The Parliamentary and Scientific Committee

INFORMATION FOR MEMBERS

From the

Parliamentary Office of Science and Technology

Briefing Note

29

November 1991

DNA PROFILING IN FORENSIC SCIENCE

Since 1986,DNA profiling techniques have been increasingly used in forensic science. While there is no dispute over the ability of this test to provide very strong evidence of guilt or innocence, debate continues over its effective use, quality control and the use of the information generated.

This briefing note reviews the current status of DNA profiling and highlights some of the issues which arise from its use in criminal cases.

DNA PROFILING

Traditional genetic markers, such as ABO blood groups have been used in forensic work since the turn of the century. While these tests can exclude a suspect, they are not sufficiently discriminatory to positively identify a suspect beyond doubt. In contrast, DNA profiling can identify an individual with virtual certainty[1], through analysis of their DNA in samples of body tissues and fluids (see box).

Since its initial use in the UK in 1986, DNA profiling has expanded as a means of providing conclusive evidence in criminal casework. The number of laboratories offering DNA profiling services has also increased (see Figure 1). These trends are mirrored overseas; forensic science laboratories in all EEC countries, as well as Scandinavia, Switzerland, Austria, Canada and elsewhere, carry out DNA profiling, as do 27 crime labs in the USA. Comprehensive data on the use of DNA profiling in the UK are not available, but the number of cases recorded by the Home Office Forensic Science Service (HOFSS) rose from 4 in 1986 to ca. 700 in 1990. At the Metropolitan Police Laboratory, 420 cases were submitted in 1989 and 540 cases during 1990.

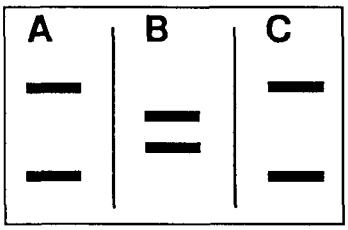
DNA profiling is used primarily to resolve serious crimes against the person (particularly rape - see Figure 2). Biological material at the scene of the crime such as bloodstains, semen, hair roots or other body tissues or secretions, may all contain sufficient DNA for profiling. Mixtures of biological fluids can be reliably analysed: a rapist's DNA profile can be separated from that of his victim; men involved in multiple rape can be separately identified. In cases involving sexual abuse of children, DNA profiling can provide evidence of sexual assault. In criminal paternity cases, DNA profiling may prove

❑ DNA Profiling Technology

With the exception of identical twins, each person is genetically unique. Our chromosomes are made of the hereditary material DNA (see Briefing note 15) which contains 3,000 million 'base pairs' whose order provides a complete set of instructions, or genetic blueprint for each person. Our genetic uniqueness results from the many small differences in the precise sequence of the bases, many of which are concentrated at certain points in the DNA (called hypervariable loci) where there is very high variability between individuals.

DNA profiling concentrates on these hypervariable loci and distinguishes between the various forms that exist. If one form is only found in 1% of the population, then the chance of DNA profiles from two people matching by chance is 1 in 100. If further tests are carried out at other hypervariable loci, then the chances of an exact match between individuals decrease accordingly. Thus a match between **unrelated** people based on 3 tests could be certain to a level of 1 in a million. (With close relatives however, the ability to discriminate is reduced because of similarities in their DNA). A single test to compare the variations in the DNA at a single location uses a single locus probe (SLP). Those which look at several locations use multilocus probes (MLPs) or a series of SLPs.

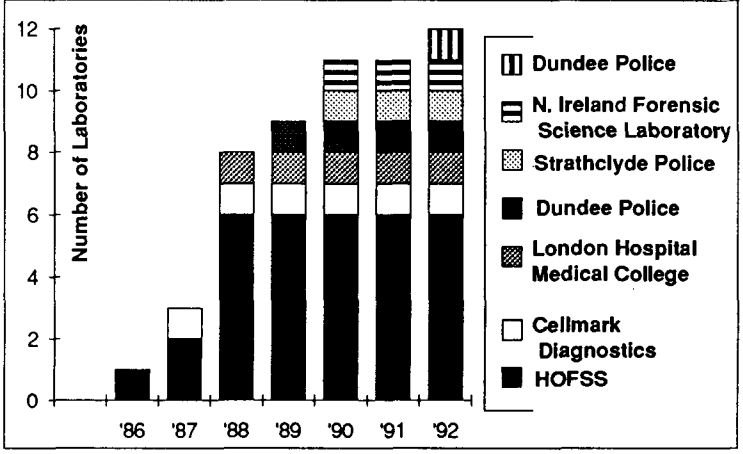
In practice, human DNA extracted from blood, semen, mouth swabs, etc., is cut into discrete fragments by 'restriction' enzymes. The different sized pieces are separated according to length by a process called gel electrophoresis and transferred to a nylon membrane. Those fragments which originate from the locus concerned are identified via a radioactively- labelled probe. MLPs give a picture on X-ray film which resembles a bar code; each SLP gives two bands as shown:



A: Profile from semen stain from scene of crime
B and C: Profiles from blood samples from suspects B and C, showing that there is a match between sample A and suspect C.

New PCR (Polymerase Chain Reaction) techniques which amplify very small quantities of DNA will soon be available. These allow tests on microscopic samples and also reduce the time for a profile in casework from 2 months to 1 week. In the latest developments (from Prof. Jeffreys' laboratory[1]), the results can be expressed as a digital code - i.e. a 'number' which is 50-70 digits long and therefore offers the potential of being a unique identifier.

Figure 1.
NUMBER OF UK FORENSIC SCIENCE DNA PROFILING LABS.



1. Also known as DNA fingerprinting, the initial technique was developed in 1984 by Professor Alec Jeffreys of Leicester University.

that a man is the father of a child conceived as a result of unlawful sexual intercourse, incest or statutory rape. Occasionally, DNA profiling is used to identify a body, particularly of a missing child, since the DNA profile of the remains can be matched to those of the possible parents (see Table 1).

Although DNA profiling is often reported as powerful evidence for the prosecution, it can be equally effective in establishing a suspect's innocence. The Metropolitan Police (Met), the HOFSS and the US FBI have all found that a suspect's DNA differs from that in crime samples in about 25% of cases - thus exonerating suspects identified through circumstantial evidence, fallible testimony or false confession. In cases of sexual assault, its use may increase the likelihood of a guilty plea, thus sparing the victim the stress of a court appearance. DNA profiles may be commissioned by the defence under legal aid.

VALIDITY AND RELIABILITY

The utility of any forensic test is closely linked to its scientific validity and reliability. In the case of DNA profiling, there is no dispute over the validity of the genetic and molecular principles underlying the tests; indeed the US Congress' Office of Technology Assessment concluded that "questions about the validity of DNA typing are red herrings that do the courts and public a disservice". Reliability is however, dependent on maintaining the quality of analytical procedures, and also on the methods of interpretation used.

Evidence in court is presented by the expert witness in terms of a statistical probability - the odds that a match could have occurred by chance. This is calculated by combining the results of several single locus probes (SLP - see box on p1), each of which produces two bands which occur with known frequencies in the population. These frequencies are determined through studies of population genetics, and vary somewhat with different ethnic groups. Separate population genetics statistics have thus been collected for the main groups (white Caucasian, Asian and Afro-Caribbean) to give reliable estimates of band matching. In a match based on 3 or 4 SLPs, the likelihood that someone other than the suspect could have been responsible for the crime sample is (except with relatives), often as low as 1 in a million.

CRIMINAL RECORDS AND DATA STORAGE

DNA profiles using single locus probes can be expressed simply as a group of numbers (these relate to the size and position of the DNA bands), and are readily amenable to storage, search and retrieval using computer databases. This also applies to the digital codes produced by new PCR techniques (see box), Stored DNA profiles can prove extremely useful in tracking down re-offenders, especially in cases of serial rape or indecent assault, where recidivism is common.

Table 1. THE USE OF DNA PROFILES

1. A man confessed to one of two linked murders. He was cleared when his DNA profile did not match that of semen at the scenes of the crimes.
2. A serial murderer avoided giving blood in a mass screening programme, but confessed when the deception was discovered and his DNA profile matched that of semen at the crime scenes.
3. A man arrested for rape pleaded guilty to murder when his DNA profile was found to match that of evidence from an unsolved murder.
4. A partial match between a rape suspect's DNA and that from semen on the victim's underwear implicated a close relative; the suspect's brother's DNA was found to match.
5. Bones from a body buried for 8 years yielded a DNA profile. Comparison with the profiles of the possible parents showed that the corpse was almost certainly that of their missing daughter.

The Home Affairs Committee recommended in 1989 that DNA profiles from those convicted of a serious arrestable offence should be stored as part of their criminal records. Currently, two databases are maintained - one is to provide population frequencies where all profiles taken are stored anonymously, the other is used for criminal intelligence purposes and contains only profiles of those convicted, charged and awaiting trial, and profiles from unsolved cases. Once court proceedings are completed or a case is solved, the relevant DNA profiles are reclassified and removed from the database where appropriate.

Using the (criminal) database, links have been identified between rapes and other offences. Arrangements are in hand to link the databases of the HOFSS and the Met to enable searches for links between crimes occurring in the different police jurisdictions. As yet, no operational need has been identified for a database profile to be exchanged between the UK and elsewhere.

CURRENT ISSUES

DNA Profiles as Evidence

There are a number of restrictions on the use of DNA profiling which arise from the Police and Criminal Evidence Act (PACE), 1984.

One concerns that of taking samples from a suspect for forensic analysis. DNA profiles are best conducted on a blood sample; this is classed as an intimate sample and can only be taken (by a doctor) with the suspect's consent, and only where a serious arrestable offence is involved. Where consent is refused, courts in England and Wales lack powers to require a sample to be taken, while in Scotland the suspect could, in certain circumstances, be required to give one. Blood samples are treated differently from fingerprints which can be required by police in England and Wales under PACE.

Mouthswabs are an alternative source of DNA. In Northern Ireland these are defined as a non-intimate sample (Criminal Justice Act, 1988) and may be taken without the need for consent where a serious arrestable offence is involved. In England and Wales however, a

mouthswab is an 'intimate' sample subject to the same conditions as a blood sample. While it is more difficult to get a good DNA profile from mouth swabs than from blood, PCR amplification techniques (see box on p1) may resolve this problem and the RUC hope to use them for terrorist casework from April 1992. However, even with PCR widely available, blood samples will remain the preferred source of DNA for profiling.

The police see the restrictions in England and Wales as limiting the use of DNA profiling and its value in speeding up resolution of guilt or innocence. They see a need to resolve disparities between different parts of the UK, and to consider whether the collection of blood samples might be allowed in certain circumstances (e.g. in 13 US States, police may take blood samples from convicted sex offenders). Civil liberties groups on the other hand, continue to see a need for special safeguards for 'intimate' samples and also believe that it should be compulsory to inform the suspect of their right to refuse consent. They also see the ability of PCR techniques to work with non-intimate samples as effectively allowing a DNA profile to be taken without consent, and believe this should be reviewed.

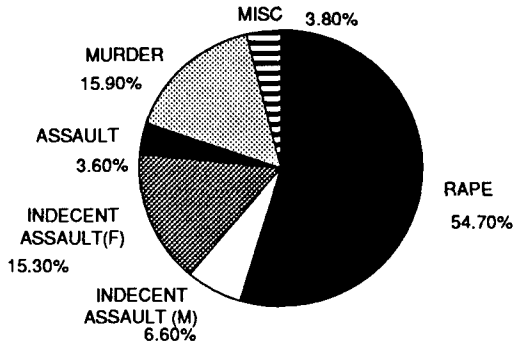
There are also problems over compliance with provisions in PACE which specify that samples from a convicted person may be retained; all others must be destroyed after they have fulfilled their purpose. These provisions do not however take into account the fact that the DNA profile test is carried out as a comparison, and therefore a single membrane (see Box) contains samples from the scene of crime and from other suspects as well as that of the convicted person. Since it is impractical to separate profiles and to destroy them would destroy material evidence, they may in some cases be retained in technical contravention of PACE.

The Royal Commission on Criminal Justice is currently considering *inter alia*, issues concerning defence access to forensic evidence. DNA profiling raises specific questions due to the time taken for a profile (currently 2 months -see box on p1). Some consider that the Crown Prosecution Service should notify the defence as soon as it is decided that a DNA profile will be used in evidence, to give the defence sufficient time for an independent analysis. Others feel that in view of the unambiguous nature of profiling results and the cost of repeating the test, the current practice (where the defence has access to the prosecution's test documentation and the scientists involved) is adequate.

Databases

The storage of DNA profiles on computer data bases is already underway (see earlier). Some see the inclusion of profiles in criminal records as involving no different principle to the storage of fingerprints, height, eye colour etc. Others see a DNA profile as describing an

Figure 2. CASES SUBMITTED FOR DNA PROFILING IN 1990 TO THE METROPOLITAN POLICE LABORATORIES



aspect of an individual's genetic make-up and express concern that data from DNA profiling could reveal functional genetic information (e.g. on susceptibility to disease) which could be of interest to other parties, such as insurance companies or potential employers.

The SLPs used at present in the UK are unrelated to functional parts of the DNA, so the profiles are unlikely to reveal useful genetic information. However, some US-made probes are related to functional regions and thus the possibility that profiles could reveal genetic information cannot be excluded. Most thus see it as important to restrict database access to law enforcement agencies. This however may be complicated by the increasing number of labs offering DNA profiling, which includes the police, HOFSS Executive Agency, and private companies such as Cellmark Diagnostics.

There are also ambiguities in the law on the maintenance and use of DNA profiling databases. Firstly, there are no clear guidelines on whether profiles from scene of crime samples or from people eliminated from enquiries should be included. Although it is against the law (Criminal Justice Act 1988) to access computer data on fingerprints of people eliminated from enquiries, there is currently no legal restriction on access to DNA profiles on computer. Many concerned with civil liberties argue that, at a minimum, laws similar to those concerning databased fingerprints and fingerprint records, should be introduced. On the other hand there is a case for keeping such information for future reference in the event of a query, and to assist in detection.

Where a database search results in a match between a DNA profile and one from a crime sample, the courts may exclude the use of that evidence. In view of the suspect's right to refuse a new sample, some see this restriction as preventing suspects from facing justice, but others feel that in view of the Data Protection Act, a DNA profile taken for one offence should not be used to incriminate a suspect for another offence.

The Home Office has stated that it will consider legal and ethical questions relating to DNA databases. But its recent scrutiny of the National Criminal Records sys-

tem contained no recommendations concerning DNA databases. The Data Protection (DP) Registrar has also supported a publicly accountable National Criminal Records Agency with statutorily regulated disclosures. In his 7th report (1991), he indicated that several issues, namely the obtaining, disclosure, relevance, accuracy, retention and security of sensitive data required careful consideration.

However, it is likely that all legislation relating to Data Protection will be reviewed once the European Commission has settled on a common data protection policy and practice. This may include consideration of the right to privacy which is not addressed by the DP Act. These rights will become even more important if, as suggested by the Home Affairs Committee, creation of a DNA profile database of the entire male population 'would provide considerable benefits for the police'.

Quality Assurance (QA) and Standards

Recent appeals have focused attention on the quality of some forensic evidence. Over the past 15 years, there has been a substantial tightening of QA procedures (developed by the HOFSS) throughout the main public forensic labs. Nevertheless, there are a number of specific technical issues related to quality assurance of DNA profiling covering technical standards (which enzyme, probes, methods of analysis including statistics, etc. should be used), accreditation of laboratories performing the tests, and training and advice in expert evidence-giving.

The first issue concerns standards used within Europe (and to a lesser extent the USA), and bears on the use of DNA profiling in transfrontier cases. The European DNA Profiling Group (EDNAP), initiated by British forensic scientists, has been responsible for encouraging technical and operational harmonisation within Europe. EDNAP now includes representatives from the major DNA profiling laboratories in Europe which have agreed to use common enzymes, probes and test conditions, to allow the results between countries to be compared.

The second issue concerns how to ensure that laboratories' QA procedures lead to reliable and accurate results. In the USA, at least one case revealed shortcomings in the technical execution and presentation of evidence so that admissibility in court was restricted. Similar cases have not occurred in the UK, although evidence from DNA profiles (e.g. the statistical probabilities quoted) has become more frequently contested by the defence.

In the UK, general quality assurance systems have been adopted by forensic science laboratories. Each court statement is checked by a peer scientist or one of greater seniority before it is released. 'Blind' trials are used to independently verify laboratory techniques. Specific QA procedures for DNA profiling have been developed by the HOFSS and independent laboratories (principally Cellmark). Through EDNAP, other quality assurance initiatives have been developed for European DNA profiling laboratories.

In their 1989 report, the Commons Home Affairs Committee felt it would be premature to consider creation of a statutory body to regulate standards in the FSS. Since then, the move of the HOFSS to an Executive Agency and the devolved power of police forces to purchase their own forensic services may encourage further local provision of DNA profiling services. The Met, HOFSS and Strathclyde Police are currently moving towards accreditation with the National Measurement Accreditation Service (run by the DTI), which deals principally with meeting laboratory and technical standards and the proper documentation of procedures. BS 5750, awarded to Cellmark Diagnostics, addresses the quality of the systems in place in an organisation, auditing etc. These forms of accreditation are mutually compatible, but conforming with their standards is expensive and relatively few laboratories are likely to afford it. Another possibility is to use EDNAP's less formal quality assurance scheme operated through the HOFSS's Aldermaston facilities.

The only formal requirement for DNA profiling labs relates to paternity testing, where laboratories must employ an appointed blood tester. But this does not involve any specific consideration of QA on DNA profiling tests carried out. Many thus see an urgent need to resolve the accreditation requirements at an early stage to ensure that the credibility of DNA profiling is not endangered by one poor quality result escaping the control procedures. Some feel this should be done by requiring specific QA management systems for all DNA profiling labs, but others consider that court examination of the data or repeat tests by independent experts provide sufficient QA.

There have also been suggestions for a professional register of forensic scientists operated by a legally established body with powers to remove members who fall short of its standards, as well as calls for a Forensic Science Board to consider, *inter alia*, issues of registration and accreditation.

FURTHER READING

Additional details and background information are available from POST, 2 Little Smith St., London SW1P 3DL, tel: (071)-222-2688.