski et al. 1990). The Laboratory has validated the criteria used for declaring a match or mismatch between profiles (Roberts et al. 1992). Subsequently, three judges in the County Court of Victoria and one Supreme Court judge in Queensland have ruled that the qualifications and expertise of forensic scientists from the State Forensic Science Laboratory in the areas of statistics and population genetics are adequate. The courts have overwhelmingly accepted that DNA evidence is of assistance to a jury, and have decided that a jury would be able properly to evaluate the weight of such evidence (R v. Le Gallienne, County Court of Victoria, June 1992; R v. Soper, Supreme Court of Queensland, July 1992; R v. Percerep, County Court of Victoria, August 1992).

**Conclusion**

It is evident from this analysis that DNA evidence is rarely crucial at the trial. The main value of DNA profiling is in the earlier stages of an investigation, before the trial. It is a powerful investigative tool for excluding people falsely suspected of involvement in a crime. It can provide very strong evidence of involvement, and this is often enough to induce the defendant to plead guilty, or to fight the case on consent.

**References**


