

DNA Fingerprinting

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DNA fingerprinting or profiling has been described as 'the most significant scientific advance in crime fighting since the introduction of fingerprints'. The technique can also be used for paternity testing and for a range of biological applications; an overview is provided in this paper which also describes how a DNA profile is produced, explains the strengths and possible limitations of the technique and discusses the controversies surrounding DNA evidence.

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Summary

Genetic fingerprinting has had a 'meteoric career'¹, being 'widely hailed as the greatest breakthrough in forensic science this century'². The term DNA or genetic 'fingerprinting' was first coined by Alec Jeffreys of Leicester University in 1985^{3,4}, but today the term 'profiling' is more often used, which helps avoid confusion with conventional fingerprinting and more truly indicates the type of results obtained. Although the terms 'fingerprinting' and 'profiling' are also sometimes used to denote different analysis techniques there is no hard definition and they are used flexibly in this paper.

In April 1995 the Home Office and Forensic Science Service (FSS) launched the national DNA database which the FSS has described as 'the most significant scientific advance in crime fighting since the introduction of fingerprints'⁵. The power to take non-intimate samples for DNA testing and to store the results (for persons cautioned or convicted of a recordable offence) was provided by the *Criminal Justice and Public Order Act 1994*. It is thought that there may be four million profiles on the database in five years' time⁶.

Some commentators have welcomed such moves and have even called for a database to be compiled of the entire population but others, notably civil rights campaigners, see this as an erosion of civil liberties. They fear that such information may one day be required when a person is seeking to obtain insurance, a mortgage, or employment⁷.

DNA profiling has been used in several mass screening programmes as part of murder investigations. This is only feasible if it is suspected that the murderer is one of a limited population, such as the inhabitant of a village, or a member of a certain profession. The technique can be used to confirm or disprove paternity, and between June 1995 and January 1996 500 cases dealt with by the Child Support Agency resulted in an alleged absent parent being proved to be the father in nearly nine out of ten cases⁸. The technique is increasingly being applied to novel areas such as wildlife crime (determining whether rare and expensive animals have been captive bred or illegally taken from the wild).

¹*The Lancet* vol 345 24 June 1995 'Beyond all reasonable DNA' pp1586-8

²*Nature* vol 368 24 March 1994 'How convincing is DNA evidence?' pp285-6

³Jeffreys, Wilson and Thein 1985 'Hypervariable minisatellite regions in human DNA' *Nature* **314** pp.67-73

⁴Jeffreys *et al*, 1985 'Individual specific 'fingerprints' of human DNA' *Nature* **316** pp76-79

⁵'DNA Database goes live' FSS News Release 10 April 1995 FSS 1/95

⁶*New Scientist* 19 November 1995 'Crime match'

⁷for example *Independent* 28 March 1994 'Seduced by the gene genie' p.21

⁸HC Deb 28 February 1996 cc623-4w

It can be argued that many such applications rely almost as much on the reputation of DNA profiling as on its capabilities. For instance, one way in which a murderer may be caught is by refusing to come forward for screening. Nevertheless, the reliability and significance of DNA evidence has been questioned, particularly by defence lawyers. As well as this, there has been extensive scientific debate concerning the way in which some aspects of DNA evidence should be quantified, and presented to a court.

I. Technical background

This section of the paper goes into some detail regarding profiling techniques, but an understanding is useful if the value and potential of the method and possible pitfalls are to be appreciated.

A. Individual genetic makeups

The instructions which determine our physical characteristics and run our bodies are encoded in the chemical DNA. DNA is suited elegantly to its functions⁹; it carries the genetic code written in only four letters (called 'base pairs'). A 'gene' is simply a sequence of base pairs that 'codes for' substances that a cell needs to produce.

We inherit our DNA blueprint from our parents; half from our mother (in an egg cell) and half from our father (in a sperm cell). Both carry only half the genetic material needed for an individual; on fertilisation or fusion the full complement, or 'genome', is restored.

When the egg and sperm cells with their half complements of DNA are created in the parents, the genetic material is purposefully jumbled up or reshuffled. Otherwise all sisters and brothers would receive identical sets of genetic information from each parent and on fertilisation would end up with identical genetic makeups. In practice however even two sisters or brothers, although very closely related genetically, will not have an identical genetic makeup - they will each have been dealt different genetic hands from each parent. (This is very important because evolution cannot work without variation.)

⁹see Library research paper 93/55 *Genetically modified organisms, transgenic animals and animal patenting*

Individual humans can only have identical genetic makeups if they are identical siblings (most usually twins); these result from the fertilised egg with its full restored genetic complement then splitting into two or more embryos with identical DNA sets.

Our genetic makeup directly determines some of our characteristics in a very simple and direct way (anyone who has inherited genes for blue eyes from both parents will have blue eyes), although it is far more common for features to be determined by a combination of genes acting in concert. Moreover there is often a pronounced environmental influence; the height to which we grow might be influenced by diet or exercise, although we may also have inherited a propensity to be tall from one or other or both parents.

Every cell in the human body carries a complete copy of the individual's gene set inside its nucleus, with the important exception of red blood cells which do not have a nucleus. It follows then that since everyone (other than identical twins) has their own DNA makeup, in theory just one cell will carry a 'genetic fingerprint' unique to that person and sufficient to identify that individual. The trick lies in reading the fingerprint. It is not feasible to produce a sequence of the entire individual genome; this has not yet been done even generally for humans, despite a multinational effort to that end.

B. Fingerprinting or profiling

The DNA code of an individual contains about 100,000 genes but there is plenty of space left over. Only around 20% of the genome is taken up with genes and gene related sequences, and even within the gene stretches about 90% of the DNA does not code for anything. In total probably 98% of the human genome does not code for anything or appear to have any obvious function¹⁰. This has traditionally been called 'junk' DNA and it is spliced out of the code when it needs to be used, but like the rest of the genome it is copied faithfully from generation to generation, so it can be used to identify individuals and species¹¹.

While most genes are unique (they occur only once on the genome), some stretches of the junk DNA repeat themselves again and again in the genome, for varying numbers of times. In all about one-third of the genome consists of repetitive sequences. Because these do not code for anything they are reasonably free to mutate and develop high variability between individuals, or a high degree of 'polymorphism'. Some repeats may occur only a few times per genome, but other sets may repeat a million times or more. They may vary in the number

¹⁰*DNA Fingerprinting* M Krawczak and J Schmidtke 1994 p.13

¹¹There is intense interest in the function of the non-coding DNA that occurs inside and outside gene sequences but a consideration is beyond the scope of this paper

of elements per repeat and in the positions at which they occur along the genome, giving rise to 'hypervariable regions' of DNA.

Conversely, the coding parts of the genome are able to show far less variation from individual to individual because we cannot risk losing vital instructions. While certain genes occur in different stable forms (for example the gene for eye colour exists as green, blue, brown and so on without causing any harm), mutations of other genes may have a devastating effect¹², so the coding parts of the genome are highly conserved for very good reason. It would be no use looking at such areas when trying to find differences between individuals.

The highly variable sequences which occur repetitively are known as satellite, minisatellite and microsatellite regions¹³. These three 'repeat classes' are together referred to as 'variable number of tandem repeat' (VNTR) regions. Some particular mini and microsatellites are notably variable in sequence and in the number of times they repeat in different people^{14,15}.

All in all this gives rise to regions of the genome which vary greatly between individuals but which are passed down reliably through generations according to the classic laws of inheritance; the basis of a DNA 'fingerprint'¹⁶.

C. Techniques

The traditional method of producing a DNA profile is described below but the technology is continually expanding and improving and there are now various DNA analysis techniques; in general the molecular biology revolution is probably one of the most significant advances of our time, and certainly comparable to the electronics revolution.

¹²see Library Research Paper 93/66 *Gene Therapy*

¹³Microsatellites consist of short sequences (just 1-6 base pairs) repeated 10-100 times, up to 10⁵ times per genome, while minisatellites have 9-100 base pairs repeated 10-1000 times over, occurring thousands of times per genome. Satellites, less useful in DNA profiling, are much bulkier sequences which repeat far fewer times on the genome.

¹⁴For instance, one particular minisatellite, called D1S7, has a 9 base pair repeat unit and may have over 24,000 different repeat lengths; it has also been found to exist in two forms (different versions being inherited from the mother and father) in over 99% of individuals tested so far. Another minisatellite, D1S8, has a 29 base pair repeat unit and has at least 50 different lengths. In addition it shows mutations in some of its repeat unit sequences, giving rise to further variations, and moreover the original and mutated sequences occur in different orders along the repeat giving rise to yet further variation

¹⁵*DNA Fingerprinting* M Krawczak and J Schmidtke 1994 p31

¹⁶Jeffreys AJJ, Wilson V, Thein SL 1985 Hypervariable minisatellite regions in human DNA, *Nature* **314**, pp.67-73

To produce a genetic fingerprint, first the DNA has to be extracted and purified from the cell using a variety of agents. The purified DNA is then cut up using 'restriction enzymes', which are simply biological molecules which can home in on specific DNA sequences ('recognition sequences') and cut the DNA molecule at that site. The length or size of the fragments produced vary; the basis of the size variation differs between techniques.

For example, there may be variations in the recognition sequences themselves which may mean that some are not recognised, giving rise to 'restriction fragment length polymorphism' (RFL polymorphism). Alternatively, the space length between recognition sequences may vary because of different numbers of repeating units in these areas (VNTR polymorphism).

Different sized fragments have different weights, and these can be separated by a very standard process called electrophoresis, which drags the fragments through a gel using an electric current; the smaller lighter fragments move more quickly and further. (It is rather like ink being sucked up a blotting paper.) If stained with a non-specific dye there would be a smear right along the gel corresponding to a continuous spectrum of fragment sizes. This would be of little use; specific fragments need somehow to be picked out and stained.

This is done using a 'probe'. The DNA molecule is made of two complementary strands; if split, a fragment of single strand can easily find and attach to its exact counterpart in a mixture of DNA sequences that contains up to 200 million times its own complexity. A probe then is essentially just a short DNA molecule complementary to the DNA of interest (usually the 'core' sequence of a VNTR).

So after the DNA fragments have been stretched out on the gel they are treated to remove one DNA strand. The remainder are then transferred¹⁷ to a nylon membrane where they are fixed in place, and they are exposed to a probe. This will usually be radioactively labelled so after it has attached itself to segments of particular interest it can be exposed to X-ray film to produce the profile itself.

The profile will then be a pattern of the DNA fragments which the probe has attached to, now visible as a series of black bands which look just like a supermarket bar code. The presence of a band shows whether or not a particular DNA sequence is present, and its position gives a measure of its size.

Modern techniques tend increasingly to use VNTR polymorphisms which do not require quite such good qualities or quantities of DNA because the repeat units can be copied or amplified

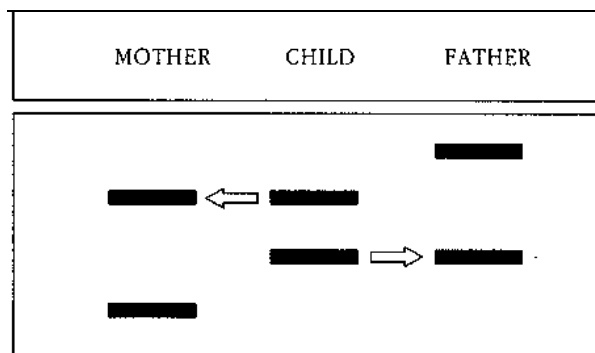
¹⁷using a technique called Southern blotting

up using a technique called PCR (see section I.E below). This may negate the need to use restriction enzymes to cut up the DNA, although it also increases the risk of errors associated with PCR creeping in. Because large quantities of DNA get generated VNTR techniques may even negate the need to use a probe to label the band, which may instead simply show up by itself.

D. Multilocus and Single locus probes

A single probe will attach to one given sequence of DNA and will thus show up as two bands in an individual's DNA profile, a band of different size being inherited from each parent (although occasionally the same sized band will have been inherited from both parents so apparently only one band will show up)¹⁸.

Figure 1¹⁹ *A child inherits half of its DNA from each parent. In this single locus probe result the child has two bands, one of which has been inherited from its mother, the other from its father.*



If possible, more than one probe will be applied. The first probe is simply washed off the nylon membrane (to which the DNA is fixed) and another probe, which is seeking to attach to a different DNA sequence ('locus'), is applied. The DNA would normally be analysed sequentially with up to four such probes. This is known as a 'single-locus' approach. Single locus probes are quite sophisticated and have been developed more recently than 'multi-locus' probes; they are increasingly the method of choice for forensic and paternity work.

'Multi-locus' approaches can involve either using several single locus probes simultaneously, or, more traditionally, by applying a probe that detects several loci. Great care must be taken that a multilocus pattern is interpreted as such and not in the same way as a superimposed

¹⁸What is DNA profiling? Forensic Science Service 1993 p.3

¹⁹Reproduced with kind permission from *DNA Paternity Testing*, Cellmark Diagnostics Zeneca 1995

single-locus pattern; there have been some 'serious' misinterpretations of such patterns.

By definition single locus probes detect unique sequences, so applying one after the other will always expand the information available. On the other hand, multi-locus probes overlap between the loci to which they hybridise, so applying several multilocus probes will **not** necessarily provide more information²⁰, even though they look more 'impressive' and detailed and are most people's idea of a genetic fingerprint. The problems of interpretation and the likelihood of chance matches occurring are discussed in section II.

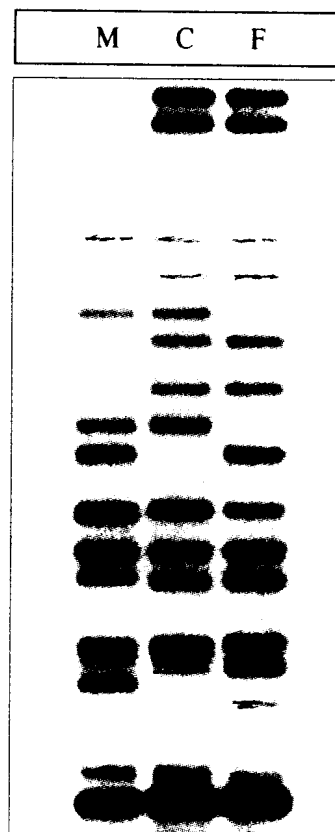


Figure 2²¹ *Multilocus probe result. Every band in the child is present in either its mother's or its father's pattern. The scanning and reproduction methods used for this paper make the bands look less distinct than the results would be in reality.*

The probes which Jeffreys used in his pioneering work²² were 'multilocus probes' called 33.6 and 33.15 which detect certain minisatellite core sequences and which, it turns out, co-detect only 1% of fragments. Several other multilocus probes have now been developed. However, single locus probes are used for forensic and paternity

²⁰*DNA Fingerprinting* M Krawczak and J Schmidtke 1994 p.32

²¹Reproduced with kind permission from *DNA Paternity Testing*, Cellmark Diagnostics Zeneca 1995

²²Jeffreys AJJ, Wilson V, Thein SL 1985 Hypervariable minisatellite regions in human DNA, *Nature* **314**, pp.67-73

work²³ and the more of these that are applied, the more discriminating a test gets (the less likely it is that a chance match is occurring; see section II.A).

Discrimination should improve all the time. Presently the Forensic Science Service (FSS) is developing the use of highly discriminating simultaneous single probe approaches²⁴. Some researchers have demonstrated a set of five probes which have a calculated probability of less than one in 3×10^{13} of producing identical DNA profiles of two unrelated individuals. These produce profiles which are quite complex in appearance but which result from simple imposition of complete single-locus patterns, not multi-locus probes, so there is no chance of co-detection or overlap²⁵.

E. Samples

DNA can be extracted from any sample that contains cells with a nucleus, so hair roots, muscle, organs and bones can be used. A swab can be taken from inside the cheek, and saliva may or may not contain enough cells. The amniotic fluid can be used if prenatal analysis is required²⁶. Because sperm cells are rich in DNA, profiling has been 'most effective in providing forensic evidence in sexual assault cases' particularly from vaginal swabs, although the technique cannot be used if the semen has come from a vasectomized man²⁷.

The main exceptions to all this are red blood cells which do not have a nucleus. This is because these are highly specialised cells designed to be expendable and rapidly produced and to do just one job, which is to carry as much oxygen around the body as possible. But of course DNA fingerprints are obtained from blood samples, and this is possible because blood contains not only red but white blood cells, plus other components. Provided the sample is fresh and large enough to contain sufficient white blood cells, an analysis can be performed.

It is worth bearing in mind that the freshness and size of the sample will have direct bearing on the ease with which DNA fingerprinting can be used; DNA will eventually deteriorate in cells outside the body. The larger the sample and the fresher it is, the better the results will be. If a post-mortem sample is badly decomposed however, it may be possible to extract DNA samples from the tooth cavities or deep muscle tissue. The DNA analysis in the recent case involving the exhumation of 'Bible John' suspect John McInnes for comparison with a sample found on the clothing of one victim has taken longer than expected. This is because

²³*What is DNA Profiling?* Forensic Science Service undated p.6

²⁴HC Deb 23 January 1996 c120w

²⁵*DNA Fingerprinting* M Krawczak and J Schmidtke 1994 p.30

²⁶*DNA Fingerprinting* M Krawczak and J Schmidtke 1994 p.17

²⁷*What is DNA profiling?* Forensic Science Service 1993 p.4

the analysis was being carried out 16 years after the death of McInnes, so the DNA would not have been in good condition and has had to be extracted from bone samples, not body fluids²⁸.

The technique called the 'polymerise chain reaction' (PCR) has revolutionised many aspects of molecular biology- it can be used to 'amplify' sections of a DNA sample which may be degraded, or to copy the minute amount of DNA found in just one cell millions of times over in a test tube, until the sample is usually large enough to be seen with the naked eye;

'The increased sensitivity offered by [PCR] enables bloodspots of approximately 1mm diameter, trace amounts of semen and single hair roots to be typed. However, such sensitivity will only be obtained with fresh, good quality samples, whilst older or degraded samples will invariably require greater quantities of material. Fifteen year old bloodstains have been successfully typed as have more specialised samples such as saliva on envelope flaps, aspermic semen, body tissue and bone²⁹.

PCR won its inventor the Nobel prize in 1994; this and other molecular biology tools are constantly being improved and will give rise to increasing sensitivity and discriminating power. PCR is usually used when looking at VNTR polymorphisms (see section I.C).

Problems may occur when a sample has arisen from more than one source and cells and tissues cannot be separated. The probes used on human DNA will not react with bacterial DNA so the presence of bacteria is not a problem. However, human probes will pick up on (hybridise to) DNA sequences from other species of 'higher' organisms. If more than one human individual is involved (mixed blood spots for instance) then typing will be needed to separate the two. Problems of contamination are further discussed in section II.E. The legalities of obtaining and retaining samples are discussed in section III.B.

²⁸eg *Herald*, 13 March 1996 'Police resist pressure on 'Bible John' DNA'; *Guardian* 16 March 1996 p.12

²⁹*What is DNA profiling?* Forensic Science Service 1993 p.5

II. Interpretation and reliability

The genetic makeup of an individual is unique, except in the case of identical twins, and the term 'fingerprint' suggests a unique and incontrovertible identifier. 'Genetic fingerprinting' was thus readily promoted and accepted as scientifically infallible on its introduction. Yet the reliability and credibility of DNA fingerprinting and profiling have been challenged in court cases, and academics have argued over and misinterpreted the statistical significance of genetic profiling. How much faith can be placed in the technique?

Imagine that two DNA profiles, from a suspect and a scene of crime sample, are presented to a court as evidence. First the jurors need to be satisfied that a match between the two samples exist; for instance, some of the bands might not be in exactly the same place. If a match appears to exist, the jurors then need to be content that an innocent or randomly arrested member of the population would not match to this by chance. Even if the forensic scientists calculate an extremely small probability that a match is by chance they then need to convey the level of significance to the jurors and to tell them exactly what this means, without misleading them. There has been a running debate in the scientific press about match probabilities.

On top of these problems are the purely technical ones such as degradation, contamination of samples or sloppy lab procedures which can give rise to false results, although many such problems arose early in the use of DNA analysis when firms started up with little experience. It should be remembered also that analysis procedures and molecular biology techniques are advancing continuously.

A. Chance match probability

The key point to bear in mind is that genetic profiles are *not* complete fingerprints or profiles of the whole genome. They are looking only at tiny parts of the genome, albeit parts which vary widely between individuals. So the technique is relying on the variation in these 'markers' being sufficient in the population for them to be enough to identify an individual. If the same band has been picked out by a single locus probe in two samples then one needs to know how many people in the population as a whole would also have that band. If four or even five unique probes are applied in sequence then of course the probability of a chance match are decreasing each time.

Scientists work out the probabilities of chance matches using a mixture of observation and statistical modelling. They take into account how often a band occurs in a 'reference DNA

database'; this is not the same as the national DNA database but a database of perhaps several hundred or a thousand or so individual anonymous profiles built up by the scientists themselves. Ideally the reference database will be of the same ethnic group as the suspect (see section II.D). There is no national reference database³⁰; organisations performing profiling build up their own to suit the type of profiling and probes they use. For example, a research group using a probe which picks up certain bands or sequences need to know about the frequency of those sequences, not others, in the population.

It is crucial to grasp the concept of what is meant by a 'chance match'. This is the probability of finding a particular profile by chance in the population. So if a probability of 1 in 1 million is stated for a particular profile presented at a trial, one could expect, in a city of 5 million people, to find 5 individuals who would match that by chance. It is not the chance that the defendant is guilty, nor innocent³¹. The chance match probability will probably be higher than 1 in 1 million if four single locus probes have been used on a good sample.

In its explanation of profiling the FSS states³²

'If, for example, a match is found between a sample and good quality crime sample with four independent probes, then we might hope to show that the possibility of a person, not involved with the crime, having a matching profile by chance is about 1 in 100 million. Whether it is beyond reasonable doubt that the crime and suspect samples must have come from the same person will depend on the circumstances of the case.

'Such tiny probabilities [1 in 100 million] imply evidential strengths far greater than have been achieved previously in the mainstream of forensic science. However, there are circumstances where the evidential implication of a particular case may not be so strong. The most obvious of these would be the quality and quantity of crime stain. If either of these is inadequate then four probes cannot be used because there is simply not enough DNA. If a result is achieved from only a single probe then we may be able to quote probabilities (of a chance match) of the order of 1 in 50 or even larger.'

³⁰source: FSS 26.3.96

³¹*New Statesman and Society* 8 December 1995 'Whose DNA is it anyway?' pp20-21

³²*What is DNA profiling?* Forensic Science Service 1993 p.6

B. The prosecutor's fallacy

One possible pitfall of presenting such evidence to jurors is known as the 'prosecutor's fallacy' and involves confusing the probability of a chance match with the probability of innocence. Note that the FSS above is careful to state that the possibility relates to an innocent individual matching the sample by chance. This is emphatically not the same as saying that the individual (suspect) is guilty or innocent, given that there is a match.

In 1987 someone called Andrew Deen was convicted of three rapes on the basis of DNA evidence which had used multi-locus probes³³ (see section I.D) but the Court of Appeal later quashed the conviction on a technicality concerning the presentation of the DNA evidence. The forensic scientist who gave evidence was reported as having stated the match probability in terms of 'the probability of the semen having originated from someone other than Andrew Deen', and as later going on to agree that 'the likelihood of [the source of the semen] being any other man but Andrew Deen is one in three million'. Both reported statements were incorrect; the match chance is simply that of a person chosen randomly from the population having a match to the crime sample. During the summing up the Judge had also made remarks about the quality of the DNA match, saying that a one in three million figure 'approximates pretty well to certainty'.^{34,35}

This was the first UK case in which DNA evidence was challenged. The use of a multi-locus probe which had scored 10 matching bands had resulted in a chance match probability of 1 in 700,000 being calculated by some other forensic scientists involved; already small compared to the sequential single locus probes more commonly in use today. Moreover, one German scientist had stated at the Appeal that he thought there were only 6 matching bands, four neutral and two discrepancies; he thought the DNA evidence showed a match probability of only 1 in 33³⁶. It must be stressed that more accurate and easily interpreted single locus probes are used in succession or simultaneously today and so the methods of calculating chance match probabilities for multi-locus probes will not be dealt with further here. (In 1995 the defence accepted the DNA evidence and Deen entered a guilty plea³⁷.)

³³*The Lancet* 'Commentary: Beyond all reasonable DNA' vol 345 June 1995 p.1587

³⁴*BMJ* vol. 308 2 April 1994 pp.874-5 'DNA evidence may have been misleading to courts'

³⁵*New Scientist* 16 April 1994 'Improving the odds on justice?' pp.12-13

³⁶*Nature* vol 368 24 March 1994 'How convincing is DNA evidence?' pp.285-6

³⁷*The Lancet* vol 345 24 June 1995 p.1587

C. Need for other evidence

Two mathematicians at Queen Mary and Westfield College London have pointed out that even if a tiny match probability is stated, this does not in itself constitute proof of guilt especially if there is little or no other evidence in the case.

They envisage a hypothetical case in a large city where someone has been arrested who matches the crime sample. However, without DNA evidence the person has only a one in a half million chance of being guilty because perhaps 500,000 other individuals in the city might have been thought as likely to have committed that crime. Given a one in one million chance match probability they then calculate, using a particular probability theory³⁸, that when the DNA evidence is included the probability of innocence is one in three³⁹.

However, there are drawbacks to this. While agreeing that the probability of innocence depends upon the totality of evidence, geneticists have highlighted other shortcomings in the above scenario, calling it 'not helpful'. For instance, the figure of a one in a half a million chance of being guilty is an entirely arbitrary choice not relevant to real cases, where suspects are profiled precisely because they are suspects⁴⁰;

'Forensic scientists have better things to do with their time than to screen systematically, with DNA profiling, individuals who have only a one in half a million chance of being guilty of the crime that is being investigated. In the future it may be true that suspects emerge through the screening of databases containing very many individuals, whose profiles were not collected in connection with the crime being investigated. In these circumstances it may indeed be true that suspects have very low prior probabilities of guilt, and this should be incorporated in the standard way using [standard statistical methods]'.

Another correspondent pointed out that in not one of 124 cases in the USA where DNA had been admitted in evidence had DNA been the sole evidence implicating the suspect in the crime, and went on to mention that 'Rhetorical criticisms of DNA testing can only generate confusion among judges and jurors alike. Significant effects of population substructure or not [see next section], the rarity of any specific hypervariable multilocus DNA profile is a biological fact'⁴¹.

³⁸*New Scientist* 16 April 1994 'Improving the odds on justice?' pp.12-13

³⁹*Nature* vol 368 24 March 1994 'How convincing is DNA evidence?' pp.285-6

⁴⁰*Nature* Correspondence vol 369 2 June 1994 'DNA profiling on trial' p.351

⁴¹*ibid*

Problems might indeed arise however if the national database grows large enough to throw up occasional chance matches. According to a Lecturer and Reader in Haematology at the London Hospital⁴²

'The important issue is this. The use of a national STR [short tandem repeat polymorphism] database will inevitably give rise to chance matches between crime samples and individuals on that database. How, in the absence of other evidence, will this information be used by the police? That is a question of concern for us all'.

In the case of mass screenings⁴³ individuals may be tested whether or not they are suspects. In such cases a trade or profession, or a place of residence (such as a village) may be the only other evidence linking such persons to a crime.

D. Relatedness, reference populations and ethnicity

According to the FSS⁴⁴

'...whereas the probability of a chance match between the crime profile and a random member of the population may be 1 in 100 million (for example) the probability of a chance match with the defendant's brother may be of the order of 1 in 200'.

In one case a scientist quoted a match probability after applying three single locus probes as 1 in 49,000 for unrelated individuals but added that a relative would be more likely to have the same profile. The probability of a match from a particular brother would be about 1 in 16, and that particular defendant had five brothers. If there had been no other evidence the probability that the defendant was innocent was thus have been more than 1 in 5; 1 in 17 even if there had been only one brother⁴⁵.

Such possibilities can be relatively easily taken into account, being matters of simple intuition. However, perhaps the most hotly debated problems concern ethnic sub-groups of populations;

⁴²letter to the *Guardian*, 29 December 1994 p.19 'Police use of a DNA database'

⁴³see section III.C

⁴⁴op cit

⁴⁵*Nature* vol 368 24 March 1994 'How convincing is DNA evidence?' pp.285-6

this raises complex and contentious issues. Human populations may not be statistically uniform, and if there is a sub-population from which the suspect or crime sample has been taken, then the chances of the same bands occurring may be increased. This would of course affect chance match probabilities.

When match probabilities are calculated (as mentioned in section II.A above) the band is compared to a reference population to see how often it occurs in the population. It used then to be common practice to simply multiply the probabilities for each band together to give infinitesimally small overall probabilities of a match (the so-called 'product rule'⁴⁶) but it has been pointed out that some gene sites or bands may occur in conjunction with one another (may not be randomly spread in the population) and so cannot be treated as independent and multiplied together in this way. Today a range of more sophisticated modelling methods is used⁴⁷.

It is now accepted that rather than there being a completely even spread of genes in populations, some repeat lengths or genes are more frequent in some ethnic groups than in others⁴⁸. While this does not (at least yet) open the possibility of identifying ethnic origin from a DNA sample (the sequences used are not found exclusively in one racial group and there are no sharp divisions between groups; the gene regions vary widely between individuals irrespective of race⁴⁹), it does mean that one should try to use a reference population database of the same ethnic group if possible. This may not always be available.

In 1991 two Harvard geneticists argued that the FBI's planned DNA database would be flawed in having only three ethnic groups as reference populations (Caucasians, Blacks and Hispanics) and by not taking into account ethnic subgroups. Their work had indicated that ethnic groups had multiple subpopulations in which certain genes might be more common than predicted theoretically; the subpopulations differed more than did the major ethnic groups themselves. They thus said that the reference groups would consist of hotpoches of subpopulations, which meant that assumptions being made about independence and homogeneity of genetic distribution could be flawed⁵⁰.

This might mean that if a murder was committed by an Amish then DNA from another innocent Amish would more closely resemble the crime scene sample than would DNA from the reference groups, and similar arguments could be made for other ethnic subgroups such

⁴⁶*Science* vol 259 5 February 1993 'Geneticists attack NRC report as scientifically flawed' pp755-6

⁴⁷*Nature* vol 371 27 October 1994 'DNA fingerprinting dispute laid to rest' pp735-8

⁴⁸*New Statesman and Society* 8 December 1995 'Whose DNA is it anyway?' pp20-21

⁴⁹*New Scientist* 8 July 1995 'Genes in black and white' pp34-37

⁵⁰*Science* vol 259 5 February 1993 'Statistical Evaluation of DNA Fingerprinting: A Critique of the NRC's Report' pp748-837

as Poles or Italians⁵¹. To put it another way, if DNA distribution is clumpy rather than randomly smooth, then small groups may exist which have increased chances of a false match⁵². The results concerning sub-population diversity found their way into a US National Research Council report on DNA fingerprinting in 1992, and were subsequently used by defence lawyers to argue a case for a lack of scientific consensus regarding population genetics and DNA analysis^{53,54}.

However, it has subsequently been argued that the above are, for a number of reasons, minority views. Other studies have investigated genetic diversity in subpopulations and have found, for instance, about twice the diversity between ethnic groups as exists within ethnic groups. In another study subpopulation diversity was much less than that among ethnic groups and both sources of diversity were far smaller than individual diversity⁵⁵. While the substructure argument has been debated with vigour in the scientific press, according to one review 'both sides conceded that substructure could matter in principle, but many doubted that its effect could be significant in practice'⁵⁶.

It is pointed out that in the US in particular, intermarriages have homogenised overall population structure (removed some clumpiness), while increasing genetic diversity among individuals (giving them a mix of inherited genes from parents from different ethnic backgrounds). This all adds up to a consensus that most variation derives from an individual level; which is what is needed for successful profiling⁵⁷.

E. Technical sources of error

According to one account, when DNA typing was first introduced it was in many cases offered by companies which were 'biotech start-up companies with good intentions but no track record in forensic science' which meant that DNA typing was 'marred by several early cases involving poorly defined procedures and interpretation'. For example, there were poorly defined procedures for defining a 'match', experiments without controls, contaminated probes and samples, and sloppy interpretation of fingerprints⁵⁸. Today criteria have been set for

⁵¹*Nature* vol 370 25 August 1994 pp588-9

⁵²*The Lancet* vol 345 24 June 1995 'Beyond all reasonable DNA' p.1587

⁵³*Science* vol 259 5 February 1993 'Statistical Evaluation of DNA Fingerprinting: A Critique of the NRC's Report' pp748-837

⁵⁴*Nature* vol 371 27 October 1994 'DNA fingerprinting dispute laid to rest' pp735-8

⁵⁵*Science* vol 259 5 February 1993 'Statistical Evaluation of DNA Fingerprinting: A Critique of the NRC's Report' pp748-837

⁵⁶*Nature* vol 371 27 October 1994 'DNA fingerprinting dispute laid to rest' pp735-8

⁵⁷*Science* vol 259 5 February 1993 'Statistical Evaluation of DNA Fingerprinting: A Critique of the NRC's Report' pp748-837

⁵⁸*Nature* vol 371 27 October 1994 'DNA fingerprinting dispute laid to rest'

determining whether a match exists between bands and, with more exacting laboratory procedures, bands match more clearly in any case. Clearer single locus probes applied together or in succession are also easier to interpret than multi-locus probes.

According to one of the Harvard researchers who criticised the FBI's use of only three reference populations⁵⁹, 'The problem of laboratory reliability has been greatly exacerbated by the increasing use of PCR technology to amplify small samples ... as everyone who uses the PCR knows, the probability of a false match by contamination of the minuscule crime scene sample, to be amplified, from the large sample taken from the accused, by mislabelling, aerosols, carelessly unchanged pipette tips and other similar laboratory sloppiness, is very great and many orders of magnitude greater than the tiny match probabilities calculated for forensic purposes'.

Another classic criticism is the possibility of a piece of dandruff from the lab technician falling into a sample. Yet while these possible problems need to be acknowledged, many arose from early teething problems or inexperience in conveying probabilities to juries, and it would be fair to say that a good deal of faith can now be placed in DNA profiling as evidence. Alec Jeffreys, who first developed fingerprinting, remains adamant that DNA profiling is reliable and almost all scientists would agree. One of the QMC mathematicians who has criticised the sole use of DNA evidence has said that his criticisms of the technique have been exaggerated, and that even if statistical methods were altered to take his points into account, match probabilities might fall from 10 million to one to one million to one⁶⁰. With four single locus probes producing probabilities of 100 million to one and the possibility of applying five such probes, this would probably make little difference so far as lawyers were concerned in most cases.

It must also be remembered that while the above arguments have focused on the fears of convicting an innocent individual, the very great advantage of DNA profiling is the ability to establish innocence or free someone from a paternity claim.

⁵⁹see above; *Nature* vol 372 1 December 1994 Correspondence 'Forensic DNA typing dispute' p.398

⁶⁰*New Scientist* 23 January 1993 'Doubts over DNA evidence 'exaggerated" p.6

III. Some applications

A. Paternity testing

Section I.A described the way in which our individual genetic makeups, encoded in DNA, are inherited half from our mothers and half from our fathers. This means that DNA profiling can therefore be very useful in determining fatherhood in cases of disputed paternity (for immigration and civil litigation cases), or in cases of concealed birth or abortion or childbirth following rape or an incestuous relationship⁶¹.

The great advantage of DNA profiling over conventional blood tests is that profiling can prove whether a man is a child's father, whereas other tests can only exclude various men and may be inconclusive. A DNA profile is created for the child, mother, and putative father(s). By comparing the profiles of the mother and child the bands inherited from the mother can be identified very simply by matching up their positions. With these maternal bands now eliminated, the rest of the bands must have been inherited from the father. If the alleged father's profile contains all these paternal-specific bands, paternity is confirmed. Alternatively, if the child's profile contains bands not present in the alleged father's profile, the child must have been fathered by someone else (Figure 3, next page).

The first paternity cases used complementary multilocus probes such as those developed by Jeffreys to give a complex pattern of bands. Today four or five single locus probes are usually applied in sequence to give a profile as described in section I.D, although a more complex fingerprint arising from the application of multi-locus probes may be used to analyse more complex relationship issues. An example was shown in Figure 2. Because one band in the child's profile will have come from the mother, this leaves only one band per probe which can be compared to the father, so profiling for paternity with a given number of bands is not as powerful a tool as for unrelated crime samples⁶².

The Child Support Agency introduced a discounted DNA scheme in July 1995, to provide alleged absent parents with a means of resolving paternity disputes without having to go to court. According to a recent PQ⁶³, from July 1995 to 25 January 1996 500 cases had been subject to tests; the alleged absent parent tested was proved to be the father in 433 (87%) of these cases. In other words, in 13% of cases the man was shown not to be the father.

⁶¹*What is DNA Profiling?* Forensic Science Service undated p.6

⁶²*ibid*

⁶³HC Deb 28 February 1996 cc623-4w

DNA testing has also been used in Argentina in the case of children separated from their parents by the military dictatorship of the 1970s. In some such cases the children's natural parents were political detainees who were murdered in detention and their babies given to illegal foster parents including military couples⁶⁴.

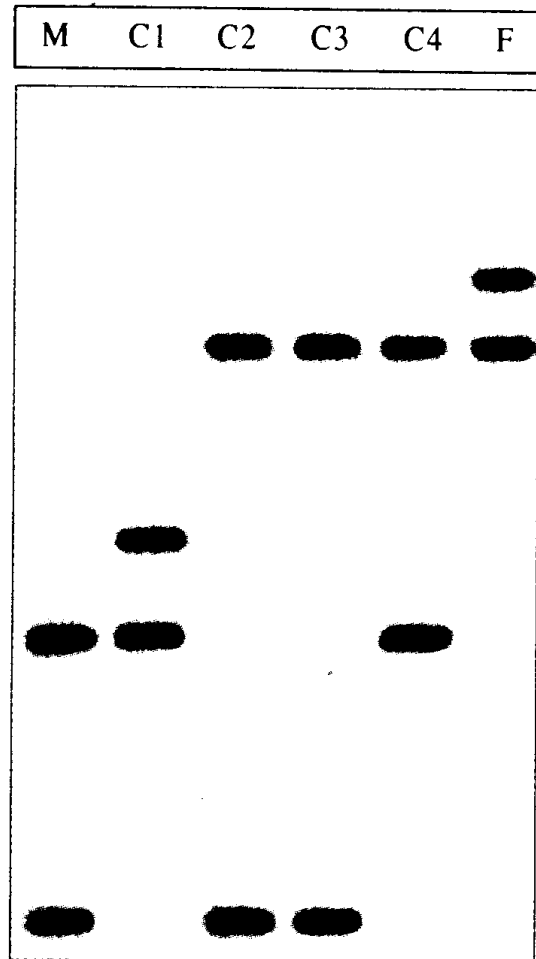


Figure 3⁶⁵ *Single locus probe result. A family with four children. M is the mother. F is the father of C2, C3 and C4, but not C1.*

B. National DNA Database

In February 1994 during the passage of the *Criminal Justice and Public Order Bill 1993/94*, Michael Howard announced a pilot study on the running of a national DNA database⁶⁶.

Section 57 of the *Criminal Justice and Public Order Act 1994* extended the provisions of

⁶⁴*Sunday Times* 25 June 1995

⁶⁵Reproduced with kind permission from *DNA Paternity Testing*, Cellmark Diagnostics Zeneca 1995

⁶⁶Home Office news release 24/94 3 February 1994 'DNA database- cracking crime through new technology'

section 64 of the *Police and Criminal Evidence Act 1984* regarding the retention of samples and fingerprints taken in evidence, and created an exemption in certain circumstances to the requirement that such samples be destroyed following an acquittal or the abandonment or discontinuance of a case.

Under the 1984 Act (with the authorisation of a police superintendent or above) non-intimate samples could be taken without consent from a suspect only if there were reasonable grounds for believing that the sample might confirm or disprove a suspect's involvement in a serious arrestable offence. An intimate sample could be taken if the suspect gave his written consent. DNA could be taken only in connection with serious arrestable offences such as murder, manslaughter, rape, certain terrorist offences, and serious thefts and only where it would tend to confirm or disprove the suspect's involvement in the offence.

The 1994 Act empowered the police to take DNA samples⁶⁷ from anyone charged with a recordable offence whether or not the DNA was immediately relevant to the particular offence under which the person was charged⁶⁸. Recordable offences are those for which convictions may be recorded in national police records; usually offences which carry a sentence of imprisonment on conviction. The director of Liberty (formerly the National Council for Civil Liberties) has noted that they include 'shoplifting and not paying your fare on the train'⁶⁹.

The reasoning behind this is that violent and sexual offenders often have previous convictions for more minor offences; the 1994 Act implemented the five recommendations of the Royal Commission on Criminal Justice on this subject in 1993⁷⁰. It has been pointed out that profiles would only be retained in searchable form if the suspect was convicted or cautioned for a recordable offence or if action against the individual was ongoing. In April 1995 the computerised national DNA database, run by the Forensic Science Service (FSS) at Birmingham, became operational⁷¹.

In May 1995 the new powers were used to take DNA samples from 911 people charged or cautioned after arrest (many for offences related to burglary) following 1,531 early morning searches. This prompted the legal officer for Liberty to comment⁷²:

⁶⁷Non-intimate samples including hair roots, mouth swabs and saliva without consent

⁶⁸Home Office Press Notice 24/94 3 February 1994 'DNA database - cracking crime through new technology'

⁶⁹*Independent* 28 February 1996 'Does DNA hold all the clues?...'

⁷⁰*Police Review* 28 April 1995 p.15 'Taking samples Code of Practice'

⁷¹Forensic Science Service News Release FSS 1/95 10 April 1995 'DNA database goes live'

⁷²*Guardian* 3 May 1995 'DNA samples taken after dawn raids'

'There is an independent check on police invading a suspect's home. They need a search warrant. There is no similar check when they want to invade a suspect's mouth for a saliva sample'.

It is estimated that 675,000 offenders are dealt with for recordable offences each year, so a substantial number of people may end up on the database⁷³. Although many see no ethical problems because in effect the database would differ little from conventional fingerprint or mugshot portfolios, Liberty opposed a national database for offenders which would be 'open to abuse', although it accepted the idea of one limited to sex offenders and murderers⁷⁴. Questioning the cost and reliability of DNA evidence, an editorial in the *Independent* on the provisions of the Criminal Justice and Public Order Act pointed to an 'alarming' 'potential invasion of privacy'. According to the *Independent* the 'most worrying aspect of the enterprise' was the prospect of 'intimate personal information about heredity and susceptibility to disease' being stored and susceptible to leakage⁷⁵.

The first match between the database and a sample taken from the scene of a crime was made in August 1995⁷⁶. By December 1995 118 matches had been made between DNA samples on the database, which held over 19,000 samples; the Home Office envisaged that the database could 'eventually hold the DNA profiles of every convicted criminal'⁷⁷. Over 35,000 samples have now been received to put on the database, and announcing the FSS's last annual report the Home Secretary Michael Howard said⁷⁸:

'The FSS is the indisputable world leader in forensic science. It has the world's first DNA database, and represents the cutting edge in scientific development.

'This pioneering system will revolutionise the way in which persistent offenders are recorded, and the message to the criminal is simple. DNA can catch you'.

The Minister of State for the Home Office Mr Maclean recently said that the database was having some 'fantastic successes' which would become public once sub judice restrictions had been lifted⁷⁹. The cost of launching the DNA database and of its first year of operation, 1995-96 has been forecast at £3.2 million⁸⁰.

⁷³*Police Review* 28 April 1995 pp.15-16 'Taking samples. Codes of Practice'

⁷⁴*Nature* 25 August 1994 p.588 'UK to set up DNA database of criminals...'

⁷⁵*Independent* 20 August 1994 'Beware the abuse of intimate gene data'

⁷⁶Home Office news release 175/95 11 August 1995 'The first match on the DNA database'

⁷⁷Home Office press notice 7 December 1995 283/95 'Over one hundred matches on the dna database'

⁷⁸Home Office Press Release 158/95 18 July 1995 'A world leader - the Forensic Science Service'

⁷⁹HC Deb 15 February 1996 cc1123-4

⁸⁰HC Deb 23 January 1996 c121w

The popularity of the database with police forces has been such that a backlog of 60,000 samples and a six month wait for results from the National Database Centre have built up⁸¹. It was recently announced that the FSS is to be merged with the Metropolitan Police Forensic Science Laboratory on 1 April 1996 to create a new national Forensic Science Service⁸² and although this will have its headquarters in Birmingham it is also anticipated that a second DNA unit in London will help clear the database backlog, which has now grown to 74,500 samples⁸³.

All police forces in England and Wales presently have access to the national database and in December 1995 the eight Scottish police forces decided to link to the scheme⁸⁴. Provisions of the *Criminal Justice (Scotland) Act 1995* which will come into force at the end of March 1996 will provide for the establishment of a DNA database for Scotland. Separate legislation will also allow for a database for Northern Ireland⁸⁵. The Scottish database will be held by the FSS at Birmingham alongside that for England and Wales and cross-searching will be allowed between the two⁸⁶.

It is thought that the database may hold four million profiles in five years' time⁸⁷.

C. Mass screening

One of the first successes for DNA profiling involved the separate murders of two 15 year old girls in Leicestershire. The male populations of two villages volunteered to take part in a mass blood test; the incidents were later rather luridly recounted in the book *The Blooding*⁸⁸. Over 5,000 males were tested; one man who had confessed to the killings was proved to be innocent, and another man, Colin Pitchfork, who had refused to take the test (and persuaded a friend to give blood in his place) was finally arrested and sentenced to life imprisonment for murdering the two girls. This case occurred at a time when the technique was still in its infancy and it was not coincidental that it took place near Leicester University, where Alec Jeffreys' research took place. Today mouth-swabs are used rather than blood tests, being a

⁸¹*Times* 26 February 1996 'DNA hold ups'

⁸²Home Office press notice 29 February 1996 'New forensic science service to help the police fight crime'

⁸³*Guardian* 1 March 1996 'Police get new scientific aid'

⁸⁴*Scotsman* 5 December 1995 'Police in DNA link-up'; *Police Review* 8 December 1995 p.13 'Scottish forces prepare to join the DNA database'

⁸⁵HC Deb 18 December 1995 c961w

⁸⁶Scottish Grand Committee Law and Order Monday 15 January 1996 c18

⁸⁷*New Scientist* 19 November 1995 'Crime match'

⁸⁸Joseph Wainburgh 1989

cheaper and quicker method.

A mass screening of 1250 people has been carried out by South Wales police in the search for the murderer of Claire Hood, and in October 1995 the Forensic Science Service was able to isolate a DNA profile of the murderer of 15 year old Naomi Smith who was sexually assaulted and killed in September. Launching a mass screening of over 800 males aged 15 to 28 in the area where Naomi lived, the Detective Superintendent in charge of the investigation said that⁸⁹

'..we have a genetic blueprint of someone involved in this murder. It is a question of when, and not if, we catch the offender'.

In November a 19 year old man was charged in connection with Naomi's murder. Most recently police have announced for the first time a national mass screening as part of the investigation of the murder of Celine Figard. In this case the population to be tested is not limited geographically but by profession, since there is reason to believe the murderer was a lorry driver. DNA taken from swabs from Celine's body will also be checked against the national DNA database.⁹⁰

Someone who refused to take such a test would clearly be placed under suspicion (although they would legally be entitled to do this) so it can be argued that the reputation of DNA profiling as a technique is in such cases almost as important as any results obtained. Liberty has alleged that in mass screenings there could be a danger of sample contamination or of a false match, leading to the conviction of an innocent man⁹¹.

Regarding retention of samples and profiles, David Maclean recently gave the following answer to Alan Beith⁹²;

'When members of the public volunteer to give samples as part of a mass screening, the samples taken are compared only with the sample found at the scene of the crime under investigation. The samples are destroyed at the end of the investigation and the profiles derived from them are not retained on the DNA database.

⁸⁹*Times* 24 October 1995 'Murder village on alert as 800 men face DNA check'

⁹⁰for example *Independent* 13 January 1996 'DNA test for 1,200 lorry drivers'

⁹¹*Guardian* 25 October 1995 'Warning on DNA murder tests'

⁹²HC Deb 31 October 1995 c166w

'When a sample is taken from a person suspected of involvement in a recordable offence, the police are required to inform the person that the sample may be the subject of a speculative search. Whether and for how long the samples will be retained is dependent upon the outcome of the investigation.

'Samples taken from persons convicted of or cautioned for a recordable offence will be retained for the same period as the offender's criminal record on Phoenix. Samples taken from people who are acquitted or not proceeded against will be retained if another person from whom a sample has been taken in the same investigation is convicted of an offence. These samples may be needed for further comparative analysis if it is subsequently suggested that there has been a miscarriage of justice.

'DNA profiles will be retained in a searchable form on the DNA database only if the suspect is convicted of or cautioned for a recordable offence or if action against that individual is ongoing'.

Names themselves are not held on the database but each record is labelled with a conventional barcode system which links with the Police computer holding the names. The barcode labels also preclude any chance of duplication or incorrect identification. Records are actually weeded off once they fail to fulfil any of the above criteria, rather than being for instance kept on in a non-searchable form⁹³.

D. Wildlife

Schedule 4 to the *Wildlife and Countryside Act 1981* lists bird species which must be registered and ringed if kept in captivity, and these in the past included most falcons and other birds of prey. The system was introduced because of major declines in populations of birds of prey in the wild; many eggs were being taken from the wild for use in falconry or in private collections and passed off as being captive bred.

Since the Act's introduction, populations of sparrowhawks, kestrels and common buzzards, in particular, have recovered in the wild, although one would not wish to be complacent. Although the RSPB had "urged the DoE to continue to require registration of these important

⁹³source: FSS 26.3.96

species"⁹⁴, in April 1994 following a DoE consultation exercise on the bird registration scheme Mr Atkins announced that the sparrowhawk, kestrel and common buzzard (all UK breeding species) and non-native and irregular visitor birds of prey would be removed from the Schedule. The idea is that by reducing "unnecessary regulation" the controls and resources of the DoE Wildlife Inspectorate can be targeted more effectively, for instance to protect more endangered species such as the hobby, red kite and golden eagle.

In addition, this will free resources for the DNA testing of captive birds to see whether these are really captive-bred as claimed or have instead been wild-caught; in effect this is paternity and maternity testing for birds. In the case of falcons, if both putative 'parents' are available for testing geneticists at Nottingham have estimated that the chances of a deception going undetected is less than 1 in 100 million⁹⁵.

In the first such case in Scotland but not in England and Wales, 22 police forces in association with the RSPB recently took blood samples from 131 birds (choughs, merlins and hobbies) from 30 sites across Great Britain. DNA profiling will be carried out at Nottingham University to see whether the birds were indeed bred in captivity or have been taken from the wild. The birds are all believed to have been sold by two people from Harlow, Essex; the couple have been arrested. The birds' owners (parks, zoos, falconry centres and private homes who purchased the birds through specialist magazines), are expected to be treated as victims of crime. The chough, a bird which is faced with extinction in Scotland, can fetch up to £500 per bird⁹⁶. More spectacular species such as the goshawk can cost up to £1000; there are perhaps only 300 breeding pairs left in Britain. The RSPB has said that⁹⁷

'DNA is such a powerful deterrent that people will realise that stealing birds is not worth the risk'.

This has been manifest in a 'sudden fall' in the number of birds claimed to have been bred in captivity. Following the seventh case of its kind, in which more than 20 out of 30 'captive bred' peregrines had been shown to be unrelated to the 'parents', the investigations officer for the RSPB said that the acceptance of the courts of DNA profiling had replaced the need to catch egg thieves in the act⁹⁸;

'Genetic fingerprinting is a very powerful forensic tool and the sudden decline in captive breeding figures [for peregrine falcons and goshawks] is because it

⁹⁴BIRDS Magazine Summer 1994

⁹⁵for more details see *Genetic Variation in Birds of Prey, Phase IV Final Report* 10 August 1994 by the University of Nottingham for the DoE

⁹⁶*Scotsman* 20 February 1996 'Police crackdown on rare bird dealers'

⁹⁷*Daily Telegraph* 5 April 1995 'Birds of prey to get DNA protection'

⁹⁸*Nature* vol 373 26 January 1995 p.275 'DNA fingers illegal trade in hawks'

is acting as a deterrent.'

The Act still protects birds of prey in the wild and prevents the injuring, killing or taking of wild birds. The RSPCA and others had expressed fears that freedom of movement within the EU would allow illegal trade in non-native birds, so some globally threatened species originally proposed for deregistration were allowed to remain on Schedule 4⁹⁹, and the birds left on Schedule 4 are all non-native species.

Around 1,000 chimpanzees are exported every year from Africa to Europe, the United States and Japan, despite the fact that the Convention on International Trade in Endangered Species (CITES) bans the capture and trade of all great apes in the 122 countries which are signatories to the Convention. It is said that zoos and circuses disguise the illegal trade by simulating live births and replacing adults with juvenile animals. DNA profiling in chimpanzees has now been developed by Italian researchers and used as evidence leading to a conviction for false veterinary certification of birth in captivity in Italy, and for reporting illegal exportation under CITES following DNA analysis on plucked hairs. The researchers hope that by building up a database of profiles of their captive animals they will eventually be able to eradicate the illegal introduction of specimens into the country¹⁰⁰.

DNA profiling techniques have long been applied to captive animals to maximise the success of breeding programmes. London Zoo has profiled species such as the Mauritius pink pigeon and Arabian oryx to determine whether potential partners come from the same or different populations¹⁰¹. This will help prevent inbreeding (mating two animals which are more closely related than would be desirable) and thus increase the fitness of the offspring and population that result. Of course such techniques may be used worldwide across zoos to design breeding programmes and maximise genetic variability.

On a more fundamental level, DNA analysis may be able to help distinguish between separate but outwardly very similar species. Modern systematic biology will, as well as considering traditional morphological and behavioural characters, be increasingly able to call on genetic techniques to help precisely split or join species, or even to determine relatedness or the evolutionary positions of different species¹⁰². This will help add some much needed depth to our understanding of the nature and extent of the biodiversity on Earth.

⁹⁹DoE *News Release* 271 25 April 1994

¹⁰⁰*Nature* vol 367 24 February 1994 'Chimpanzee DNA profiles on trial' pp692-3

¹⁰¹*Science for Conservation The research of the Zoological Society of London* June 1991 p.18

¹⁰²for instance see evidence given to the Lords Select Committee on Science and Technology, *Systematic Biology Research* Session 1991-92 1st report Volume II HL Paper 22-II

Further reading

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- *Genetic Variation in Birds of Prey, Phase IV Final Report* 10 August 1994 by the University of Nottingham for the DoE
- *DNA Paternity Testing* Cellmark Diagnostics Zeneca 1995 (pamphlet)
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Abbreviations/glossary

DNA	Deoxyribonucleic acid (the molecule that carries our genetic code)
Base pairs	The letters of the genetic code along the DNA molecule
Genome	The entire genetic makeup or complement carried in the cells of an organism
Gene	A sequence of DNA base pairs which codes for (writes the instructions for) a cell product
'Junk' DNA	DNA sequences which have no obvious function/do not seem to code for anything, and where variation and mutation rates can thus be higher than with gene sequences
VNTR	Variable number of tandem repeats (of certain sequences; a source of variation in the genome). 'Repeat classes' are termed minisatellites, microsatellites and satellites according to length and number of repeats
STR	Short tandem repeats
RFLP	Restriction fragment length polymorphism (another source of variation)
PCR	Polymerase chain reaction (a method of multiplying or amplifying up DNA samples in the test-tube)
Probe	A short length of DNA with a label attached, which will attach to its counterpart in the split DNA molecule, so identifying sequences of interest
FSS	Forensic Science Service
CITES	Convention on International Trade in Endangered Species