General Certificate of Education
Advanced Level Examination
June 2011

Science in Society SCIS4/PM

Unit 4

Preliminary Material

- This Source Material should be opened and issued to candidates on or after 1 May 2011.
- A clean copy of the Pre-released Source Material will be provided at the start of the Unit 4 examination.

Information

- This case study source material consists of extracts from five sources (**A-E**) on the subject of DNA and human rights.
- This material is being given to you in advance of the Unit 4 examination to enable you to study the content of each extract in preparation for questions based on the material in the examination. Consider the scientific explanations and the ideas about how science works that are involved, as well as the issues raised in the sources.
- You may write notes on this copy of the case study source material, but you will not be allowed to bring this copy, or any other notes you may have made, into the examination room. You will be provided with a clean copy of this case study source material, together with one additional source, **Source F**, at the start of the Unit 4 examination.
- You are not required to carry out any further study of the topic than is necessary for you to gain an understanding of the ideas described and to consider the issues raised. You are not required to understand any detailed **science explanations** beyond those outlined in **Sources A-E** and those in the *Science in Society* specification.
- It is suggested that a minimum of three hours detailed study is spent on this pre-release material.

Source A

Adapted from an article on www.thenakedscientists.com written by Dalya Rosner, a PhD student at Cambridge University, May 2004

How does DNA Fingerprinting Work?

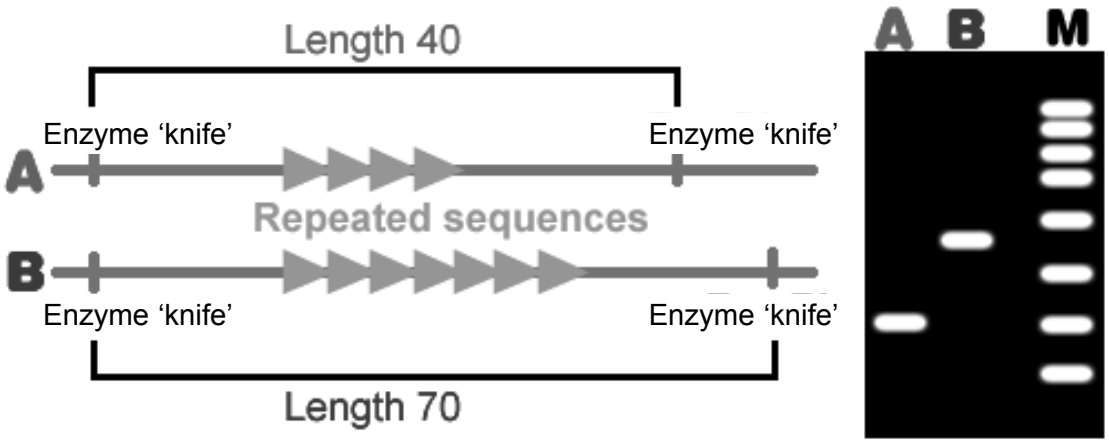
DNA fingerprinting is a technique for determining the likelihood that genetic material came from a particular individual or group. 99% of human DNA is identical between individuals, but the 1% that differs enables scientists to distinguish identity.

The DNA alphabet is made up of four building blocks – A, C, T and G, called bases, which are linked together in long chains to spell out the genetic words, or genes, which tell our cells what to do. The order in which these 4 DNA letters are used determines the meaning (function) of the words, or genes, that they spell.

But not all of our DNA contains useful information; in fact a large amount is said to be "non-coding" or "junk" DNA which is not translated into useful proteins. Changes often crop up within these regions of junk DNA because they make no contribution to the health or survival of the organism. But compare the situation if a change occurs within an essential gene, preventing it from working properly; the organism will be strongly disadvantaged and probably not survive, effectively removing that altered gene from the population.

For this reason, random variations crop up in the non-coding (junk) DNA sequences as often as once in every 200 DNA letters. One type of variation is known as a short tandem repeat (STR). These are short sections of DNA where a pattern of bases is repeated e.g. CATGCATGCATG. These STR are usually in non-coding DNA and don't make any difference to the organism. The place on the chromosome where the STR is found is known as the locus.

DNA fingerprinting takes advantage of the STR changes at different loci and creates a visible pattern of the differences to compare DNA from two or more organisms.

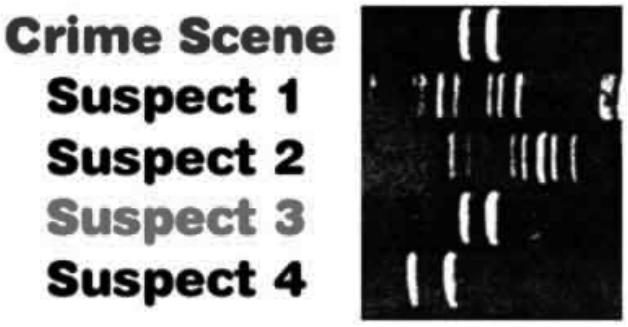


In junk regions of the genome, sequences of DNA are frequently repeated. In the example shown above, person A has only 4 repeats whilst person B has 7. When their DNA is cut at either end of the repeated sequence using enzymes, the DNA fragment produced by B is nearly twice as big as the piece from A, as shown in the picture. The lane marked M contains marker pieces of DNA that help determine the sizes. If lots of pieces of DNA are analysed in this way, a 'fingerprint' comprising DNA fragments of different sizes emerges.

Stretches of DNA can be separated from each other by cutting them up at these points of differences or by amplifying the highly variable pieces. 'Bands' of DNA are generated; the number of bands and their sizes give a unique profile of the DNA that it came from. The more genetic similarity between two people, the more similar the banding patterns will be, and the higher the probability that they are identical.

DNA fingerprinting is commonly used to probe our heredity. Since people inherit the arrangement of their base pairs from their parents, comparing the banding patterns of a child and the alleged parent generates a probability of relatedness; if the two patterns are similar enough (taking into account that only half the DNA is inherited from each parent), then they are probably family. However, DNA fingerprinting cannot discriminate between identical twins since their banding patterns are the same.

Perhaps best known is the use of DNA fingerprinting in forensic science. DNA samples gathered at a crime scene can be compared with the DNA of a suspect to show whether or not he or she was present.



In the example shown on the left, DNA collected at the scene of a crime is compared with DNA samples collected from 4 possible suspects. The DNA has been cut up into shorter pieces by enzymes. The pieces of DNA can be separated according to their size using a technique called electrophonesis. Each piece of DNA forms a band (the white lines). The lines from the DNA of suspect 3 match those left at the scene of the crime, betraying the guilty party.

Source B

Extracted from a speech given by the Home Secretary to the Intellect Trade Association, 16 December 2008

Home Secretary's speech: Protecting rights, protecting society

Today I'd like to address one of the most pressing questions we face as a modern society – how we secure our rights and liberties as individuals, at the same time as ensuring the wider protection of all in our society against terrorism, crime and disorder.

Balancing these individual and collective rights has always been a key responsibility of government. And in an era of rapid technological change, it is right that we should constantly satisfy ourselves that we have got the balance right.

When we talk about fingerprints…CCTV cameras…DNA swabs…or scanning machines at airports…I think that people instinctively understand that these technologies, used properly, are vital tools against crime, terrorism and illegal immigration.

But I also recognise the absolute necessity of getting the balance on privacy right.

… I've found there are few areas where the balance between rights and protections comes into such stark relief as on DNA.

In May 2002, Kensley Larrier was arrested for the possession of an offensive weapon. His DNA was taken and loaded to the DNA database, although the proceedings were then discontinued. Two years later, DNA from a rape investigation was speculatively searched against the database and matched his sample. This was the only evidence in the case, and when found guilty Larrier received a 5 year custodial sentence and was entered on the sex offenders register for life.

[This case] and others tell me that the DNA database is crucial to public protection. It not only helps to lead to the guilty. It helps to prove innocence and to rule people out as suspects. There is more we can do to strengthen the dividing line between guilt and innocence. For those who have committed a serious offence, our retention policies need to be as tough as possible.

But for others, including children, I am convinced that we need to be more flexible in our approach.

The DNA of children under 10 – the age of criminal responsibility – should no longer be held on the database. There are around 70 such cases, and we will take immediate steps to take them off.

For those under the age of 18, I think we need to strike the right balance between protecting the public and being fair to the individual. There's a big difference between a 12 year old having their DNA taken for a minor misdemeanour and a 17 year old convicted of a violent offence, and next year I will set out in a White Paper on Forensics how we ensure that that difference is captured in the arrangements for DNA retention.

We will consult on bringing greater flexibility and fairness into the system by stepping down some individuals over time – a differentiated approach, possibly based on age, or on risk, or on the nature of the offences involved.

That may mean letting the 12 year old I mentioned come off the database once they reach adulthood. And it could mean limiting how long the profiles of those who have been arrested but not convicted of an offence could be retained.

We are also re-examining retention arrangements for samples. Physical samples of hair and saliva swabs that represent people's actual DNA are much more sensitive than the DNA profile that is kept on the database – which only uses a small part of non-coding DNA.

This was a key point flagged up when we set up the Ethics Group under the National DNA Database Strategy Board, and we will pursue improvements to the safeguards around the handling of samples.

These changes will see some people coming off the system. But, as I said, we need to strengthen the dividing lines between innocence and guilt – and so I want to do more to ensure we get the right people on to the system as well.

No matter when they were convicted, I want to see the most serious offenders on the database. That's why we are working with the police to increase the number of convicted offenders on it, starting with those now serving time in prison for rape and murder. And we will also look at whether we need to extend powers so that the police can take DNA samples for a longer period after conviction and from those convicted overseas when they return to the UK.

As I said at the beginning, the use of DNA in investigations is one of the breakthroughs for modern policing. And it's an area where I'm proud to say that Britain is leading the world.

The strengths of the DNA database can only be safeguarded if they enjoy the confidence and trust of the public – and so the changes we will set out in the White Paper will deliver a more proportionate, fair and common sense approach.

At a time when technology is moving more quickly than ever before, and in an age where the public has never been better informed and more rigorous in their scrutiny of authority, it is fitting that the age-old question of how we get the balance right between individual and collective protections should continue to be asked.

Over the next few months, I want to engage the public in a discussion based on the protections and security we all derive from getting this balance right.

The public are our best defence against crime and terrorism. But I know they will not thank us if the systems we design to protect them are too intrusive. And so I will continue to put safeguards and openness, a sense of proportion and above all common sense, at the heart of everything we do.

Source C

Extracted from a Press Release from the Equality and Human Rights Commission

Commission says Government DNA database proposals will still break the law

7 August 2009

In its response to the Government's consultation, 'Keeping the Right People on the DNA Database', the Equality and Human Rights Commission believes further changes are required to the way DNA profiles are stored and used by the state if it is to comply with the law.

Under the Government's proposals, even if someone has not been charged with committing a crime their DNA profile can be kept for up to 12 years, or indefinitely if they have been found guilty of any offence.

The Commission believes this proposal does not meet the European Court of Human Rights requirement for the UK Government to have clear, justifiable reasons for holding on to DNA data from people who have not been convicted of a crime. The Commission's response is based on advice from Michael Beloff QC.

…Recommendations to bring [the Government] in line with the Council of Europe's guidance on the use of DNA in the criminal justice system:

- DNA profiles must be destroyed once a final decision has been made in a case, with only a few exceptions. A person's DNA profile should only be kept for a limited period if they have been convicted of a serious crime and where destroying that information is likely to pose a risk to the public.
- There must be more of a balance between someone's right to privacy and the right of other people to be protected from a crime that might be committed.
- The rules should also differentiate more between children and adults. The Commission argues that it is not proportionate – and therefore unlawful – to keep the DNA profile of a 10-year-old child arrested for a minor offence for the same length of time as an adult.

The Commission also wants an independent adjudicator to be put in place to oversee the system. This would give innocent people a way of challenging the need to keep their DNA profile on file.

John Wadham, Group Director Legal at the Commission, said:

'We recognise that the DNA database is a vital tool in the fight against crime, but people have a right to have their privacy protected. The proposed changes to the national DNA database are a step in the right direction, but we think there is no reason why the police should be allowed to keep anyone's DNA profile indefinitely. There also needs to be better protection for innocent people.'

The Commission also calls into question the validity of the research used by the Government to support its proposals, noting that the evidence has been criticised by other experts. The response also drew attention to issues that the consultation paper failed to address, but which the Commission thinks are highly relevant:

- In Scotland DNA samples and profiles must generally be destroyed if the individual is not convicted or is granted an absolute discharge, unless it relates to a violent or sexual offence. The Government has not pointed to any evidence that this is having a detrimental effect on crime in Scotland.
- The proposals do not tackle the fact that there is a disproportionate number of black men, particularly young black men, on the database.
- There are disproportionate numbers of vulnerable people on the database, including children as young as ten and people with mental illnesses.
- DNA is being collected for a broad range of offences, even when it may not be relevant as evidence.

Notes to Editors

- The UK Government has to revise how it stores fingerprints, DNA samples and DNA profiles following a case at the European Court of Human Rights (the Court). In the case of S and Marper v United Kingdom, (Dec 2008) the Court found that the 'blanket and indiscriminate' nature of the powers of retention of fingerprints, DNA samples and profiles of persons suspected but not convicted of offences in England and Wales interferes with their right to respect for their private lives (Article 8). Such a retention regime is not proportionate and fails to strike a fair balance between the competing interests. The court emphasised the general principle that an interference with an individual's right to privacy will only be considered 'necessary in a democratic society' for a legitimate aim if it answers a 'pressing social need' and, in particular, if it is proportionate to the legitimate aim pursued and if the reasons adduced to justify it are 'relevant and sufficient'.

The Equality and Human Rights Commission

The Commission is a statutory body established under the Equality Act 2006, which took over the responsibilities of Commission for Racial Equality, Disability Rights Commission and Equal Opportunities Commission. It is the independent advocate for equality and human rights in Britain. It aims to reduce inequality, eliminate discrimination, strengthen good relations between people, and promote and protect human rights. The Commission enforces equality legislation on age, disability, gender, race, religion or belief, sexual orientation or transgender status, and encourages compliance with the Human Rights Act. It also gives advice and guidance to businesses, the voluntary and public sectors, and to individuals.

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Source D

Extracted from a letter sent to *Science*, Volume 326, 18 December 2009, pp1631–1632

Time for DNA Disclosure

1. The legislation that established the U.S. National DNA Index System (NDIS) in 1994 explicitly anticipated that database records would be available for purposes of research and quality control "if personally identifiable information is removed". However, the Federal Bureau of Investigation (FBI), which controls the database, has published no research derived from NDIS and has declined to disclose these records to academic scholars. The National Research Council recently noted that "methods developed in crime laboratories to aid in law enforcement" would benefit from the contributions of academic scientists. We believe the time has come for the FBI to release anonymized NDIS profiles to academic scientists for research that will benefit criminal justice.
2. Disclosure of NDIS profiles would allow independent scientists to evaluate some of the population genetic assumptions underlying DNA testing using a database large enough to allow more sensitive evaluation of population structure. The publicly available population databases used to date for statistical estimation of the frequency of DNA profiles are relatively small ($N \approx 1000$), consisting of convenience samples analyzed over a decade ago. In contrast, NDIS has grown to over 7 million complete 13-locus short tandem repeat (STR) genotypes. Analysis of these data would allow more powerful tests of independence within and between loci.
3. The large sample size also allows real-world tests of propositions that previously have been addressed only by simulation. For example, it would allow tests of the frequency with which three-person mixtures could produce profiles consistent with two contributors; kinship analysis could allow assessment of how match probabilities are affected by the number of close relatives in the database.… Access to the anonymized 13-locus genotypes would allow more powerful analyses of these important issues than was previously possible.
4. Analysis of NDIS can also yield valuable insights into the frequency and circumstances under which certain typing errors may occur. A review of a government database from Victoria, Australia, containing 15,021 9-locus STR profiles shows how important such a review can be for "quality control purposes". The study found an error rate of about 1 in 300 for the typing of reference samples, which raises concerns about missed opportunities to develop investigative leads.
5. The profiles in the Victoria, Australia, database have been widely circulated for years with no known harm occurring. The U.S. government regularly argues to courts that broad mandatory DNA collection statutes are not unconstitutional precisely because the 13 genetic loci are noncoding and thus have no power to reveal any sensitive information.
6. Open access to data is a fundamental tenet of science. The need for openness was reinforced by the recent National Research Council report, which called for greater involvement of the academic community in assessment, validation, and improvement of forensic science methods. Law enforcement should honor the norms of science and open the NDIS and other government DNA databases to independent scientific scrutiny. Doing so poses no meaningful risk and can only strengthen the quality of forensic DNA analysis.

This letter was signed by 41 authors from 39 institutions including universities in UK and USA, private companies and individuals in USA. Details removed.

References

The letter included 12 references which have been removed for clarity.

Source E

Extract from: *The Journal of Law, Medicine and Ethics*, Volume 34, Issue 2, pp 248-262, 2006

Family Ties: The Use of DNA Offender Databases to Catch Offenders' Kin

Henry T. Greely, Daniel P. Riordan, Nanibaa' A. Garrison, and Joanna L. Mountain

1. Just after midnight on March 21, 2003, a drunk stood on a footbridge over a motorway in a village in Surrey in southern England. After eight pints of beer, he was drunk enough to decide to drop a brick from the overpass into traffic to see if he could hit something; unfortunately, he was not so drunk that he missed. The brick crashed through the windshield on the driver's side of a truck. It hit the driver, Michael Little, in the chest, triggering a fatal heart attack. He stayed conscious long enough to pull the truck safely to the side of the road, thereby perhaps saving other motorists; then he died. The crime was widely publicized, as was the driver's role in preventing any further accidents.

2. The police had no suspects, but they did have a clue – the brick had on it a mixture of DNA from the victim and someone else, presumably the perpetrator. The police also had blood from a nearby car that had been broken into that evening. The DNA from that blood matched the DNA on the brick. The police analyzed the DNA and compared it to their British DNA database, but found no match. Interviews began in the village, and voluntary DNA samples were taken from over 350 young men in the area, but without success. A £25,000 reward for information was offered, but nothing useful appeared. The police were eager to solve this crime, but, after six months, they had no suspect.

3. So the British police decided to check the DNA database for less than perfect matches in the hope that the perpetrator had a relative in the database. They set the search to pull up any offender in their database who matched at least eleven of the twenty DNA markers used by the British system. At first, they found too many matches to investigate. But after restricting the search to young white males from Surrey and Hampshire, two counties near the crime scene that are home to about 2.6 million people, they found about twenty five partial matches, one of which matched on sixteen of the twenty DNA markers. The police interviewed the person with the closest match and discovered he had a twenty-year-old brother living near the village where the crime had occurred. The brother, Craig Harman, denied involvement, but did agree to give a DNA sample. His DNA matched the DNA isolated from the blood on the brick; when confronted with the DNA match, Harman confessed. In April 2004 he started serving a six year prison term for manslaughter.

4. Genes run in families. If you have information about one person's genome, you know something about the likely composition of the genomes of his or her biological relatives. This fact is now beginning to be used in criminal investigations. As of 2004, the British had used this method about twenty times, gaining valuable information about a quarter of the time. In the United States, this method has been used successfully at least once. Willard Brown was convicted of a rape-murder from twenty years earlier after crime scene DNA provided a partial match to the DNA profile of his brother, Anthony Brown, which was in the North Carolina database. The same DNA test exonerated a man who had spent eighteen years in prison for the crime.

…

5. The legal and policy implications of this kind of "family forensic DNA," which the British call "familial searching," have been discussed only rarely and briefly. This article provides both a fuller explanation of the science and technology behind such uses, and a different analysis of their policy implications. … We do not view this article as providing definitive answers to the issues it raises, but we hope it will help start an informed discussion that can lead to useful policies concerning this technique.

Forensic DNA and DNA Databases
The Science of Forensic DNA Identification

6. The average human adult is made up of about fifty to one hundred trillion human cells, almost all of which contain, in their nuclei, forty-

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six chromosomes made up of deoxyribonucleic acid (DNA) wrapped around a protein backbone. Those chromosomes – pairs numbered one through twenty-two, plus either two X chromosomes (in most women) or one X and one Y (in most men) – comprise each human's "genome," the total of all that person's genetic information, apart from a very small amount of DNA in the cells' mitochondria. Each human's genome is made up of two copies of a variant of the human genome, one from his mother, one from his father. Each of these parental genomes, consisting of one each of the twenty-two pairs of chromosomes, plus the X or Y chromosome, is made up of about 3.2 billion "base pairs" of DNA. Each base pair makes up one "rung" of the double helix that is the DNA molecule, and comprises either an adenine connected to a thymine, a cytosine connected to a guanine, a guanine connected to a cytosine, or a thymine connected to an adenine: A-T, C-G, G-C, or T-A. The order of the A's, C's, G's, and T's is the genetic "sequence."

7. About three percent of the genome contains DNA instructions for making ("coding for") a particular version of ribonucleic acid (RNA), which in turn either provides the instructions for the cell to make a particular protein or has an independent function of its own. About four percent of the genome is not known to code for RNA but seems, from the fact that very similar genetic sequences are found in many species, to perform some important, but thus far unknown, function. The three percent that codes for RNA and probably much of the four percent with some other function make up the 20 000 to 25 000 "genes" that humans have. Some of the remaining ninety-three percent of our genomes appears to play structural roles in the physical activity of the chromosome; most of it, thus far at least, seems to have no function.

8. Nested within the vast majority of our genomes that have no known function are many different stretches of sequence called "microsatellites," or "short tandem repeats" ("STRs"). These are stretches of DNA where the DNA replicating mechanism appears to "stutter," resulting in different numbers of copies of repeated sequences. One common set of STRs involves repeated sequences of four bases – for example, the sequence ACGT. Some people inherited from one parent a stretch of DNA with four repeats of ACGT; others inherited a stretch with six repeats, or one, or ten, or twenty-five.

9. Each stretch with a different number of repeats is a different "allele." The fact that these stretches of DNA have a different number of these repeats makes them useful as "markers." Because their location on the chromosomes is known, they "mark" the location of genes that are nearby; because any individual will often have inherited a different length STR from his mother and his father, they can "mark" which chromosome came from which parent. Each of these STRs is found at one spot on one particular chromosome, a location known as a "locus."

10. These repeats, as far as scientists know, have no function. They do not code for RNA, and they do not seem to be responsible for any difference in the structure or functioning of the people of who carry them. In other words, a person whose genome has two copies (one from each parent) of a marker, with twelve repeats, seems no different from a person with two copies of the marker with five repeats, or someone with one copy with seventeen repeats and another copy with three repeats. These STRs can be used for identification. In the United States, crime laboratories typically use a set of thirteen STRs, known as the "CODIS markers," named after the FBI's Combined DNA Information System. These STRs are spread over twelve chromosomes. Each individual has two copies of each of the thirteen STRs. On average, one of the CODIS markers has twelve different lengths, or alleles, found in significant numbers of the population, but the least variable CODIS marker has seven alleles and the most variable has twenty-three. One person might have two copies of the first marker that are four and eight repeats long, copies of the second that are eleven and twenty-three copies long, copies of the third that are three and ten copies long, and so on through all thirteen markers. That person – someone, possibly the perpetrator, who left DNA at a crime scene; someone who left DNA on some important evidence to a crime; or an unidentified person whose remains have been found – can thus be identified as thirteen pairs of numbers, one pair for each of the thirteen STRs. Those numbers constitute a "genotype" of the individual for those STRs (based on the alleles they have of those STRs).

11. The odds that an unrelated person shares the same set of thirteen pairs are normally infinitesimal – at most one in several hundred billion, compared with a total of 6.3 billion living humans. Two random Americans will share, on average, about two or three alleles. On the other hand, identical twins will share all thirteen pairs

– and first degree relatives (parent, sibling, or child) on average will share at least half. This much higher rate of sharing among relatives is the reason for this paper.

Forensic DNA Databases

12. Many nations and every American state have established forensic DNA databases. The United Kingdom has one of the oldest and largest ones. Since 1995, it has collected and analyzed DNA samples from all those convicted of felonies and, indeed, often from those arrested but not convicted. The British analyze the DNA … and put the analyzed genotype (the … numbers that represent the length of the STRs for each of the pairs) into a database.

13. Their database now contains genotypes for over 2.5 million Britons, about five percent of the United Kingdom's population, and, as most of the samples are from adult males, it contains this genetic information on about one-tenth of the country's men. Every jurisdiction in the United States has established its own "offender" database. States set out different requirements about who must provide DNA for these databases. …

…description of different databases removed…

The Scientific Basis of Family Forensic DNA

14. DNA runs in families. Two people who are closely related genetically are likely to share more alleles than two people who are not closely related. The patterns of these similarities depend, however, on the type of familial relationship.

…

15. Determining, however, whether a high match is the result of a genetic family relationship between the offender in the database and whoever left the crime scene sample is not simple. It depends both on the nature of the postulated relationship and on the rarity of the genotype (set of alleles) involved.

16. First degree relatives share, on average, about fifty percent of each other's DNA variants, including STR lengths, by descent (as a result of their very recent shared ancestry). These are genetic parents, siblings, and children. Second degree relatives – uncles or aunts and nephews or nieces, grandparents and grandchildren, half-brothers and half-sisters – share one quarter of their DNA variations by descent; third degree relatives (first cousins or great-grandparents and great-grandchildren, among others) share one-eighth. First degree relationships are most likely to be useful for this investigative technique, but all first degree relationships are not the same.

17. While two unrelated people usually share only a few CODIS alleles, a genetic parent, say a father, and his child *must* match at no fewer than thirteen alleles, and are most likely to match at fourteen, fifteen, or sixteen alleles. One of the child's two alleles at each of the thirteen CODIS markers came from the father; except for the unusual event of a mutation in one of those alleles in the sperm that was part of the child's conception, those thirteen alleles *must be* the same. In addition, by chance, the father may share with the child's mother some of the thirteen alleles that he did not pass on to the child. For example, if two alleles of an average CODIS marker in genetically unrelated people, such as the child's mother and father, are likely to be identical by chance fifteen percent of the time, then the child will likely get two alleles from his mother that match the father's genotype. That child would match the father at one of two alleles at eleven markers (where the matching allele came only from the father), and at both alleles at two markers, where one came from the father and the other one, which came from the mother, happened by chance to be the same as the father's second allele. The most likely number of alleles shared between parent and child will vary from population to population because the extent to which unrelated individuals share alleles is somewhat different in different populations.

18. Using the Caucasian population (for which good published data exists) as an example, a father and his genetic child will share, on average, 15.7 of the twenty-six CODIS alleles, whereas two completely unrelated Caucasian individuals will share on average 8.7 alleles. However, it is also important to recognize that not only are a parent and child likely to have more total matching alleles than two unrelated people, but also that the way these genetic matches will occur between a parent and child is highly characteristic of that specific relationship – namely, every marker will have at least one of the two alleles in common, and relatively few markers will have more than one allele in common. The unusual pattern of parent-child matches – that they *must* match at one allele at each marker – makes them a particularly useful kind of partial match. The FBI has published the frequency in the CODIS database of the different

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length variations for all thirteen of the markers it examines. This data allows one to calculate how likely it is that a genotype that matches a profile in the Offender Index at a certain number of alleles comes from a relative of the person in the Offender Index, versus an unrelated person.

19. On average, the chance that an unrelated person's genotype will match the genotype from crime scene DNA at thirteen or more of the twenty-six alleles, allowing for all possible ways of distributing the matches across markers, is around three percent. However, the chance that two unrelated people match at thirteen or more sites with every marker having at least one match (as will occur for parent-child pairs) is about one in two thousand. Although these odds are low, with genotypes from 2.75 million people in the CODIS Offender Index, there should be many spurious matches at this level in the database. For an average genotype, around 2 000 to 3 000 people in the Offender Index are likely to have one or more matching alleles at all thirteen markers.

20. For a rare genotype, the number will be much lower – perhaps none. In fact, if we consider the rarest possible genotype in the Caucasian population – corresponding to someone who has two copies of the least frequent allele at every single marker – then the chance of an unrelated individual randomly matching that genotype at least once at every marker is around one in ten trillion quadrillion. Therefore, in this best-case scenario, a partial match at this level is considerably less likely to be spurious than is the typical perfect genotype match (which is on average around one in ten quadrillion for the Caucasian population).

21. On the other hand, in an extreme worst-case scenario (corresponding to someone who has one copy of each of the two most frequent alleles at every single marker), the chance of randomly matching at least once at every marker is just under one percent. In this worst-case scenario, it is unlikely that a true relative could be reliably identified by familial searching since there would be so many spurious matches.

22. It is possible for two siblings to share anywhere from zero to all twenty-six markers, but on average they share around 16.7 alleles. Thirteen of the shared alleles are expected to occur, on average, due to common inheritance of the same alleles from their parents, whereas the additional matches can occur either when the two parents share some alleles with each other, or when either parent has two copies of the same allele. Again, not only the number of overall matching markers, but also the *pattern* of how the matches are distributed across the genetic markers are characteristic of a relationship between two siblings – a few markers are expected to have no shared alleles, most have one allele in common, and a fair number of markers have two shared alleles.

23. For the same Caucasian population, an average pair of siblings has about one marker with no common alleles, about seven with exactly one shared allele, and about five with two alleles in common. Although there is no simple pattern of partial genotype matching that can perfectly distinguish all sibling pairs from spurious partial matches, the difference in genotype matching patterns between siblings and unrelated individuals does provide considerable information that can be used to successfully identify pairs of siblings some of the time. If a false positive rate of one in two thousand were tolerated (comparable to the false positive rate at which one hundred percent of parent-child pairs can be detected), then about sixty percent of true sibling pairs could be reliably identified.

24. However, about twenty percent of true siblings could be detected at a level that would be expected to yield only one in 100 000 matches by random chance, such that only around twenty to thirty spurious matches would result from searching the Offender Index. Thus, for a substantial fraction of cases, partial genotype match patterns can be used to reliably identify pairs of siblings with false positive rates comparable to or even much lower than observed for parent-child pairs (with one hundred percent detection).

25. Usually, the partial match by itself will not be overwhelming evidence that the person who left the crime scene DNA *is* a relative of the person in the Offender Index who provided a partial match. It will usually be the case that such a partial match could be made to many of the world's 6.3 billion people who do not have a relative in the CODIS Offender Index. The partial match is only a lead – a relatively weak one for a common genotype though possibly a very strong one for a rare genotype.

26. How strong or weak the lead is can be estimated. One should be able to estimate how many people in the overall population, or perhaps in defined subpopulations, match any given genotype at thirteen specified sites.

Ultimately, though, the partial match would only need to function as a lead and not as evidence in court. If a suspect were identified as a result of the partial match, his DNA could then be taken and analyzed (voluntarily, through a search warrant, or after arrest) and compared to the crime scene DNA, leading to a conclusive match or non-match.

27. Algorithms could be created easily to look for both parent-child and sibling-sibling matches. The first are more distinctive, because at least one allele at each site must match, but, at least in the early years of a database's existence, the second are likely to be more useful. ….

Legal and Policy Implications of Family Forensic DNA

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Suspicion by Family Ties

28. Dan Krane, a DNA specialist at Wright State University in Dayton, Ohio, says familial searching "puts someone in jeopardy of investigation simply because his brother committed a crime…that's the sins of the father being visited on the son…[it is] contrary to the whole idea of our criminal justice system." The "family suspicion" aspect of family forensic DNA clearly troubles people. It may well be responsible for the fact that this emerging technique is already being hedged round with limitations.

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Conclusion

29. This paper has argued that family forensic DNA has substantial potential to extend the usefulness of DNA databases in generating investigational leads from crime scene DNA. Several plausible enhancements could make it even more useful. Using DNA from offenders to help catch their relatives is, at the least, unsettling.

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30. Our goal in this paper is to explore and bring to light these possibilities and implications of family forensic DNA, not to propose a general "solution" to the issues it raises. We believe that our society needs to discuss these issues broadly and reach an open and politically legitimate resolution.

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The article contained 53 references which have been removed for clarity.

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