Title
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ISBN
9780080970868

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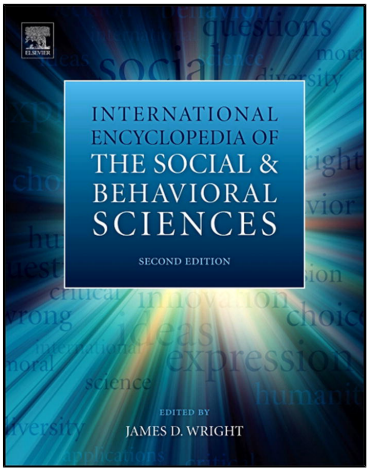
Publication Date
2015

DOI
10.1016/b978-0-08-097086-8.82050-4

Peer reviewed



From Cole, S.A., Prainsack, B., 2015. Genetics and Forensics. In: James D. Wright (editor-in-chief), International Encyclopedia of the Social & Behavioral Sciences, 2nd edition, Vol 9. Oxford: Elsevier. pp. 955–961.
ISBN: 9780080970868

Genetics and Forensics

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Abstract

This article provides an overview of the use of genetic science and technologies for criminal investigation and forensics. After discussing the history of the use of biological markers for criminal identification and forensics, we outline the emergence of DNA analysis as the 'gold standard' in forensic identification. We then discuss a range of more recent practices and issues in the field of forensic genetics. We conclude with a brief discussion of novel technologies that have not yet found entrance into most courtrooms and routine crime scene work.

Background: Forensic Identification

In criminal investigation, it often is useful to try to link bodily traces to suspect bodies, which may have been present at sites of interest or had contact with persons or objects of interest. The earliest uses of such traces probably involved handwriting, but the paradigmatic forensic identifier is the fingerprint, which came into regular use for forensic purposes in the early twentieth century. Unlike handwriting, which is a volitional act, fingerprints are bodily attributes, which are in some sense outside the control of the individual, and which we call *markers*. Other forensic identification marker sources included hair, dentition, and blood. The foundation of bodily forensic identification lies in the claim that traces can, with some degree of accuracy and precision, be linked to the suspect body. A 'known' fingerprint trace is compelled from the suspect body (i.e., by 'rolling' the suspect's fingerprint), and the 'known' and 'unknown' traces are compared. For most of the twentieth century, fingerprinting often was called the 'gold standard' in forensic identification and, indeed, forensic science more generally. Fingerprints were particularly useful for forensic science for a number of reasons: People generally deposited fingerprints fairly prolifically, making the recovery rate high, despite rather well-known and easily adopted countermeasures like wearing gloves. Whereas handwriting identification may have been particularly useful in investigation of crimes like fraud and other financial crimes involving documents, fingerprints were deposited by the anatomical organs of touch, which were frequently employed in the commission of crimes, especially violence and theft. Fingerprints could be compelled from suspects easily and cheaply. These known prints could be indexed according to their 'pattern types' by trained classifiers and could be stored relatively cheaply. At the same time, however, the utility of fingerprinting was limited. Not all crime scenes yielded incriminating prints. If suspects were not identified by investigative means, the examiner would have no 'known' to which to compare the crime-scene print. The database could be consulted, but this often was prohibitively difficult because most databases were indexed according to the pattern types on all 10 fingers, whereas crimes scenes rarely yielded complete sets of 10 prints. This issue placed severe limits on the utility of fingerprint databases in suspectless crimes until the flourishing of computerized searching in the 1980s.

Development of Forensic DNA Profiling

Forensic serology, the analysis of blood, was not new. It had been possible for decades to analyze blood stains for blood type and also for the presence of various enzymes. These tests, however, were not sufficiently discriminating. Serology was mostly helpful for the exclusion of suspects (e.g., if a trace that must have come from the perpetrator had a different blood type than the suspect). Fingerprinting, at this time, was claiming to be able to achieve infinite discrimination, that is, to the level of a single individual (this claim later was found to be unfounded). With criminal investigation paying increasing attention to biological information, the utility of treating genetic information as a forensic marker was clear. It was in this milieu that the utility of Alec Jeffreys' discovery of probes for visualizing variable-number tandem repeats (VNTRs) (*see* Forensic Genetic Databases: Ethical and Social Dimensions) for forensic identification gradually became apparent. Given a sample of bodily fluid, the forensic investigator could posit the same sort of link between a bodily trace and a suspect body that had been posited for fingerprints. Indeed, Jeffreys called the technique 'DNA fingerprinting' in a deliberate evocation of the older technique (Lynch et al., 2008). Because DNA profiling – the term that later became standard – targeted a different type of bodily trace than fingerprint analysis did, it could be used in investigations in which no usable fingerprints were found, but in which body fluids from the likely perpetrator were present.

Briefly, Jeffreys' method utilized the fact that human beings have different variants of DNA in certain places on their chromosomes; in healthy individuals, DNA is the same in every cell in the body. The higher the number of places (loci) in the chromosome that are being analyzed, the smaller is the likelihood that two individuals have the same DNA variants in all the same places (unless they are identical twins or multiples; for more details, *see* Forensic Genetic Databases: Ethical and Social Dimensions). Jeffreys' method of DNA analysis went far beyond serology in the sense that it allowed for the testing of several genetic loci, which offered much higher discrimination in the analysis of bodily fluids. The technique was famously first put to use in 1986 in a serial rape-murder cases in Narborough in the United Kingdom near Jeffreys' laboratory at the University of Leicester. There was a prime suspect, who even confessed to the crimes; genetic analysis, however, excluded him as a possible

International Encyclopedia of the Social & Behavioral Sciences, Second Edition, 2015, 955–961

perpetrator. The police then conducted what later would be called a 'DNA dragnet,' demanding blood samples from every eligible male in the area. This pressure eventually flushed out a witness who reported that a suspicious colleague was seeking to evade giving a sample. Several themes that would become important for genetics and forensics, including the exoneration of the innocent, DNA dragnets, and the power of the perception of the technique's accuracy to flush out other kinds of evidence (e.g., Machado and Prainsack, 2012), were presaged in this case.

The technique's potential for forensics was apprehended relatively quickly, and some law enforcement agencies, including the (now defunct) UK Forensic Science Service (FSS) and the US Federal Bureau of Investigation (FBI) moved quickly to develop it. In addition, a number of private companies arose to market the DNA profiling services (Daemmrich, 1998). The technology developed rapidly. Systems based on the polymerase chain reaction (PCR) were developed during the 1990s. These systems enabled 'amplification' of forensic traces – small amounts of genetic material could be copied until sufficient material to test was available, and testing also became faster. But early PCR markers lacked the discrimination of Jeffreys' original technique. The technique of counting short tandem repeats (STRs) – repetitions of chains of nucleotides in given loci – combined PCR's amplification capability with high discrimination power. STR-based techniques remain the most common DNA profiling system used in criminal justice. According to the latest survey carried out by Interpol on this topic, 120 of the 172 Interpol member countries reported to have used DNA profiling (Interpol, 2008).

Effects on Criminal Investigations

The effect of genetics on criminal investigation may usefully be divided into two categories: cases in which genetic analysis inculpates a suspect and cases in which it exculpates a suspect or convict. There is, obviously, a quite significant third category of cases in which genetic analysis neither inculpates nor exculpates. Examples of such cases might include those in which the genetic samples are degraded, contaminated, or otherwise not fully analyzable a relatively robust profile is not linked to a person of interest in solving the crime, or the DNA profile of the suspect could have ended up at the crime scene for legitimate reasons (see following section).

Inculpation

Genetic analysis is of obvious utility in providing inculpatory evidence in the investigation of crimes. Evidence of a possible common source between a crime scene sample and a sample from a known individual can provide strong evidence of that individual's presence at the crime scene. Depending on the nature of the sample, that evidence may be more or less incriminating in the actual commission of the crime. For instance, consistency with a genetic sample derived from a semen sample from a rape-murder case may be incriminating. On the other hand, consistency with a genetic sample derived from a sweat stain at a public, highly trafficked location may be far less incriminating. Incriminating links need not necessarily involve a crime scene. For example, links might be drawn between a genetic sample recovered from a suspect's property and a genetic sample known to come from the crime victim.

Although it is commonplace to speak of genetic 'matches' in forensics, in fact, an association between two genetic samples is probabilistic in nature. Genetic forensic scientists assess whether the two samples appear to have the same alleles at the designated loci. This assessment sometimes can require interpretation, as when there are ambiguities as to whether a given result is genuine or is the result of noise, contamination, mixture with DNA from another source, and so on. Even if the findings of consistency are assumed to have been interpreted correctly, however, a further question concerns whether other individuals in the population might have the same array of alleles at the designated loci. A monozygotic (identical) twin is an obvious possibility. Even setting twins or multiples aside, however, genetically distinct individuals might by chance have the same array of alleles at the loci examined. It is, therefore, necessary to try to estimate the rarity of the array of alleles. A common such estimate used in forensics is a 'random match probability' (RMP). Genetic information has an unusual data structure not shared by most other forensic disciplines – the alleles are numbers of repeats and thus exist in nature as discrete whole-number values, rather than continuous measures – which makes the calculation of such estimates relatively straightforward in simple (e.g., nonmixture) cases.

It is necessary to take this estimate into account when considering the inculpatory value of a finding of genetic consistency between two samples. Many forensic scholars believe strongly that the proper framework for such considerations is what generally is known as a 'logical' or 'Bayesian' approach based on a likelihood ratio, but the penetration of this approach into routine practice and courtroom proceedings has been mixed. The approach has been embraced in some countries such as the Netherlands and Sweden, and it was strongly promoted by the FSS in the United Kingdom. In other countries, like the United States, however, the Bayesian approach remains rare. Instead, most forensic genetic expert witnesses testify about RMPs. In some cases, when RMPs are low, experts testify, controversially, to source attributions – that is, a single individual *is* the source of the forensic trace.

One paradox of advancing technology is that, as genetic tests have become increasingly sensitive, their incriminating value is in some cases beginning to erode. In the earliest days of genetics and forensics, blood or semen were necessary to contain sufficient genetic material to analyze. This left a lot of crime scenes impervious to genetic forensics, but if an association between a suspect and a blood or semen sample was established, that association often was quite difficult for the suspect to explain away without reference to increasingly fanciful scenarios involving other attackers or other less probable events. Contemporary technology, however, is capable of recovering analyzable samples from quite small traces of genetic material derived from sweat, hair, skin cells, body oils, and so on – so-called 'touch DNA.' This makes genetic analysis deployable for a much broader variety of crimes from auto thefts to financial crimes. At the same time, it increases greatly the number of samples that will be recovered from a particular crime of object, but it also increases the probability that the

sample came to be at the scene 'innocently' through a prior visit or transfer, thus increasing the range and plausibility of defense that might explain a finding of association. Planting scenarios are also increasingly plausible.

Exculpation

Many exculpations, presumably, occur at the investigative state. How many is not well understood, although during the early years of DNA profiling, the FBI reported that around one-quarter of prime suspects were exculpated by DNA analysis (Connors et al., 1996). The more spectacular exculpations are those that have occurred postconviction. Because biological evidence is sometimes preserved, the development of genetic forensics in the 1980s made it possible to retest evidence postconviction. This resulted in a significant number of spectacular exonerations of individuals who often had served long prison sentences, under sentence of death in some cases, for crimes they apparently did not commit. The primary locus of such exonerations has been the United States, possibly because of better preservation of evidence, where there have been more than 300 'post-conviction DNA exonerations' since 1989 (Garrett, 2011). Although there always have been other means of exonerating the convicted – and, indeed, during the period since 1989, there have been more non-DNA exonerations than DNA exonerations – DNA exonerations have an ability to achieve 'epistemological closure' (Aronson and Cole, 2009) that other exonerations usually lack. This means that 'innocence skeptics' are less disposed to challenge the factual basis of exonerations when they are vouched for by genetic evidence. In most cases, such challenges rely on changing the government's theory of the crime from single- to multiple-perpetrator rape. In recent years, initiatives of lawyers working *pro bono* to exonerate the wrongfully convicted have become more widespread in countries outside of the United States, including Canada, France, New Zealand, and Australia.

In the United States, the wave of DNA exonerations has been extremely influential in stimulating the movement for criminal justice system reform and also for supporting opposition to capital punishment. Seemingly paradoxically, a small number of the US DNA exonerations have involved individuals who also were convicted on the basis of genetic evidence, evidence that was either misinterpreted or presented in a misleading manner (Garrett, 2011). It has long been predicted that DNA exonerations eventually would wane, as DNA testing at the investigation stage became routine, but thus far exonerations continue.

Use as Evidence in Court

The 'DNA Wars'

During the earliest years in which DNA evidence was introduced, its admissibility was extensively litigated in the United States, in a series of contests sometimes labeled as the 'DNA wars' (Aronson, 2007; Kaye, 2010) and in the United Kingdom (Lynch et al., 2008). In the earliest US cases (in the late 1980s and very early 1990s), DNA evidence was either not challenged or not challenged expertly, and it was routinely admitted. In later cases, however, defense attorneys enlisted well-credentialed molecular biologists who were able to gradually expose sloppy practices, failure to adhere to protocols, and unprincipled (and biased) interpretations of data. These interventions and criticisms produced a few cases in which state courts excluded DNA results, perhaps most famously in *People v. Castro*. Not insignificantly, these exclusions quickly were followed by federal courts deeming similar evidence admissible. The courts also exercised caution in early cases in the United Kingdom and Australia (Edmond et al., 2013). In the United Kingdom, proposals to employ Bayesian statistical analysis to interpret DNA evidence were rejected by the courts (Lynch et al., 2008).

Successful challenges to the admissibility of DNA evidence in the United States drew on population genetics to challenge calculations of RMPs. Drawing on debates among geneticists about human mating patterns, defendants argued that the state's RMP calculations were not accepted in the scientific community. These cases helped to trigger the intervention of the US National Academy of Sciences – through its National Research Council (NRC) committees. The NRC issued two reports, in 1992 and 1996, each of which endorsed two different ways of estimating the RMP, the 'ceiling principle' and the 'product rule,' respectively. Following the first report, some courts excluded RMPs proffered by the government. The second report, however, practically eliminated admissibility challenges based on population genetics; to the extent that the government asserted that it was adhering to the NRC recommendation, admissibility challenges were unlikely to succeed, and subsequent cases upheld admissibility. Since the mid-1990s, DNA evidence in general has been universally admissible in the United States and many other countries (Edmond et al., 2013; Hindmarsh and Prainsack, 2010). Another consequence of these reports was that the issue of accounting for human mating patterns was resolved by allowing for the calculation of RMPs based on gross categories labeled 'race' (Kahn, 2008): for example, in the United States the categories were 'White,' 'Black,' and 'Hispanic'; in the United Kingdom, they were 'White,' 'Black,' and 'Asian.'

Perhaps the most famous motion to exclude DNA evidence was one that was never filed. Although the defendant's 'dream team' prepared an extensive motion to exclude DNA evidence in *People v. O.J. Simpson*, sometimes called 'the trial of the century,' they withdrew the motion early in 1995. Instead, the defense team famously – and generally, it would appear, successfully – opted to attack the weight of the evidence at trial by alleging sloppy procedures, inadvertent contamination, and possible planting of evidence (Aronson, 2007: p. 173; Kaye, 2010: p. 152).

Targeted admissibility challenges still are made. One avenue of challenge concerns how a 'cold' database search affects the calculation of the RMP. Because statisticians disagreed about how the fact that a DNA association was generated through a database search should be handled (although all agreed that it mattered), some defendants argued, unsuccessfully, that the government's method of calculating the RMP was not 'generally accepted.' There has been some litigation on statistical grounds about the conclusions that DNA analysts should be permitted to state in their testimony and that prosecutors

should be permitted to state in their summations. There has thus far been only a small amount of litigation over DNA profiling techniques more exotic than the STR testing that has become standard. The admissibility of Y-STR haplotyping (the analysis of DNA in the male sex chromosome, which can provide information on paternal lineage) and mitochondrial DNA profiling (i.e., the analysis of DNA that is not contained in the nucleus of the cell; mitochondrial DNA is passed on from mothers to children) both have been upheld in the United States (Kaye, 2010). In England, Wales, and Australia, there has been litigation concerning low-copy-number (LCN) DNA profiling (Gilder et al., 2009). LCN DNA profiling is a particularly sensitive method of analysis for which only tiny amounts of DNA – a few cells – are needed. Disagreement remains about the reliability of LCN DNA markers.

As the technology has improved and RMPs – that is, the chances that a DNA profile obtained from a crime scene matches the profile of a randomly drawn profile from the general population – have gotten smaller, the arguments for the importance of race and racial mating patterns have diminished. It originally was argued that race-specific calculations of RMP were important to generate more accurate RMPs, but some social scientists now argue that the purported accuracy gains derived from using racial categories 'fade into irrelevance' and are outweighted by the harms of reifying crude notions of genetic races (Kahn, 2008).

Effects on Forensic Technologies

Although the legal admissibility battles generally have resulted in admissibility for genetic forensics, they had unexpected and unintended consequences for other areas of forensic science. The sophisticated disputes involving high-powered scientists over population genetics and the statistical reporting of results provoked the question of how these issues were addressed in other areas of forensic identification, such as fingerprinting, firearms and toolmarks, bitemarks, handwriting, microscopic hair comparison, and so on. It turned out that all these fields had been indifferent to statistics, viewing it as unnecessary to seek to statistically characterize the size of the potential donor pool of forensic traces. Instead, these fields had relied on semantic workarounds that evaded these issues, and courts, historically, had permitted them to do so. The admission of genetic forensics into court thus had the effect of prompting defendants and scholars to demand that these older fields of forensic science account for themselves probabilistically, resulting in admissibility challenges to venerable techniques whose admissibility had seemed long settled. These developments have lent momentum to a movement toward probabilistic thinking in forensic science, which has held that all evidence can, and should, be understood probabilistically. It also generated a phenomenon that has been called an 'inversion of credibility' in which genetic forensics, which in the early 1990s routinely described itself as similar to, but weaker than, venerable techniques like fingerprinting, came to be widely seen as stronger. The period in which proponents of genetic forensics sought to bolster their credibility by invoking analogies with fingerprinting was followed by a period in which genetics was widely seen as having succeeded fingerprinting as the 'gold standard' in forensics (Lynch et al., 2008).

The 'CSI Effect'

Another potential unintended consequence of the development of DNA profiling is the so-called CSI effect, a term used somewhat inconsistently to mean a variety of things. Increasing awareness of the existence of forensic genetics may reasonably be expected to have a legitimate effect on the behavior of various criminal justice system actors, including attorneys, judges, jurors, police, and offenders. This effect has been dubbed the 'tech effect' to distinguish it from the 'CSI effect,' which generally is used to denote an exaggerated, and often false, belief in the power and capabilities of forensic science, including forensic genetics, fostered by popular fictional media, especially television programming. The most common claims have been advanced in regard to US juries, which, it is claimed, are acquitting in cases in which they would have convicted but for television programming. Evidence in support of such strong claims of a 'CSI effect' is thus far weak, but claims of changes in attorney, offender, or even individual juror behavior are better supported (Shelton et al., 2006; Cole and Dioso-Villa, 2007; Machado and Prainsack, 2012; Cole, in press).

Databases

In the early years, the results of DNA analysis from crime scene and suspect samples were compared in an ad hoc manner – that is, in the context of specific investigations and trials – and stored in a decentralized manner, if they were stored at all. In the mid-late 1990s, as an effect of both the increasing importance of STR-based DNA analysis in forensic investigation, and in court, centralized databases for police and forensic use were set up in many countries. The ways in which these databases were set up differ greatly in terms of when they were started, what the criteria for inclusion and expungement of profiles are, and in whose custody the database is held (*see* Forensic Genetic Databases: Ethical and Social Dimensions; see also Hindmarsh and Prainsack, 2010; Krimsky and Simoncelli, 2011). Not all countries had or have dedicated laws governing the establishment, maintenance, and use of DNA databases. What all DNA databases for police and forensic uses have in common, however, is that they include DNA profiles from crime scenes and from convicted offenders; some databases store profiles of suspects (e.g., England and Wales) or volunteers (e.g., Portugal) as well.

All databases allow so-called speculative searching, which means that profiles obtained from crime scene stains, or from subjects, can be compared against all other profiles in the database, without there being a particular suspect, or a hypothesis regarding how specific profiles could be linked. By making speculative searching possible, centralized DNA databases have given rise to many new 'cold case' investigations, that is, the reopening of investigations of cases that had ran out of leads (Innes and Clarke, 2009). At the same time, speculative searching has led to concerns about false matches within the database. This is particularly relevant in the context of very large databases, or networks of databases, such as the Prüm network: The Prüm network is based on EU law that obliges member states to render their DNA databases searchable to each other (at a hit–no hit basis; supposed profile matches then are followed up bilaterally by the countries between which the databases matches were

obtained; see Prainsack and Toom, 2013). In 2011, databases within the Prüm network included more than 9 million profiles (ENFSI, 2011); comparing all these profiles with each other would lead to several trillions of DNA comparisons. This means that, depending on how many loci are searched, even with very low RMPs (e.g., several billions), numerous false matches could result from every single search within the database. It is also for this reason that the European Union has endorsed the inclusion of five new markers in the European Standard Set of Loci (ESSOL; a voluntary agreement on a minimum set of genetic loci that DNA analysis in all countries include, irrespective of what chip and software they use for analysis; see Gill et al., 2006; European Union (EU) Council, 2009). These five markers will be added to the Combined DNA Index System (CODIS) used in the United States and elsewhere to ensure compatibility of DNA profiles across countries also outside of Europe, and within the DNA Gateway run by Interpol (*see* Forensic Genetic Databases: Ethical and Social Dimensions).

Function Creep

Over the course of the development of genetic forensics, the functions and uses of some DNA databases have expanded beyond those originally intended. This phenomenon has been referred to as function creep (Dahl and Sætnan, 2009). For example, the fact that biologically related people share more genetic markers in common than unrelated people could be used to search for biological relatives of a person in connection with a crime in the DNA database. Another example is the increasing use of genetic information to generate 'intelligence' about hitherto unknown crimes or linkages between crimes (Ribaux et al., 2010). Still another example is the linkage of DNA analysis results carried out by handheld devices with DNA databases. This may emerge as a result of the development of ultrarapid (i.e., a few hours) DNA analysis that does not need to be carried out in a laboratory but can be performed by crime scene officers, and analyses could be compared automatically against profiles held in the database (currently, this is not done in any country). Such a scenario would raise questions about accreditation, and about data leakage and misuse, and about the exacerbation of biases against certain population groups if handheld DNA analysis devices also would be used in police patrols. A third example is the storage of so-called single-nucleotide polymorphisms (SNP)-data or Y-chromosome STR data, alongside conventional STR-profiles in centralized DNA databases. SNPs are variations at the level of single nucleotides (G, A, T, C); forensic SNP-based tests scan places across the entire genome for such variants and highlight those that have been found to correlate with particular physical traits (e.g., eye color) or ethnicity in scientific studies (e.g., Kayser and Schneider, 2009). The question about storage of SNP-data thus emerges in connection with phenotypic profiling. Y-chromosome analysis, which can give clues about paternal lineage and – in societies where surnames are patrilineal – allow probabilistic inferences about a person's family name, has been used in criminal investigation for decades; however, the results of Y-chromosome analysis are not stored routinely in centralized DNA databases and thus are not regularly available for speculative searches. There is a Y-chromosome haplotype reference database that can aid case workers by providing Y-STR haplotype frequency estimates, but this database does not contain profiles of particular convicted individuals.

Access for Researchers

There is great variety between jurisdictions as to whether or not they allow researchers to access DNA samples or profiles stored in the database. Any research carried out with these materials or data normally will have gone through institutional research ethics reviews, and personal identifiers (names, addresses, etc.) will not be disclosed to researchers, although recent studies have shown that anonymization is no guarantee that reidentification will be impossible (Gymrek et al., 2013). The STR-based DNA profiles currently stored in centralized DNA databases are not useful for sensitive research into, for example, correlations between genetic markers and antisocial or criminal behavior, intelligence, disease propensity, character traits, sexuality, and ethnicity. This is because STR profiles – which are stored as strings of numbers in the database – represent random genetic variants of which we have millions, most with yet unexplored, or nonexisting, function. DNA *samples* – the physical material, such as blood or saliva stains, from which DNA profiles are derived – are stored in the laboratories carrying out the analysis in some countries, while other countries require that labs destroy samples once the DNA profile has been obtained. Retained DNA samples potentially could be useful for such future research, as would the analysis of existing SNP data (see next section on phenotypic profiling). In addition, some scholars have called for database custodians to allow increased access to (purportedly) anonymized data to better facilitate research on issues related to forensics, such as, for example, the statistical independence of the alleles used for profiling (Krane et al., 2009). Paradoxically for some, database custodians have emerged as defenders of the personal privacy of convicted offenders in these debates. The use of DNA samples and data collected and generated during criminal investigation for research raises a range of political and ethical concerns, especially in cases in which no transparent and publicly accessible information about the purposes and terms of such research exists.

Surreptitious Sampling

Concerns have been raised also about the practice of so-called surreptitious sampling. In the context of forensic genetics, the term 'surreptitious sampling' refers to analyses carried out on DNA that has not been obtained with the knowledge of the originator (the 'owner' of the DNA). Surreptitious sampling could use DNA left behind by suspects on coffee mugs, chewing gum, cigarette ends, or clothes, for example (Joh, 2006). At present, DNA profiles obtained by means of surreptitious sampling cannot be uploaded to the DNA database in most countries; they can be used only for comparisons in concrete cases, if at all. The issues around surreptitious sampling – or better, 'convenience' sampling in this context – would become pressing if portable DNA testing devices were deployed locally (e.g., at police stations or on police patrols), and if the people operating the device could compare the profile generated by the portable device with the profiles stored in the national database. This could lead to a situation in which individuals from whom it is not legal to demand a DNA sample could be submitted to

testing 'through the backdoor.' Moreover, as members of ethnic minorities are more likely to be affected by routine police controls in many countries ('racial profiling'), this practice also would exacerbate existing ethnic and racial biases.

Phenotypic Profiling

Another, more recent, practice that is sometimes discussed in connection with racial profiling so-called phenotypic profiling. Conventional, STR-based forensic DNA profiling looks only at genetic markers that do not allow for any meaningful inferences regarding a person's externally visible traits, such as her hair or eye color. If different markers on the genome are analyzed, however – namely, those that have been found to correlate with externally visible traits (Keating et al., 2012) – then inferences can be made regarding the person's likely characteristics. These inferences always will be probabilistic, because the predictive power of the genetic markers regarding expressed phenotypic traits is limited. In principle, this method can be highly useful in the context of some criminal investigations – for example, if there are no known suspects, phenotypic profiling could give an indication of what the perpetrator could look like and also what ethnic background she or he is likely to have. The latter is the case because some of the same markers that correlate with externally visible traits are typical for specific population groups. This example illustrates both the potential benefits as well as the potential risks inherent in phenotypic profiling (M'charek, 2008). Because the information obtained by this method is, as mentioned, not deterministic but probabilistic, its utility is limited, not only as evidence at court, but also as intelligence in a criminal investigation. For example, if in the context of a dragnet, the information that the perpetrator is 67% likely to have blue eyes, were used to target only people with blue eyes, the real perpetrator could be missed. (As dragnets are highly intrusive for those asked to volunteer a DNA sample, there is wide agreement among experts that they should be used sparingly, and only in a range of narrowly defined contexts; see Zadok et al., 2010.) DNA information used for phenotypic profiling purposes currently is not stored routinely in centralized DNA databases. Phenotypic profiling could be useful particularly in narrowing down the range of missing persons to which unidentified bodies could be matched (also in the context of disaster victim identification).

Familial Searching

'Familial searching' – which arguably would be more accurately called 'genetic proximity testing,' as not all genetically related people feel part of a family, and vice versa – can be performed if a DNA profile derived from a crime scene stain results in a partial match with a profile in the database. For example, two profiles might not match in all of the analyzed genetic loci but in most of them (e.g., in 11 of the 13 loci used in the US Federal CODIS database; for this reason, some authors call it 'low stringency,' 'near-miss matching,' or 'kinship matching'; e.g., Kaye, in press). The 'hit' in the database could have identified a genetic relative of the person who actually left the DNA at the crime scene. Police could then investigate whether the person identified has any biological relatives who could have committed the crime. In the United Kingdom and in the United States, for example, several cases have been solved by this method. In other countries, genetic proximity testing is explicitly or implicitly forbidden. Critics of genetic proximity testing argue that this method violates the privacy of the person whose profile has been identified on the database through the matching attempt, as well as the genetic privacy of the (genetic) relatives of the person whose DNA profile has been identified, and that it could reinforce the stereotype of 'crime running in families.' Moreover, some critics are concerned that a criminal investigation using genetic proximity testing could reveal a genetic link (or lack thereof) between members of a family that they currently were unaware of (Haimes, 2006, see also Greely et al., 2006). Finally, genetic proximity testing is seen to repeat existing demographic and ethnic biases within criminal justice systems. For example, in the forensic DNA database for England and Wales, black individuals are clearly overrepresented: According to the 2011 census in England and Wales, about 3.5% of those who filled in the questions about ethnicity self-identified as 'black' (in addition, just over 1% were of 'mixed' ethnicity, including 'black'; Office for National Statistics, 2011). The proportion of DNA profiles from 'black' individuals stored in the National DNA Database (NDNAD) is currently at more than 8%, that is, almost exactly double the proportion of black individuals in the general population (National Policing Improvement Agency, 2012). Although no data are available regarding socioeconomic background in this respect, a similar overrepresentation of certain groups is a possible, if not likely, scenario. This, in turn, means that individuals genetically related to people whose DNA profiles are stored in the database have a higher risk of being the subject of an investigation than genetic relatives of individuals whose profiles are not in the database, which raises questions about equity and fairness.

Future Prospects: Gene Expression and Microbiome Analysis

A limitation of the use of any kind of DNA analysis for forensic identification purposes is that it is impossible to differentiate monozygotic twins or multiples with this method. At least two strands of current research could provide solutions for this: The first is the comparison of gene expression profiles that do not look at the sequence of nucleotides but rather at what genes are expressed. Although monozygotic twins and multiples – who have developed out of one single egg and sperm – carry the same nucleotide sequences in their genes, this does not mean that all of these genes are *expressed* in the same manner. Lifestyle, environmental exposures, and other factors have the potential to switch certain genes 'on' or 'off' epigenetically without altering their structure. Epigenetic modifications – such as DNA methylation, that is, the addition of a methyl group to a DNA nucleotide – affect how genes are transcribed and expressed (see e.g., Bell and Spector, 2011; Li et al., in press). Another way of distinguishing people who carry identical DNA sequences is looking at the microbes they carry in their bodies, for example, in their throat. 'Throat-swabs' disclosing the composition of microbes could complement the traditional buccal swabs, in which cells are taken from the inside of the mouth to isolate DNA. It is unlikely that microbiome profiles will ever replace DNA profiles, however, because

they have the disadvantage that they are not stable – the composition of microbes changes over people's lifetime. For concrete investigations, however, micobiome profiles could distinguish people with identical DNA profiles in the future.

See also: Bioethics in the Post-genomic Era; Bioethics: Genetics and Genomics; Ethical, Legal, and Social Implications Program at the National Human Genome Research Institute; Expert Testimony; Genetic Admixture; Genetic Ancestry Testing; Genetics and Disaster Victim Identification; Genetics and Social Justice; Genetics and Society; Genetics and Sociology; Genetics and the Media; Genetics: Legal Aspects; Genetics: The New Genetics; Genomics, Ethical Issues In; Human Genome Project: History and Assessment; Law and Society: Development of the Field; New Genetics and Race; Privacy: Theoretical and Legal Issues; Race: Genetic Aspects; Science and Law; Science and Technology Studies: Experts and Expertise; Scientific Controversies; Scientific Evidence: Legal Aspects; Truth and Credibility in Science.

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