

# DNA profiling

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**DNA profiling** (also called **DNA testing**, **DNA typing**, or **genetic fingerprinting**) is a technique employed by forensic scientists to assist in the identification of individuals by their respective DNA profiles. **DNA profiles** are encrypted sets of numbers that reflect a person's DNA makeup, which can also be used as the person's identifier. DNA profiling should not be confused with full genome sequencing.<sup>[1]</sup> It is used in, for example, parental testing and criminal investigation.

Although 99.9% of human DNA sequences are the same in every person, enough of the DNA is different to distinguish one individual from another, unless they are monozygotic twins.<sup>[2]</sup> DNA profiling uses repetitive ("repeat") sequences that are highly variable,<sup>[2]</sup> called variable number tandem repeats (VNTRs), particularly short tandem repeats (STRs). VNTR loci are very similar between closely related humans, but so variable that unrelated individuals are extremely unlikely to have the same VNTRs.

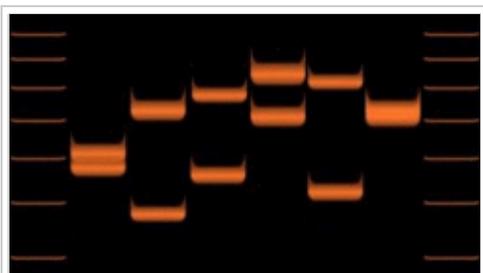
The DNA profiling technique was first reported in 1984<sup>[3]</sup> by Sir Alec Jeffreys at the University of Leicester in England,<sup>[4]</sup> and is now the basis of several national DNA databases. Dr. Jeffreys's genetic fingerprinting was made commercially available in 1987, when a chemical company, Imperial Chemical Industries (ICI), started a blood-testing centre in England.<sup>[5]</sup>

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## DNA profiling process



Variations of VNTR allele lengths in 6 individuals.

The process begins with a sample of an individual's DNA (typically called a "reference sample"). The most desirable method of collecting a reference sample is the use of a buccal swab, as this reduces the possibility of contamination. When this is not available (e.g. because a court order may be needed and not obtainable) other methods may need to be used to collect a sample



Alec Jeffreys, the pioneer of DNA profiling.

of blood, saliva, semen, or other appropriate fluid or tissue from personal items (e.g. toothbrush, razor, etc.) or from stored samples (e.g. banked sperm or biopsy tissue). Samples obtained from blood relatives (biological relative) can provide an indication of an individual's profile, as could human remains which had been previously profiled.

A reference sample is then analyzed to create the individual's DNA profile using one of a number of techniques, discussed below. The DNA profile is then compared against another sample to determine whether there is a genetic match.

### RFLP analysis

*Main article: Restriction fragment length polymorphism*

The first methods for finding out genetics used for DNA profiling involved restriction enzyme digestion, followed by Southern blot analysis. Although polymorphisms can exist in the restriction enzyme cleavage sites, more commonly the enzymes and DNA probes were used to analyze VNTR loci. However, the Southern blot technique is laborious, and requires large amounts of undegraded sample DNA. Also, Karl Brown's original technique looked at many minisatellite loci at the same time, increasing the observed variability, but making it hard to discern individual alleles (and thereby precluding parental testing). These early techniques have been supplanted by PCR-based assays.

### PCR analysis

*Main article: polymerase chain reaction*

In 1985 (see Mullis and Faloona 1987) a process was reported by which specific portions of the sample DNA can be amplified almost indefinitely (Saiki et al. 1985, 1988). This has revolutionized the whole field of DNA study. The process, the polymerase chain reaction (PCR), mimics the biological process of DNA replication, but confines it to specific DNA sequences of interest.

In this process, the DNA sample is denatured into the separate individual strands. Two DNA primers are used to hybridize to two corresponding nearby sites on opposite DNA strands in such a fashion that the normal enzymatic extension of the active terminal of each primer (that is, the 3' end) leads toward the other primer. In this fashion, two new copies of the sequence of interest are generated.

Repeated denaturation, hybridization, and extension in this fashion produce an exponentially growing number of copies of the DNA of interest. The denaturation is generally performed by heating, and in this case using, replication enzymes that are tolerant of high temperatures (Taq DNA polymerase). Instruments that perform thermal cycling are now readily available from commercial sources. This process can produce a million-fold or greater amplification of the desired region in 2 hours or less.

With the invention of the polymerase chain reaction (PCR) technique, DNA profiling took huge strides forward in both discriminating power and the ability to recover information from very small (or degraded) starting samples. PCR greatly amplifies the amounts of a specific region of DNA, using oligonucleotide primers and a thermostable DNA polymerase. Early assays such as the HLA-DQ alpha reverse dot blot strips grew to be very popular due to their ease of use, and the speed with which a result could be obtained. However they were not as discriminating as RFLP. It was also difficult to determine a DNA profile for mixed samples, such as a vaginal swab from a sexual assault victim.

Fortunately, the PCR method was readily adaptable for analyzing VNTR, particularly STR loci.

In recent years, research in human DNA quantitation has focused on new "real-time" quantitative PCR (qPCR) techniques. Quantitative PCR methods enable automated, precise, and high-throughput measurements. Interlaboratory studies have demonstrated the importance of human DNA quantitation on achieving reliable interpretation of STR typing and obtaining consistent results across laboratories.

## STR analysis

*Main article: Short tandem repeats*

The method of DNA profiling used today is based on PCR and uses short tandem repeats (STR). This method uses highly polymorphic regions that have short repeated sequences of DNA (the most common is 4 bases repeated, but there are other lengths in use, including 3 and 5 bases). Because unrelated people almost certainly have different numbers of repeat units, STRs can be used to discriminate between unrelated individuals. These STR loci (locations on a chromosome) are targeted with sequence-specific primers and amplified using PCR. The DNA fragments that result are then separated and detected using electrophoresis. There are two common methods of separation and detection, capillary electrophoresis (CE) and gel electrophoresis.

Each STR is polymorphic, but the number of alleles is very small. Typically each STR allele will be shared by around 5 - 20% of individuals. The power of STR analysis comes from looking at multiple STR loci simultaneously. The pattern of alleles can identify an individual quite accurately. Thus STR analysis provides an excellent identification tool. The more STR regions that are tested in an individual the more discriminating the test becomes.

From country to country, different STR-based DNA-profiling systems are in use. In North America, systems which amplify the CODIS 13 core loci are almost universal, while in the UK the SGM+ 11 loci system (which is compatible with The National DNA Database), is in use. Whichever system is used, many of the STR regions used are the same. These DNA-profiling systems are based on multiplex reactions, whereby many STR regions will be tested at the same time.

The true power of STR analysis is in its statistical power of discrimination. Because the 13 loci that are currently used for discrimination in CODIS are independently assorted (having a certain number of repeats at one locus

doesn't change the likelihood of having any number of repeats at any other locus), the product rule for probabilities can be applied. This means that if someone has the DNA type of ABC, where the three loci were independent, we can say that the probability of having that DNA type is the probability of having type A times the probability of having type B times the probability of having type C. This has resulted in the ability to generate match probabilities of 1 in a quintillion ( $1 \times 10^{18}$ ) or more. However, DNA database searches showed much more frequent than expected false DNA profile matches.<sup>[6]</sup> Moreover, since there are about 12 million monozygotic twins on Earth, the theoretical probability is not accurate.

In practice, the risk of contaminated-matching is much greater than matching a distant relative, such as a sample being contaminated from nearby objects, or from left-over cells transferred from a prior test. Logically, the risk is greater for matching the most common person in the samples: everything collected from, or in contact with, a victim is a major source of contamination for any other samples brought into a lab. For that reason, multiple control-samples are typically tested, to ensure that they stayed clean, when prepared during the same period as the actual test samples. Unexpected matches (or variations) in several control-samples indicates a high probability of contamination for the actual test samples. In a relationship test, the full DNA profiles should differ (except for twins), to prove that a person wasn't actually matched as being related to their own DNA in another sample.

## AmpFLP

*Main article: Amplified fragment length polymorphism*

Another technique, AmpFLP, or amplified fragment length polymorphism was also put into practice during the early 1990s. This technique was also faster than RFLP analysis and used PCR to amplify DNA samples. It relied on variable number tandem repeat (VNTR) polymorphisms to distinguish various alleles, which were separated on a polyacrylamide gel using an allelic ladder (as opposed to a molecular weight ladder). Bands could be visualized by silver staining the gel. One popular locus for fingerprinting was the D1S80 locus. As with all PCR based methods, highly degraded DNA or very small amounts of DNA may cause allelic dropout (causing a mistake in thinking a heterozygote is a homozygote) or other stochastic effects. In addition, because the analysis is done on a gel, very high number repeats may bunch together at the top of the gel, making it difficult to resolve. AmpFLP analysis can be highly automated, and allows for easy creation of phylogenetic trees based on comparing individual samples of DNA. Due to its relatively low cost and ease of set-up and operation, AmpFLP remains popular in lower income countries.

## DNA family relationship analysis

Using PCR technology, DNA analysis is widely applied to determine genetic family relationships such as paternity, maternity, siblingship and other kinships.

During conception, the father's sperm cell and the mother's egg cell, each containing half the amount of DNA found in other body cells, meet and fuse to form a fertilized egg, called a zygote. The zygote contains a complete set of DNA molecules, a unique combination of DNA from both parents. This zygote divides and multiplies into an embryo and later, a full human being.

At each stage of development, all the cells forming the body contain the same DNA—half from the father and half from the mother. This fact allows the relationship testing to use all types of all samples including loose cells from the cheeks collected using buccal swabs, blood or other types of samples.

While a lot of DNA contains information for a certain function, there is some called junk DNA, which is currently used for human identification. At some special locations (called loci) in the junk DNA, predictable inheritance patterns were found to be useful in determining biological relationships. These locations contain specific DNA markers that DNA scientists use to identify individuals. In a routine DNA paternity test, the markers used are

Short Tandem Repeats (STRs), short pieces of DNA that occur in highly differential repeat patterns among individuals.

Each person's DNA contains two copies of these markers—one copy inherited from the father and one from the mother. Within a population, the markers at each person's DNA location could differ in length and sometimes sequence, depending on the markers inherited from the parents.

The combination of marker sizes found in each person makes up his/her unique genetic profile. When determining the relationship between two individuals, their genetic profiles are compared to see if they share the same inheritance patterns at a statistically conclusive rate.

For example, the following sample report from this commercial DNA paternity testing laboratory Universal Genetics signifies how relatedness between parents and child is identified on those special markers:

<b>DNA Marker</b>	<b>Mother</b>	<b>Child</b>	<b>Alleged father</b>
D21S11	28, 30	28, 31	29, 31
D7S820	9, 10	10, 11	11, 12
TH01	14, 15	14, 16	15, 16
D13S317	7, 8	7, 9	8, 9
D19S433	14, 16.2	14, 15	15, 17

The partial results indicate that the child and the alleged father's DNA match among these five markers. The complete test results show this correlation on 16 markers between the child and the tested man to draw a conclusion of whether or not the man is the biological father.

Scientifically, each marker is assigned with a Paternity Index (PI), which is a statistical measure of how powerfully a match at a particular marker indicates paternity. The PI of each marker is multiplied with each other to generate the Combined Paternity Index (CPI), which indicates the overall probability of an individual being the biological father of the tested child relative to any random man from the entire population of the same race. The CPI is then converted into a Probability of Paternity showing the degree of relatedness between the alleged father and child.

The DNA test report in other family relationship tests, such as grandparentage and siblingship tests, is similar to a paternity test report. Instead of the Combined Paternity Index, a different value, such as a Siblingship Index, is reported.

The report shows the genetic profiles of each tested person. If there are markers shared among the tested individuals, the probability of biological relationship is calculated to determine how likely the tested individuals share the same markers due to a blood relationship.

## **Y-chromosome analysis**

Recent innovations have included the creation of primers targeting polymorphic regions on the Y-chromosome (Y-STR), which allows resolution of a mixed DNA sample from a male and female and/or cases in which a differential extraction is not possible. Y-chromosomes are paternally inherited, so Y-STR analysis can help in the identification of paternally related males. Y-STR analysis was performed in the Sally Hemings controversy to determine if Thomas Jefferson had sired a son with one of his slaves. The analysis of the Y-chromosome yields weaker results than autosomal chromosome analysis. The Y male sex-determining chromosome, as it is only inherited by males from their fathers, is almost identical along the patrilineal line. This leads to a less precise

analysis than if autosomal chromosomes were testing, because of the random matching that occurs between pairs of chromosomes as zygotes are being made.<sup>[7]</sup>

## Mitochondrial analysis

*Main article: Mitochondrial DNA*

For highly degraded samples, it is sometimes impossible to get a complete profile of the 13 CODIS STRs. In these situations, mitochondrial DNA (mtDNA) is sometimes typed due to there being many copies of mtDNA in a cell, while there may only be 1-2 copies of the nuclear DNA. Forensic scientists amplify the HV1 and HV2 regions of the mtDNA, then sequence each region and compare single-nucleotide differences to a reference. Because mtDNA is maternally inherited, directly linked maternal relatives can be used as match references, such as one's maternal grandmother's daughter's son. A difference of two or more nucleotides is generally considered to be an exclusion. Heteroplasmy and poly-C differences may throw off straight sequence comparisons, so some expertise on the part of the analyst is required. mtDNA is useful in determining clear identities, such as those of missing people when a maternally linked relative can be found. mtDNA testing was used in determining that Anna Anderson was not the Russian princess she had claimed to be, Anastasia Romanov.

mtDNA can be obtained from such material as hair shafts and old bones/teeth.

## DNA databases

*Main article: National DNA database*

There are now several DNA databases in existence around the world. Some are private, but most of the largest databases are government controlled. The United States maintains the largest DNA database, with the Combined DNA Index System, holding over 5 million records as of 2007.<sup>[8]</sup> The United Kingdom maintains the National DNA Database (NDNAD), which is of similar size, despite the UK's smaller population. The size of this database, and its rate of growth, is giving concern to civil liberties groups in the UK, where police have wide-ranging powers to take samples and retain them even in the event of acquittal.<sup>[9]</sup>

The U.S. Patriot Act of the United States provides a means for the U.S. government to get DNA samples from other countries if they are either a division of, or head office of, a company operating in the U.S. Under the act, the American offices of the company can't divulge to their subsidiaries/offices in other countries the reasons that these DNA samples are sought or by whom.<sup>[citation needed]</sup>

When a match is made from a National DNA Databank to link a crime scene to an offender who has provided a DNA Sample to a databank that link is often referred to as a *cold hit*. A cold hit is of value in referring the police agency to a specific suspect but is of less evidential value than a DNA match made from outside the DNA Databank.<sup>[10]</sup>

FBI agents cannot legally store DNA of a person who was not convicted of a crime. DNA collected from a suspect who was not later convicted must be disposed of and not entered into the database. In 1998 a man residing in the UK was arrested on accusation of burglary. His DNA was taken and tested, and he was later released. Nine months later, this man's DNA was accidentally and illegally entered in the DNA database. New DNA is automatically compared to the DNA found at cold cases, and in this case, this man was found to be a match to DNA found at a rape and assault case one year earlier. The government then prosecuted him for these crimes. During the trial the DNA match was requested to be removed from the evidence because it had been illegally entered into the database. The request was facilitated.<sup>[11]</sup>

## Considerations when evaluating DNA evidence

In the early days of the use of genetic fingerprinting as criminal evidence, juries were often swayed by spurious statistical arguments by defense lawyers along these lines: given a match that had a 1 in 5 million probability of occurring by chance, the lawyer would argue that this meant that in a country of say 60 million people there were 12 people who would also match the profile. This was then translated to a 1 in 12 chance of the suspect being the guilty one. This argument is not sound unless the suspect was drawn at random from the population of the country. In fact, a jury should consider how likely it is that an individual matching the genetic profile would also have been a suspect in the case for other reasons. Another spurious statistical argument is based on the false assumption that a 1 in 5 million probability of a match automatically translates into a 1 in 5 million probability of innocence and is known as the prosecutor's fallacy.

When using RFLP, the theoretical risk of a coincidental match is 1 in 100 billion (100,000,000,000), although the practical risk is actually 1 in 1000 because monozygotic twins are 0.2% of the human population.<sup>[*citation needed*]</sup> Moreover, the rate of laboratory error is almost certainly higher than this, and often actual laboratory procedures do not reflect the theory under which the coincidence probabilities were computed. For example, the coincidence probabilities may be calculated based on the probabilities that markers in two samples have bands in *precisely* the same location, but a laboratory worker may conclude that similar—but not precisely identical—band patterns result from identical genetic samples with some imperfection in the agarose gel. However, in this case, the laboratory worker increases the coincidence risk by expanding the criteria for declaring a match. Recent studies have quoted relatively high error rates which may be cause for concern.<sup>[12]</sup> In the early days of genetic fingerprinting, the necessary population data to accurately compute a match probability was sometimes unavailable. Between 1992 and 1996, arbitrary low ceilings were controversially put on match probabilities used in RFLP analysis rather than the higher theoretically computed ones.<sup>[13]</sup> Today, RFLP has become widely disused due to the advent of more discriminating, sensitive and easier technologies.

Since 1998, the DNA profiling system supported by The National DNA Database in the UK is the SGM+ DNA profiling system which includes 10 STR regions and a sex indicating test. STRs do not suffer from such subjectivity and provide similar power of discrimination (1 in 10<sup>13</sup> for unrelated individuals if using a full SGM+ profile). It should be noted that figures of this magnitude are not considered to be statistically supportable by scientists in the UK, for unrelated individuals with full matching DNA profiles a match probability of 1 in a billion is considered statistically supportable. However, with any DNA technique, the cautious juror should not convict on genetic fingerprint evidence alone if other factors raise doubt. Contamination with other evidence (secondary transfer) is a key source of incorrect DNA profiles and raising doubts as to whether a sample has been adulterated is a favorite defense technique. More rarely, chimerism is one such instance where the lack of a genetic match may unfairly exclude a suspect.

### Evidence of genetic relationship

It's also possible to use DNA profiling as evidence of genetic relationship, although such evidence varies in strength from weak to positive. Testing that shows no relationship is absolutely certain.

While almost all individuals have a single and distinct set of genes, ultra-rare individuals, known as "chimeras", have at least two different sets of genes. There have been two cases of DNA profiling that falsely suggested that a mother was unrelated to her children.<sup>[14]</sup> This happens when two eggs are fertilized at the same time and fuse together to create one individual instead of twins.

### Fake DNA evidence

The value of DNA evidence has to be seen in light of recent cases where criminals planted fake DNA samples at crime scenes. In one case, a criminal even planted fake DNA evidence in his own body: Dr. John Schneeberger raped one of his sedated patients in 1992 and left semen on her underwear. Police drew what they believed to be Schneeberger's blood and compared its DNA against the crime scene semen DNA on three occasions, never showing a match. It turned out that he had surgically inserted a Penrose drain into his arm and filled it with foreign blood and anticoagulants.

The functional analysis of genes and their coding sequences (open reading frames [ORFs]) typically requires that each ORF be expressed, the encoded protein purified, antibodies produced, phenotypes examined, intracellular localization determined, and interactions with other proteins sought.<sup>[15]</sup> In a study conducted by the life science company Nucleix and published in the journal *Forensic Science International*, scientists found that an *In vitro* synthesized sample of DNA matching any desired genetic profile can be constructed using standard molecular biology techniques without obtaining any actual tissue from that person.

In the case of the Phantom of Heilbronn, police detectives found DNA traces from the same woman on various crime scenes in Austria, Germany and France — among them murders, burglaries and robberies. Only after the DNA of the "woman" matched the DNA sampled from the burned body of a *male* asylum seeker in France, detectives began to have serious doubts about the DNA evidence. In that case, DNA traces were already present on the cotton swabs used to collect the samples at the crime scene, and the swabs had all been produced at the same factory in Austria. The company's product specification said that the swabs were guaranteed to be sterile, but not DNA-free.

## DNA evidence as evidence in criminal trials

### Familial DNA searching

Familial DNA searching (sometimes referred to as “Familial DNA” or “Familial DNA Database Searching”) is the practice of creating new investigative leads in cases where DNA evidence found at the scene of a crime (forensic profile) strongly resembles that of an existing DNA profile (offender profile) in a state DNA database but there is not an exact match.<sup>[16][17]</sup> After all other leads have been exhausted, investigators may use specially developed software to compare the forensic profile to all profiles taken from a state’s DNA database in order to generate a list of those offenders most likely to be a close relative of the forensic profile.<sup>[18]</sup> To eliminate the majority of this list, crime lab technicians conduct Y-STR analysis that confirms the familial relationships suggested by the first list. Using standard investigative techniques, authorities are then able to build a family tree. The family tree is populated from information gathered from public records and criminal justice records. Investigators rule out family members’ involvement in the crime by finding excluding factors such as sex, living out of state or being incarcerated when the crime was committed. They may also use other leads from the case, such as witness or victim statements, to identify a suspect. Once a suspect has been identified, investigators seek to legally obtain a DNA sample from the suspect. This suspect DNA profile is then compared to the sample found at the crime scene, in accordance with well established and constitutionally accepted practices, to definitively identify the suspect as the source of the crime scene DNA.

Familial DNA database searching was first used to convict Craig Harman of manslaughter in the United Kingdom on April 19, 2004.<sup>[19]</sup> Craig Harman was convicted using familial DNA because of the partial matches from Harman's brother. When the police questioned Harman's brother, the police noticed Harman lived very close to the original crime scene. Harman confessed when his DNA isolated from the DNA found on the brick, matched.<sup>[20]</sup> Currently, familial DNA database searching is not conducted on a national level in the United States. States determine their own policies and decision making processes for how and when to conduct familial searches. The first familial DNA search and subsequent conviction in the United States was conducted in Denver, Colorado in

2008 using software developed under the leadership of Denver District Attorney Mitch Morrissey and Denver Police Department Crime Lab Director Gregg LaBerge.<sup>[21]</sup> California was the first state to implement a policy for familial searching under then Attorney General, now Governor, Jerry Brown.<sup>[22]</sup> In his role as consultant to the Familial Search Working Group of the California Department of Justice, former Alameda County Prosecutor Rock Harmon is widely considered to have been the catalyst in the adoption of familial search technology in California. The technique was used to catch the Los Angeles serial killer known as the "Grim Sleeper" in 2010<sup>[23]</sup> It wasn't a witness or informant that tipped off law enforcement to the identity of the "Grim Sleeper" serial killer, who had eluded police for more than two decades, but DNA from the suspect's own son. The suspect's son was arrested and convicted in a felony weapons charge and swabbed for DNA last year. When his DNA was entered into the database of convicted felons, detectives were alerted to a partial match to evidence found at the "Grim Sleeper" crime scenes. David Franklin Jr., also known as the Grim Sleeper, was charged with ten counts of murder and one count of attempted murder.<sup>[24]</sup> More recently, familial DNA, led to the arrest of 21-year-old Elvis Garcia on charges of sexual assault and false imprisonment of a woman in Santa Cruz in 2008.<sup>[25]</sup> In March 2011 Virginia Governor Bob McDonnell announced that Virginia would begin using familial DNA searches.<sup>[26]</sup> Other states are expected to follow.

At a press conference in Virginia on March 7, 2011 regarding the East Coast Rapist, Prince William County prosecutor Paul Ebert and Fairfax County Police Detective John Kelly said the case would have been solved years ago if Virginia had used familial DNA searching. Aaron Thomas, the suspected East Coast Rapist, was arrested in connection with the rape of 17 women from Virginia to Rhode Island, but familial DNA was not used in the case.<sup>[27]</sup>

Critics of familial DNA database searches argue that the technique may be an invasion of an individual's 4th Amendment rights.<sup>[28]</sup> Privacy advocates are petitioning for DNA database restrictions, while others are arguing that the only fair way to construct of database is to make it universal.<sup>[11]</sup> Some scholars have pointed out that the privacy concerns surrounding familial searching are no more threatening than other police search techniques.<sup>[29]</sup> The Ninth Circuit Court of Appeals in *United States v. Pool* ruled that this practice is somewhat analogous to a witness looking at a photograph of one person and stating that it looked like the perpetrator, which leads law enforcement to show the witness photos of similar looking individuals, one of whom is identified as the perpetrator.<sup>[30]</sup> Regardless of whether familial DNA searching was the method used to identify the suspect, authorities always conduct a normal DNA test to match the suspect's DNA with that of the DNA left at the crime scene. Critics also claim that racial profiling could occur on account of Familial DNA testing. In the United States, the conviction rates of racial minorities are much higher than that of the overall population. It is unclear whether this is due to discrimination from police officers and the courts, as opposed to a simple higher rate of offence among minorities. Arrest-based databases, which are found in some parts of the United States, lead to an even greater level of racial discrimination. An arrest, as opposed to conviction, relies much heavier on police discretion, inevitably leading to a bias on the amount of people belonging to a certain race that are being convicted.<sup>[11]</sup>

For instance, investigators with Denver District Attorney's Office successfully identified a suspect in a property theft case using a familial DNA search. In this example, the suspect's blood left at the scene of the crime strongly resembled that of a current Colorado Department of Corrections prisoner.<sup>[31]</sup> Using publicly available records, the investigators created a family tree. They then eliminated all the family members who were incarcerated at the time of the offense, as well as all of the females (the crime scene DNA profile was that of a male). Investigators obtained a court order to collect the suspect's DNA, but the suspect actually volunteered to come to a police station and give a DNA sample. After providing the sample, the suspect walked free without further interrogation or detainment. Later confronted with an exact match to the forensic profile, the suspect pled guilty to criminal trespass at the first court date and was sentenced to two years probation.

## Partial matches

Partial DNA matches are not searches themselves, but are the result of moderate stringency CODIS searches that produce a potential match that shares at least one allele at every locus.<sup>[32]</sup> Partial matching does not involve the use of a familial search software, such as those used in the UK and Denver, or additional Y-STR analysis, and therefore often misses sibling relationships. Partial matching has been used to identify suspects in several cases in the UK and US<sup>[33]</sup> and has also been used as a tool to exonerate the falsely accused. Darryl Hunt was wrongly convicted in connection with the rape and murder of a young woman in 1984 in North Carolina.<sup>[34]</sup> Hunt was exonerated in 2004 when a DNA database search produced a remarkably close match between a convicted felon and the forensic profile from the case. The partial match led investigators to the felon's brother, Willard E. Brown, who confessed to the crime when confronted by police. A judge then signed an order to dismiss the case against Hunt.

## Surreptitious DNA collecting

Police forces may collect DNA samples without the suspects' knowledge, and use it as evidence. Legality of this mode of proceeding has been questioned in Australia.

In the United States, it has been accepted, courts often claiming that there was no expectation of privacy, citing *California v. Greenwood* (1985), during which the Supreme Court held that the Fourth Amendment does not prohibit the warrantless search and seizure of garbage left for collection outside the curtilage of a home. Critics of this practice underline the fact that this analogy ignores that "most people have no idea that they risk surrendering their genetic identity to the police by, for instance, failing to destroy a used coffee cup. Moreover, even if they do realize it, there is no way to avoid abandoning one's DNA in public."<sup>[35]</sup>

In the UK, the Human Tissue Act 2004 prohibited private individuals from covertly collecting biological samples (hair, fingernails, etc.) for DNA analysis, but excluded medical and criminal investigations from the offence.<sup>[36]</sup>

## England and Wales

Evidence from an expert who has compared DNA samples must be accompanied by evidence as to the sources of the samples and the procedures for obtaining the DNA profiles.<sup>[37]</sup> The judge must ensure that the jury must understand the significance of DNA matches and mismatches in the profiles. The judge must also ensure that the jury does not confuse the 'match probability' (the probability that a person that is chosen at random has a matching DNA profile to the sample from the scene) with the 'likelihood ratio' (the probability that a person with matching DNA committed the crime). In 1996 *R v. Doheny*<sup>[38]</sup> Phillips LJ gave this example of a summing up, which should be carefully tailored to the particular facts in each case:

Members of the Jury, if you accept the scientific evidence called by the Crown, this indicates that there are probably only four or five white males in the United Kingdom from whom that semen stain could have come. The Defendant is one of them. If that is the position, the decision you have to reach, on all the evidence, is whether you are sure that it was the Defendant who left that stain or whether it is possible that it was one of that other small group of men who share the same DNA characteristics.

Juries should weigh up conflicting and corroborative evidence, using their own common sense and not by using mathematical formulae, such as Bayes' theorem, so as to avoid "confusion, misunderstanding and misjudgment".<sup>[39]</sup>

## Presentation and evaluation of evidence of partial or incomplete DNA profiles

In *R v Bates*,<sup>[40]</sup> Moore-Bick LJ said:

“We can see no reason why partial profile DNA evidence should not be admissible provided that the jury are made aware of its inherent limitations and are given a sufficient explanation to enable them to evaluate it. There may be cases where the match probability in relation to all the samples tested is so great that the judge would consider its probative value to be minimal and decide to exclude the evidence in the exercise of his discretion, but this gives rise to no new question of principle and can be left for decision on a case by case basis. However, the fact that there exists in the case of all partial profile evidence the possibility that a "missing" allele might exculpate the accused altogether does not provide sufficient grounds for rejecting such evidence. In many there is a possibility (at least in theory) that evidence exists which would assist the accused and perhaps even exculpate him altogether, but that does not provide grounds for excluding relevant evidence that is available and otherwise admissible, though it does make it important to ensure that the jury are given sufficient information to enable them to evaluate that evidence properly”.<sup>[41]</sup>

## DNA testing in the US

There are state laws on DNA profiling in all 50 states of the United States.<sup>[42]</sup> Detailed information on database laws in each state can be found at the National Conference of State Legislatures website.<sup>[43]</sup>

## Development of artificial DNA

In August 2009, scientists in Israel raised serious doubts concerning the use of DNA by law enforcement as the ultimate method of identification. In a paper published in the journal *Forensic Science International: Genetics*, the Israeli researchers demonstrated that it is possible to manufacture DNA in a laboratory, thus falsifying DNA evidence. The scientists fabricated saliva and blood samples, which originally contained DNA from a person other than the supposed donor of the blood and saliva.<sup>[44]</sup>



CBP chemist reads a DNA profile to determine the origin of a commodity.

The researchers also showed that, using a DNA database, it is possible to take information from a profile and manufacture DNA to match it, and that this can be done without access to any actual DNA from the person whose DNA they are duplicating. The synthetic DNA oligos required for the procedure are common in molecular laboratories.<sup>[44]</sup>

*The New York Times* quoted the lead author on the paper, Dr. Daniel Frumkin, saying, "You can just engineer a crime scene... any biology undergraduate could perform this."<sup>[44]</sup>

Dr. Frumkin perfected a test that can differentiate real DNA samples from fake ones. His test detects epigenetic modifications, in particular, DNA methylation. Seventy percent of the DNA in any human genome is methylated, meaning it contains methyl group modifications within a CpG dinucleotide context. Methylation at the promoter region is associated with gene silencing. The synthetic DNA lacks this epigenetic modification, which allows the test to distinguish manufactured DNA from original, genuine, DNA.<sup>[44]</sup>

It is unknown how many police departments, if any, currently use the test. No police lab has publicly announced that it is using the new test to verify DNA results.<sup>[45]</sup>

## Cases

- In the 1950s, Anna Anderson claimed that she was Grand Duchess Anastasia Nikolaevna of Russia. In the 1980s, after her death, samples of her tissue that had been stored at a Charlottesville, Virginia hospital following a medical procedure were tested using DNA fingerprinting, and showed that she bore no relation to the Romanovs.<sup>[46]</sup>
- In 1986, Richard Buckland was exonerated, despite having admitted to the rape and murder of a teenager near Leicester, the city where DNA profiling was first discovered. This was the first use of DNA fingerprinting in a criminal investigation.<sup>[47]</sup>
  - In 1987, in the same case as Buckland, British baker Colin Pitchfork was the first criminal caught and convicted using DNA fingerprinting.<sup>[48]</sup>
- In 1987, genetic fingerprinting was used in criminal court for the first time in the trial of a man accused of unlawful intercourse with a mentally handicapped 14-year-old female who gave birth to a baby.<sup>[49]</sup>
- In 1987, Florida rapist Tommy Lee Andrews was the first person in the United States to be convicted as a result of DNA evidence, for raping a woman during a burglary; he was convicted on November 6, 1987, and sentenced to 22 years in prison.<sup>[50][51]</sup>
- In 1988, Timothy Wilson Spencer was the first man in Virginia to be sentenced to death through DNA testing, for several rape and murder charges. He was dubbed "The South Side Strangler" because he killed victims on the south side of Richmond, Virginia. He was later charged with rape and first-degree murder and was sentenced to death. He was executed on April 27, 1994. David Vasquez, initially convicted of one of Spencer's crimes, became the first man in America exonerated based on DNA evidence.
- In 1989, Chicago man Gary Dotson was the first person whose conviction was overturned using DNA evidence.
- In 1991, Allan Legere was the first Canadian to be convicted as a result of DNA evidence, for four murders he had committed while an escaped prisoner in 1989. During his trial, his defense argued that the relatively shallow gene pool of the region could lead to false positives.
- In 1992, DNA evidence was used to prove that Nazi doctor Josef Mengele was buried in Brazil under the name Wolfgang Gerhard.
- In 1992, DNA from a palo verde tree was used to convict Mark Alan Bogan of murder. DNA from seed pods of a tree at the crime scene was found to match that of seed pods found in Bogan's truck. This is the first instance of plant DNA admitted in a criminal case.<sup>[52][53][54]</sup>
- In 1993, Kirk Bloodsworth was the first person to have been convicted of murder and sentenced to death, whose conviction was overturned using DNA evidence.
- The 1993 rape and murder of Mia Zapata, lead singer for the Seattle punk band The Gits was unsolved nine years after the murder. A database search in 2001 failed, but the killer's DNA was collected when he was arrested in Florida for burglary and domestic abuse in 2002.
- The science was made famous in the United States in 1994 when prosecutors heavily relied on DNA evidence allegedly linking O. J. Simpson to a double murder. The case also brought to light the laboratory difficulties and handling procedure mishaps which can cause such evidence to be significantly doubted.
- In 1994, Royal Canadian Mounted Police (RCMP) detectives successfully tested hairs from a cat known as Snowball, and used the test to link a man to the murder of his wife, thus marking for the first time in forensic history the use of non-human DNA to identify a criminal.
- In 1995, the British Forensic Science Service carried out its first mass intelligence DNA screening in the investigation of the Naomi Smith murder case.
- In 1998, Dr. Richard J. Schmidt was convicted of attempted second-degree murder when it was shown that there was a link between the viral DNA of the human immunodeficiency virus (HIV) he had been accused of injecting in his girlfriend and viral DNA from one of his patients with AIDS. This was the first time viral DNA fingerprinting had been used as evidence in a criminal trial.
- In 1999, Raymond Easton, a disabled man from Swindon, England, was arrested and detained for seven hours in connection with a burglary. He was released due to an inaccurate DNA match. His DNA had been retained on file after an unrelated domestic incident some time previously.<sup>[55]</sup>

- In May 2000 Gordon Graham murdered Paul Gault at his home in Lisburn, Northern Ireland. Graham was convicted of the murder when his DNA was found on a sports bag left in the house as part of an elaborate ploy to suggest the murder occurred after a burglary had gone wrong. Graham was having an affair with the victim's wife at the time of the murder. It was the first time Low Copy Number DNA was used in Northern Ireland.<sup>[56]</sup>
- In 2001, Wayne Butler was convicted for the murder of Celia Douty. It was the first murder in Australia to be solved using DNA profiling.<sup>[57][58]</sup>
- In 2002, the body of James Hanratty, hanged in 1962 for the "A6 murder", was exhumed and DNA samples from the body and members of his family were analysed. The results convinced Court of Appeal judges that Hanratty's guilt, which had been strenuously disputed by campaigners, was proved "beyond doubt".<sup>[59]</sup> Paul Foot and some other campaigners continued to believe in Hanratty's innocence and argued that the DNA evidence could have been contaminated, noting that the small DNA samples from items of clothing, kept in a police laboratory for over 40 years "in conditions that do not satisfy modern evidential standards", had had to be subjected to very new amplification techniques in order to yield any genetic profile.<sup>[60]</sup> However, no DNA other than Hanratty's was found on the evidence tested, contrary to what would have been expected had the evidence indeed been contaminated.<sup>[61]</sup>
- In 2002, DNA testing was used to exonerate Douglas Echols, a man who was wrongfully convicted in a 1986 rape case. Echols was the 114th person to be exonerated through post-conviction DNA testing.
- In August 2002, Annalisa Vincenzi was shot dead in Tuscany. Bartender Peter Hamkin, 23, was arrested, in Merseyside, in March 2003 on an extradition warrant heard at Bow Street Magistrates' Court in London to establish whether he should be taken to Italy to face a murder charge. DNA "proved" he shot her, but he was cleared on other evidence.<sup>[62]</sup>
- In 2003, Welshman Jeffrey Gafoor was convicted of the 1988 murder of Lynette White, when crime scene evidence collected 12 years earlier was re-examined using STR techniques, resulting in a match with his nephew.<sup>[63]</sup> This may be the first known example of the DNA of an innocent yet related individual being used to identify the actual criminal, via "familial searching".
- In March 2003, Josiah Sutton was released from prison after serving four years of a twelve-year sentence for a sexual assault charge. Questionable DNA samples taken from Sutton were retested in the wake of the Houston Police Department's crime lab scandal of mishandling DNA evidence.
- In June 2003, because of new DNA evidence, Dennis Halstead, John Kogut and John Restivo won a re-trial on their murder conviction. The three men had already served eighteen years of their thirty-plus-year sentences.
- The trial of Robert Pickton (convicted in December 2003) is notable in that DNA evidence is being used primarily to identify the *victims*, and in many cases to prove their existence.
- In 2004, DNA testing shed new light into the mysterious 1912 disappearance of Bobby Dunbar, a four-year-old boy who vanished during a fishing trip. He was allegedly found alive eight months later in the custody of William Cantwell Walters, but another woman claimed that the boy was her son, Bruce Anderson, whom she had entrusted in Walters' custody. The courts disbelieved her claim and convicted Walters for the kidnapping. The boy was raised and known as Bobby Dunbar throughout the rest of his life. However, DNA tests on Dunbar's son and nephew revealed the two were not related, thus establishing that the boy found in 1912 was not Bobby Dunbar, whose real fate remains unknown.<sup>[64]</sup>
- In 2005, Gary Leiterman was convicted of the 1969 murder of Jane Mixer, a law student at the University of Michigan, after DNA found on Mixer's pantyhose was matched to Leiterman. DNA in a drop of blood on Mixer's hand was matched to John Ruelas, who was only four years old in 1969 and was never successfully connected to the case in any other way. Leiterman's defense unsuccessfully argued that the unexplained match of the blood spot to Ruelas pointed to cross-contamination and raised doubts about the reliability of the lab's identification of Leiterman.<sup>[65][66][67]</sup>
- In December 2005, Evan Simmons was proven innocent of a 1981 attack on an Atlanta woman after serving twenty-four years in prison. Mr. Clark is the 164th person in the United States and the fifth in Georgia to be freed using post-conviction DNA testing.

- In March 2009, Sean Hodgson who spent 27 years in jail, convicted of killing Teresa De Simone, 22, in her car in Southampton 30 years ago was released by senior judges. Tests prove DNA from the scene was not his. British police have now reopened the case.

## See also

- DNA barcoding
- DNA database
- National DNA database
- Capillary electrophoresis (CE)
- Forensic identification
- Full genome sequencing
- Gene mapping
- Genealogical DNA test
- *Harvey v. Horan*
- Identification (biology)
- Kinship analysis
- Parental testing
- Phantom of Heilbronn
- Project Innocence
- Restriction fragment length polymorphism (RFLP)
- Ribotyping
- Short tandem repeat (STR)
- State of Louisiana v. Frisard

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65. ^ CBS News (<http://www.cbsnews.com/stories/2005/11/22/48hours/main1066064.shtml>) story on the Jane Mixer murder case; March 24, 2007.
66. ^ Another CBS News (<http://www.cbsnews.com/stories/2007/07/17/earlyshow/main3065082.shtml>) story on the Mixer case; July 17, 2007.
67. ^ An advocacy site (<http://www.garyisinnocent.org>) challenging Leiterman's conviction in the Mixer murder.

## Further reading

- Anne Hart (July 2003). *The Beginner's Guide to Interpreting Ethnic DNA Origins for Family History: How Ashkenazi, Sephardi, Mizrahi & Europeans Are Related to Everyone Else* (<http://books.google.com/books?id=DHBuRpLQaKQC&pg=PP1>) . iUniverse. ISBN 978-0-595-28306-4. <http://books.google.com/books?id=DHBuRpLQaKQC&pg=PP1>.

## External links

- DNA Fingerprinting (<http://www.guardian.co.uk/science/2009/may/24/dna-fingerprinting-alec-jeffreys>) Eureka Moment
- Create a DNA Fingerprint (<http://www.pbs.org/wgbh/nova/sheppard/analyze.html>) PBS.org
- In silico simulation of Molecular Biology Techniques (<http://insilico.ehu.es>) - A place to learn typing techniques by simulating them
- National DNA Databases in the EU (<https://www.kuleuven.be/cbmer/page.php?LAN=E&ID=399&FILE=subject&PAGE=1>)
- The Innocence Record (<https://www.innocencerecord.org>) , Winston & Strawn LLP/The Innocence Project

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Categories: Applied genetics | Biometrics | DNA | DNA profiling techniques | Molecular biology  
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